

Reference List, June 4, 2008 to January 28, 2009

Adult Studies

Barf HA, Post MW, Verhoef M, Jennekens-Schinkel A, Gooskens RH, Prevo AJ. Restrictions in social participation of young adults with spina bifida. *Disabil.Rehabil.* 2008 Dec 29;1-7.

Abstract: Purpose. To determine participation restrictions of young adults with spina bifida (SB) in relation to health condition and activity limitations. Method. A total of 179 persons aged 16-25 years and born with SB participated in a cross-sectional study. The main outcome on four domains of participation (independent living, employment, education and partner relationships) was assessed using a structured questionnaire. Results. At the mean age of 21 years only 16% were living independently, more than one-third of the participants went to special secondary education, 53% of those who finished education did not have a regular job and 71% did not have a partner. Health condition variables (type of SB, hydrocephalus and level of lesion) and to a lesser extent activity limitations (wheelchair dependence and incontinence) were significant determinants for having participation restrictions. Perceived hindrances in participation included long-distance transportation (19-36%), accessibility (10-42%), physical impairments (22-40%), emotional barriers (20-32%) and financial limits (3-17%). More severe SB, defined as hydrocephalus, high level of lesion and wheelchair dependence, was related with more experienced hindrances due to long-distance transportation accessibility of buildings. Conclusions. Many young adults with spina bifida experience participation restrictions. Severity of SB was negatively related to participation. Social integration should be a major focus in the professional guidance of youngsters with physical disabilities.

[For many years, physicians have used an “illness model” when dealing with people with spina bifida, focusing on body structures and functions. The World Health Organization and others have argued that we need to look at a person’s activities and social participation as well. This article evaluated the social participation of adults with spina bifida in the Netherlands. They found that few of these adults were living independently, were gainfully employed, or had a partner. The more severe the spina bifida, the worse the social participation. The authors conclude that, “Social integration should be a major focus in the professional guidance of youngsters with physical disabilities.”]

Dicianno BE, Kurowski BG, Yang JM, Chancellor MB, Bejjani GK, Fairman AD, Lewis N, Sotirake J. Rehabilitation and medical management of the adult with spina bifida. *Am.J.Phys.Med.Rehabil.* 2008 Dec;87(12):1027-50.

Abstract: As the life expectancy of individuals with spina bifida increases, a lifelong need for management of many health issues in a rehabilitation setting has emerged in recent years. Physiatrists, in consultation with a variety of adult specialists, are particularly well suited to manage the common musculoskeletal, skin, bowel, bladder, renal, neurological, and other issues that arise in the adult population. This article reviews the last 20 yrs of literature pertinent to the rehabilitative care of this population, summarizes current evidence-based practice, and identifies key areas in which scientific evidence is lacking and future research is needed.

[This article reviews rehabilitation services provided to adults with spina bifida in the past. The

authors conclude that adults with spina bifida are living longer but encountering “a wide range of medical, rehabilitation, and psychosocial complications.” Future research will need to address these issues in order to optimize their quality of life.]

Galambos NL, Magill-Evans J, Darrah J. Psychosocial maturity in the transition to adulthood for people with and without motor disabilities. *Rehabilitation Psychology*. 2008; 53(4):498-504.

Objective: To identify individual, family, and community variables that contribute to psychosocial maturity in the transition to adulthood for people with and without motor disabilities (mean age = 25 years, range = 20 to 30). Participants/Design: Seventy-four people (34 men, 40 women) with developmental motor disabilities (cerebral palsy or spina bifida) and 72 people (31 men, 41 women) without motor disabilities from a Canadian province completed questionnaires and participated in a semi-structured interview. Results: In a hierarchical regression analysis, individual and family variables explained variance in psychosocial maturity for the total sample. More use of task-focused coping, lower levels of depression, and perceptions of fathers as autonomy fostering were associated with increased psychosocial maturity. These variables were also significant in a similar analysis for the group with motor disabilities with 1 addition; older age also was related to higher psychosocial maturity. Community variables did not explain variation in psychosocial maturity in either analysis. Conclusion: Knowledge about barriers to and facilitators of a successful transition to adulthood can be used to design services to maximize opportunities for young people with disabilities.

[This article addresses factors affecting psychosocial maturity in youth and young adults with spina bifida. Although many factors were related to the person’s family, the occurrence of depression also played a role, reminding us that depression is a treatable occurrence in these individuals.]

Guarnieri J, Vinchon M. [Follow-up of adult patients with myelomeningocele]. *Neurochirurgie* 2008 Oct;54(5):604-14.

Abstract: The adult outcome of spina bifida patients is fraught with multiple problems reflecting the multisystemic nature of the disease. These problems result in both mortality, actually caused in most cases by the shunt, and morbidity, mostly affecting locomotion and sphincter control. These patients can thus lose ambulation and suffer from worsening incontinence and kidney function. The result in the vast majority of cases is a poor functional and socioprofessional outcome. The patients' demands regarding their occupational and affective lives, as well as procreation, must be heard. This requires a multidisciplinary approach, and pediatric and adult, as well as patient education and financial support for maintaining the patient's autonomy and medical acts allowing the viability of continued medical care.

[This article documents that many adults who have spina bifida—at least in France—not only carry the problems that they had as children, but deteriorate physically. They may lose ambulation and continence and develop renal failure as well. Interventions to prevent these problems from occurring should be researched.]

van Mechelen MC, Verhoef M, Van Asbeck FW, Post MW. Work participation among young adults with spina bifida in the Netherlands. *Dev.Med.Child Neurol*. 2008 Oct;50(10):772-7.

Abstract: The aim of this study was to: (1) assess work participation among young adults with spina bifida, (2) identify problems perceived in finding employment, and (3) examine which determinants are related to work participation. This cross-sectional study was a follow-up study to the Adolescents with SPina bifida In the Netherlands (ASPINE) study. Data regarding work participation and problems finding employment were collected with questionnaire developed by the authors. Data on disease characteristics were taken from the ASPINE database. Responses of 136 participants were analyzed (77 females, 59 males; mean age 26 years 1 month [SD 3y1mo], range 21-32y). Twenty participants had spina bifida occulta and 116 had spina bifida aperta, 96 of whom also had hydrocephalus. Work participation rate was 62.5%, of which 22.4% was in a sheltered workplace. Significant determinants of having paid work for at least 1 hour a week were: level of education, level of lesion, hydrocephalus, IQ, functional independence, and ambulation. Significant determinants of full-time employment were the same, plus sex and type of spina bifida. In a multivariate backward logistic regression analysis, however, only level of education remained a significant predictor of work participation. Sex, level of education, and self-care independence were significant predictors of full-time employment. This study shows the importance of educational support and self-care independence training for children with spina bifida.

[In this Dutch study, adults with spina bifida, aged 21 to 32 years, had low levels of employment. Those with higher levels of education and more independent skills were more likely to be employed.]

Epidemiology

Racial/ethnic differences in the birth prevalence of spina bifida - United States, 1995-2005. *MMWR Morb.Mortal.Wkly.Rep.* 2009 Jan 9;57(53):1409-13.

Abstract: In 1992, the U.S. Public Health Service recommended that all women of childbearing age consume 400 microg of folic acid daily to help prevent pregnancies affected by neural tube defects (NTDs) such as spina bifida. Subsequently, the Food and Drug Administration mandated adding folic acid to all enriched cereal grain products by January 1998. During October 1998--December 1999, the birth prevalence of spina bifida in the United States decreased 22.9% compared with 1995--1996; however, by 2003--2004, no further decrease had been observed. Notably, the prevalence of NTD-affected pregnancies remained higher among Hispanic women than among women in other racial/ethnic populations. To update previously reported data and assess racial/ethnic differences, CDC analyzed birth certificate data for four periods during 1995--2005. This report summarizes the results of that analysis, which indicated that from the early postfortification period, 1999--2000, to the most recent period of analysis, 2003--2005, the prevalence of spina bifida declined 6.9%, from 2.04 to 1.90 per 10,000 live births (prevalence ratio [PR] = 0.93; 95% confidence interval [CI] = 0.87--1.00). Among infants with non-Hispanic black mothers, prevalence fell 19.8%, from 2.17 to 1.74 per 10,000 live births (PR = 0.80; CI = 0.67--0.96), while prevalence among infants with non-Hispanic white and Hispanic mothers remained nearly constant. Additional public health efforts targeting women with known risk factors (e.g., obesity and certain genetic factors) likely are needed to further reduce the prevalence of spina bifida in the United States.

[This study examined birth prevalence rates of spina bifida from 1999 to 2005 and found that while the rate fell for African American women, it did not fall for Hispanics or Whites. The

reasons for these differences are uncertain.]

Bell KN, Oakley GP, Jr. Update on prevention of folic acid-preventable spina bifida and anencephaly. *Birth Defects Res.A Clin.Mol.Teratol.* 2009 Jan;85(1):102-7.

Abstract: **BACKGROUND:** The number of countries fortifying wheat and maize flour with folic acid has increased in the past 2 years. Folic acid prevents most cases of spina bifida and anencephaly by raising serum folate levels among women capable of bearing children, as does encouraging women to consume folic acid supplements prior to pregnancy. **METHODS:** The progress in preventing these serious birth defects can be measured by tracking the number of countries now fortifying and program coverage in each. Country estimates of the number of pregnancies affected by spina bifida and anencephaly are calculated using a prefortification birth prevalence baseline and estimates of the proportion prevented by wheat and maize flour fortified with folic acid. **RESULTS:** Current fortification programs are preventing about 22,000, or 9% of the estimated folic acid-preventable spina bifida and anencephaly cases. This represents an annual global decrease of about 6,600 folic acid-preventable spina bifida and anencephaly cases since 2006. **CONCLUSIONS:** The pace of preventing these serious birth defects can be accelerated if more countries require fortification of both wheat and maize flour and if regulators set fortification levels high enough to increase a woman's daily average consumption of folic acid to 400 mcg.

[This article documents the decreasing rate of spina bifida around the world as more countries begin to fortify flour with folic acid. Progress is being made, but it is slow.]

Boulet SL, Yang Q, Mai C, Kirby RS, Collins JS, Robbins JM, Meyer R, Canfield MA, Mulinare J. Trends in the postfortification prevalence of spina bifida and anencephaly in the United States. *Birth Defects Res.A Clin.Mol.Teratol.* 2008 Jul;82(7):527-32.

Abstract: **BACKGROUND:** The prevalence of NTDs in the US declined significantly after mandatory folic acid fortification; however, it is not known if the prevalence of NTDs has continued to decrease in recent years relative to the period immediately following the fortification mandate. **METHODS:** Population-based data from 21 birth defects surveillance systems were used to examine trends in the birth prevalence of spina bifida and anencephaly during 1999-2000, 2001-2002, and 2003-2004. Prevalence data were stratified by non-Hispanic White, non-Hispanic Black, and Hispanic race or ethnicity. Prevalence ratios were calculated by dividing the birth prevalences during the later time periods (2001-2002 and 2003-2004) by the birth prevalences during 1999-2000. **RESULTS:** During 1999-2004, 3,311 cases of spina bifida and 2,116 cases of anencephaly were reported. Hispanic infants had the highest prevalences of NTDs for all years. For all infants, the combined birth prevalences of spina bifida and anencephaly decreased 10% from the 1999-2000 period to the 2003-2004 period. The decline in spina bifida (3%) was not significant; however the decline in anencephaly (20%) was statistically significant. **CONCLUSIONS:** While the prevalences of spina bifida and anencephaly in the United States have declined since folic acid fortification in the food supply began, these data suggest that reductions in the prevalence of anencephaly continued during 2001-2004 and that racial and ethnic and other disparities remain.

[During the period of this study, the combined birth prevalences of spina bifida and anencephaly decreased 10%, with Hispanic mothers having the highest prevalences of NTDs for all years. The recent decline in the birth prevalence of spina bifida has been imperceptibly slow. Not all

people have equal access to preventive measures in this country.]

Chen BH, Carmichael SL, Selvin S, Abrams B, Shaw GM. NTD prevalences in central California before and after folic acid fortification. *Birth Defects Res.A Clin.Mol.Teratol.* 2008 Aug;82(8):547-52.

Abstract: **BACKGROUND:** In many regions, NTD prevalences were already declining prior to folic acid fortification. This study examined whether the declining prefortification (1989-1996) NTD prevalences continued into the postfortification period (1998-2003) in selected California counties. **METHODS:** This population-based study used vital statistics data and birth defects registry data that were actively ascertained from medical records. The study population included all live births and stillbirths delivered in central California counties from 1989 to 2003. Cases included deliveries with NTDs during the same time period. **RESULTS:** For all NTDs combined, the slopes indicated that NTD prevalence was decreasing by 7.5 (slope: -7.5; 95% CI: -12.4, -2.5) cases per 100,000 deliveries per year before fortification, whereas NTD prevalence was no longer decreasing after fortification. Comparison of the difference in the two slopes indicated that the postfortification slope exceeded the prefortification slope by 12.6 (95% CI: 2.6, 22.6) cases per 100,000 deliveries per year. **CONCLUSIONS:** Annual NTD prevalences in central California did not continue to decrease after implementation of folic acid fortification.

[This study found that in California the birth prevalence of spina bifida and anencephaly (NTDs) did not continue to decrease after folic acid was used to fortify flour. They discuss possible reasons including a change in elective terminations, and a change in risk factors for NTDs like maternal race/ethnicity, folic acid supplement use, and maternal obesity.]

De WP, Tairou F, Van Allen MI, Lowry RB, Evans JA, Van den Hof MC, Crowley M, Uh SH, Zimmer P, Sibbald B, et al. Spina bifida before and after folic acid fortification in Canada. *Birth Defects Res.A Clin.Mol.Teratol.* 2008 Sep;82(9):622-6.

Abstract: **BACKGROUND:** In 1998, fortification of a large variety of cereal products with folic acid became mandatory in Canada. A multicentric study was carried out to assess the impact of this policy on the frequency of NTDs. The present analysis focused on spina bifida. **METHODS:** The study population included approximately 2 million livebirths, stillbirths, and terminations of pregnancies because of fetal anomalies among women residing in seven Canadian provinces, from 1993 to 2002. Spina bifida cases were divided according to the upper limit of the defect: upper (cranial, cervical, or thoracic) and lower (lumbar or sacral) defects. Based on published results of red blood cell folate tests, the study period was divided into prefortification, partial fortification, and full fortification periods. **RESULTS:** A total of 1,286 spina bifida cases were identified: 51% livebirths, 3% stillbirths, and 46% terminations. Prevalence decreased from 0.86/1,000 in the prefortification to 0.40 in the full fortification period, while the proportion of upper defects decreased from 32% to 13%. Following fortification, regional variations in the prevalence and distribution of sites almost disappeared. **CONCLUSIONS:** Results confirmed the etiologic heterogeneity of spina bifida and the more pronounced effect of folic acid in decreasing the risk of the more severe clinical presentations.

[This study found that the birth prevalence of spina bifida in Canada decreased after grain was fortified with folic acid. The decrease was more noticeable among those with higher level (more

severe) lesions.]

Mosley BS, Cleves MA, Siega-Riz AM, Shaw GM, Canfield MA, Waller DK, Werler MM, Hobbs CA. Neural tube defects and maternal folate intake among pregnancies conceived after folic acid fortification in the United States. *Am.J.Epidemiol.* 2009 Jan 1;169(1):9-17.

Abstract: Rates of neural tube defects have decreased since folic acid fortification of the food supply in the United States. The authors' objective was to evaluate the associations between neural tube defects and maternal folic acid intake among pregnancies conceived after fortification. This is a multicenter, case-control study that uses data from the National Birth Defects Prevention Study, 1998-2003. Logistic regression was used to compute crude and adjusted odds ratios between cases and controls assessing maternal periconceptional use of folic acid and intake of dietary folic acid. Among 180 anencephalic cases, 385 spina bifida cases, and 3,963 controls, 21.1%, 25.2%, and 26.1%, respectively, reported periconceptional use of folic acid supplements. Periconceptional supplement use did not reduce the risk of having a pregnancy affected by a neural tube defect. Maternal intake of dietary folate was not significantly associated with neural tube defects. In this study conducted among pregnancies conceived after mandatory folic acid fortification, the authors found little evidence of an association between neural tube defects and maternal folic acid intake. A possible explanation is that folic acid fortification reduced the occurrence of folic acid-sensitive neural tube defects. Further investigation is warranted to possibly identify women who remain at increased risk of preventable neural tube defects.

[This study, which was conducted after mandatory folic acid fortification was established, documented virtually no association between neural tube defects and maternal folic acid intake. It is likely that fortification of food prevented the differences from being seen. Biomarkers like serum/plasma or RBC folate concentration were not used, so the correlation between folic acid use and spina bifida on an individual level could not be ascertained.]

Poretti A, Anheier T, Zimmermann R, Boltshauser E. Neural tube defects in Switzerland from 2001 to 2007: are periconceptual folic acid recommendations being followed? *Swiss.Med.Wkly.* 2008 Oct 18;138(41-42):608-13.

Abstract: **BACKGROUND:** Neural tube defects (NTDs) are common congenital anomalies. Their aetiology is complex, with both genetic and environmental factors implicated. The present study was performed to analyse the birth prevalence of NTD in Switzerland from 2001 to 2007 and to identify possible risk factors. **METHODS:** Diagnosed cases of NTD in all paediatric units in Switzerland and four prenatal centres were reported to the Swiss Paediatric Surveillance Unit from January 2001 to December 2007. Patient, mother, and NTD characteristics were assessed prospectively with a questionnaire. **RESULTS:** Data of 140 newborns and fetuses with NTD were studied. The major group suffered from myelomeningocele (70%), followed by anencephaly (16%) and encephalocele (14%). The prevalence of NTD in live born children between 2001 and 2007 was 0.13 per thousand, corresponding to 9-10 affected newborns each year. About the same number of pregnancies was terminated annually. Correct periconceptual folic acid supplementation was taken by 5% of the women. Remarkably, 39% of the women with an affected pregnancy were not Swiss citizens - almost twice the proportion of foreigners living in Switzerland. **CONCLUSIONS:** NTDs remain a frequent problem in Switzerland. Although correct periconceptual folic acid

supplementation is effective in reducing the prevalence of NTD, women still do not follow these recommendations. Possible reasons are lack of awareness and communication problems. Consequently, only a public health policy that includes folic acid fortification of food is likely to result in significant prevention of NTD.

[In this Swiss study of women who had a baby with a neural tube defect (NTD), only 5% had taken their folic acid correctly. More education is needed to insure better adherence to the established guidelines for using folic acid.]

Shin M, Besser LM, Correa A. Prevalence of spina bifida among children and adolescents in metropolitan Atlanta. *Birth Defects Res. A Clin. Mol. Teratol.* 2008 Nov;82(11):748-54.

Abstract: **BACKGROUND:** Although studies have examined the prevalence of spina bifida (SB) among births, little is known about the SB prevalence among children and adolescents. We estimated the prevalence of SB among children and adolescents in metropolitan Atlanta. **METHODS:** This study used data from a population-based registry of birth defects, with information on children with SB (cases) born in five Atlanta counties from 1979-2002. The population at risk was derived from United States Census data and variations in SB prevalence were examined by race/ethnicity, sex, lesion level, age group under 20 years, 4-year birth cohort, and time period using Poisson regression. **RESULTS:** From 1979 to 2002, SB birth prevalence decreased from 6.3 to 3.2 per 10,000 live births ($p < 0.001$) and SB prevalence within each age group also declined. In 2002, there were 211 children 0-19 years old surviving with SB in Atlanta (2.4 per 10,000 children 0-19 years old); prevalence of SB was higher among non-Hispanic whites and among children with lumbosacral lesion but did not vary by sex. With the exception of the most recent birth cohort (1998-2002), within each 4-year birth cohort, the prevalence of SB was generally higher among non-Hispanic whites than among non-Hispanic blacks. **CONCLUSIONS:** This study provides minimum prevalence estimates among children and adolescents with SB in metropolitan Atlanta, and identifies race/ethnic disparities in such prevalence estimates. This information could be useful for assessing the specialized health care needs for children with SB and the possible reasons for the racial/ethnic variation in prevalence of SB.

[This study documents that from 1979 to 2002, the birth prevalence of spina bifida decreased from 6.3 to 3.2 per 10,000 live births. Whites had a higher prevalence than Blacks. These data provide baseline numbers against which future prevalences can be compared.]

Sutton M, Daly LE, Kirke PN. Survival and disability in a cohort of neural tube defect births in Dublin, Ireland. *Birth Defects Res A Clin Mol Teratol.* 2008 Oct;82(10):701-9.

Abstract: **BACKGROUND:** Neural tube defects (NTDs) are a major cause of death and disability. Periconceptional folic acid prevents up to 70% of these malformations but public health campaigns to increase use of supplements have had disappointing results: The proposed mandatory fortification of bread products in Ireland has raised concerns about possible side effects. We examined data collected on a cohort of children born with NTDs in an era before fortification/supplementation to illustrate the serious consequences in terms of survival and disability. **METHODS:** All 623 infants born with NTDs in the Dublin area between 1976 and 1987 were included. Information was collected on mortality and length of survival for those who died, and for those who survived at least 5 years, interview-based data were collected on age, place of residence, prevalence of hydrocephalus, degree of disability, schooling, and IQ. **RESULTS:** Seventy-four births

(12%) were stillborn. Of the livebirths only 41% were alive at 5 years. Factors associated with mortality were type of lesion, level of lesion, presence of other defects, hydrocephalus, year of birth, and gestation. Of the children who survived to 5 years or more, 75% had a disability and 56% were severely disabled. Type of lesion and level of lesion influenced disability risk. Of the survivors, 51% of children had mobility limitations, 59% were incontinent, 42% had hydrocephalus, and 17% had intellectual disability. CONCLUSIONS: These findings illustrate the devastating consequences of NTDs and underline the importance of effective intervention programs with folic acid for prevention.

[This study presents findings from a large, complete, and unselected cohort of infants born with NTDs in the greater Dublin area born between 1976 and 1987. 41% of the children survived to 5 years and at follow-up, survivors had undergone a mean of five surgical operations since birth, surgical repair having been performed in 98% and a shunt insertion in 39%. More than half of the survivors had a severe disability, with almost 80% of children with meningomyelocele having a "severe disability." Children born later in the study had a better prognosis.]

Exercise/Fitness

Buffart LM, van der Ploeg HP, Bauman AE, Van Asbeck FW, Stam HJ, Roebroek ME, van den Berg-Emons RJ. Sports participation in adolescents and young adults with myelomeningocele and its role in total physical activity behaviour and fitness. *J.Rehabil.Med.* 2008 Oct;40(9):702-8.

Abstract: OBJECTIVE: To assess sports participation in young adults with myelomeningocele and its association with personal, disease-related and psychosocial factors, physical activity and fitness. DESIGN: Cross-sectional study. SUBJECTS: Fifty-one persons (26 males) with myelomeningocele, mean age 21.1 (standard deviation 4.5) years. METHODS: We assessed self-reported sports participation, ambulatory status, presence of hydrocephalus, functional independence, social support, perceived competence, exercise enjoyment, objective and self-reported physical activity, peak oxygen uptake, muscle strength and body fat. Associations were studied using regression analyses. RESULTS: Thirty-five subjects (69%) participated in sports. Sports participation was not associated with disease-related characteristics, but was associated with social support from family, perceived athletic competence and physical appearance ($p < \text{or} = 0.05$), and tended to be associated with global self-worth ($p = 0.10$). Sports participants had higher self-reported physical activity levels than non-participants ($p < \text{or} = 0.05$); objective results did not support this. Furthermore, sports participants tended to be less likely to have subnormal muscle strength (odds ratio = 0.26; $p = 0.08$) and their peak oxygen uptake was 0.19 l/min higher, but not statistically significantly ($p = 0.13$). CONCLUSION: Sports participation seems to be due to personal preferences rather than physical ability; it could benefit from improving social support and perceived competence, and is associated with higher self-reported physical activity.

[Increasing evidence indicates that maintaining a minimum level of physical activity is critical for the health of adults without disabilities. This has not been emphasized in adults with spina bifida but is now getting increasing attention. This article from the Netherlands showed that participation in sports was associated with family support and perceived athletic competence. For many reasons, we should be encouraging adults with spina bifida to participate in physical activities.]

Buffart LM, van den Berg-Emons RJ, Burdorf A, Janssen WG, Stam HJ, Roebroek ME. Cardiovascular disease risk factors and the relationships with physical activity, aerobic fitness, and body fat in adolescents and young adults with myelomeningocele. *Arch.Phys.Med.Rehabil.* 2008 Nov;89(11):2167-73.

Abstract: OBJECTIVES: To describe cardiovascular disease (CVD) risk factors in adolescents and young adults with myelomeningocele (MMC) and to explore relationships with physical activity, aerobic fitness, and body fat. DESIGN: Cross-sectional study. SETTING: Outpatient clinic. PARTICIPANTS: Adolescents and young adults (N=31) with MMC (58% men) age 16 through 30 years; 13 were ambulatory and 18 were nonambulatory. INTERVENTIONS: Not applicable. MAIN OUTCOME MEASURES: We studied biologic and lifestyle-related CVD risk factors, including lipid and lipoprotein profiles, blood pressure, aerobic fitness ($\dot{V}O_2$ peak), body fat, daily physical activity, and smoking behavior. We considered subjects at increased CVD risk when 2 or more of the following risk factors clustered: systolic blood pressure, total serum cholesterol (TC), high-density lipoprotein cholesterol (HDL-C), and cigarette smoking. Relationships were studied using regression analyses. RESULTS: Levels of TC, low-density lipoprotein cholesterol, and triglycerides were elevated in 29%, 38%, and 3% of the participants, respectively. HDL-C was reduced in 19%. Hypertension was found in 20%, and 19% were current cigarette smokers. Based on the clustering of risk factors, 42% of the participants were at increased CVD risk: 15% of ambulatory participants and 61% of nonambulatory participants ($P=.03$). Adjusted for sex and ambulatory status, participants with higher aerobic fitness tended to be more likely to have no CVD risk (odds ratio=13.0; $P=.07$). CVD risk was not associated to physical activity and body fat. CONCLUSIONS: A large proportion of the study sample was at CVD risk, indicated by clustering of risk factors. Improving aerobic fitness in young adults with MMC may contribute in reducing CVD risk; this needs to be confirmed in future studies.

[This study from the Netherlands found that 42% of the teenagers and young adults were at increased risk for cardiovascular disease based on criteria like blood pressure and lipid profile. As in persons without spina bifida, those who were more fit physically were less likely to be at increased risk. Studies documenting the effect of aerobic fitness programs in people with spina bifida on cardiovascular risk factors need to be conducted.]

De Groot JF, Takken T, Schoenmakers MA, Vanhees L, Helders PJ. Limiting factors in peak oxygen uptake and the relationship with functional ambulation in ambulating children with spina bifida. *Eur.J.Appl.Physiol* 2008 Nov;104(4):657-65.

Abstract: The objective of this study is to interpret the outcomes of peak oxygen uptake ($\dot{V}O_2$ peak) in children with SB and explore the relationship between $\dot{V}O_2$ peak and functional ambulation using retrospective cross-sectional study. Twenty-three ambulating children with SB participated at Wilhelmina's Children's Hospital Utrecht, the Netherlands. $\dot{V}O_2$ peak was measured during a graded treadmill-test. Eschenbacher's and Maninna's algorithm was used to determine limiting factors in reaching low $\dot{V}O_2$ peak values. Energy expenditure during locomotion (both \dot{O}_2 rate and \dot{O}_2 cost) and percentage of $\dot{V}O_2$ peak and HR(peak) were determined during a 6-min walking test (6MWT). Differences between community and normal ambulators were analyzed. $\dot{V}O_2$ peak, $\dot{V}O_2$ peak/kg, HR(peak), RER(peak) and VE (peak) were significantly lower compared to reference values, with significant differences between normal and

community ambulators. Limiting factors according to the algorithm were mostly "muscular and/or deconditioning" (47%) and ventilatory "gasexchange" (35%). Distance walked during 6MWT was 48.5% of predicted distance. Both $\dot{V}O_2$ rate and $\dot{V}O_2$ cost were high with significant differences between normal and community ambulators [17.6 vs. 21.9 ml/(kg min) and 0.27 vs 0.43 ml/(kg m)]. Also %HR(peak) and % $\dot{V}O_2$ (peak) were significantly higher in community ambulators when compared to normal ambulators (resp. 97.6 vs. 75% and 90.2 vs. 55.9%). $\dot{V}O_2$ (peak) seems to be mostly limited by deconditioning and/or muscular components and possible ventilatory factors. For both peak values and functional ambulation, community ambulators were significantly more impaired than normal ambulators. High energy expenditure, % $\dot{V}O_2$ (peak) and %HR(peak) reflect high level of strain during ambulation in the community ambulators. Future exercise testing in children with SB should include assessment of ventilatory reserve. Exercise training in ambulatory children should focus on increasing both $\dot{V}O_2$ (peak) and muscular endurance, as well as decreasing energy cost of locomotion.

[This article found that most children with spina bifida were "out of shape" (deconditioned). Some also had lung issues that limited their endurance. The authors encourage more exercise training in these children.]

Fragala-Pinkham M, Haley SM, O'Neil ME. Group aquatic aerobic exercise for children with disabilities. *Dev.Med.Child Neurol.* 2008 Nov;50(11):822-7.

Abstract: The effectiveness and safety of a group aquatic aerobic exercise program on cardiorespiratory endurance for children with disabilities was examined using an A-B study design. Sixteen children (11 males, five females) age range 6 to 11 years (mean age 9y 7mo [SD 1y 4mo]) participated in this twice-per-week program lasting 14 weeks. The children's diagnoses included autism spectrum disorder, myelomeningocele, cerebral palsy, or other developmental disability. More than half of the children ambulated independently without aids. Children swam laps and participated in relay races and games with a focus of maintaining a defined target heart rate zone. The strengthening component consisted of exercises using bar bells, aquatic noodles, and water resistance. The following outcomes were measured: half-mile walk/run, isometric muscle strength, timed floor to stand 3-meter test, and motor skills. Complaints of pain or injury were systematically collected. Significant improvements in the half-mile walk/run were observed, but not for secondary outcomes of strength or motor skills. The mean program attendance was 80%, and no injury was reported. Children with disabilities may improve their cardiorespiratory endurance after a group aquatic aerobic exercise program with a high adult:child ratio and specific goals to maintain training heart rates.

[This study showed that a group-aquatic-aerobic exercise program was safe and well tolerated by children with disabilities, including those with spina bifida. It also encourages social participation and is fun. This is one way in which children with spina bifida can increase their physical activity, which, theoretically, could have long-term health benefits.]

Experimental/Animal

Amouee A, Memarzadeh M, Ashrafi M, Farid M, Sanei MH, Soroori S, Vahed E, Yadegari M. The effects of amniotic fluid on the histopathologic changes of exposed spinal cord in fetal sheep. *Arch.Iran Med.* 2009 Jan;12(1):35-40.

Abstract: BACKGROUND: Experimental studies have shown that in myelomeningocele,

the primary malformation is neural tissue damage resulting from exposure of neural tissue to amniotic fluid. In this study, the effects of amniotic fluid on histopathologic changes of exposed spinal cord in fetal sheep were evaluated. **METHODS:** In an experimental trial, 10 fetal sheep in two groups containing five subjects (group A) and five shams (group B) were studied. In the sheep at 90 - 100 days of gestation (term: 145 - 150 days) the lumbar skin was incised, paraspinal soft tissues were excised, laminectomy was performed at L2 - L4, and dura matter was opened. In group A, the dura matter was not dorsally closed and thus the spinal cord was left exposed to amniotic fluid, and in group B the skin was immediately closed. The lambs were delivered near term by cesarean section and were assessed clinically and morphologically. **RESULTS:** In group A, all lambs (n=5) had a complete or incomplete flaccid sensorimotor paraplegia and suffered from urine incontinence. Four lambs in this group were stool incontinent. In group B (n=4), only one lamb had paraparesis (P=0.048) and all lambs were urine and stool continent. In group A, all lambs had hypoplastic longitudinal muscles of the rectum but well-developed circular muscles. The anal sphincter muscles did not develop normally. In group B, all lambs had well-developed longitudinal and circular muscles and anal sphincter muscles developed normally (P=0.048). Histopathologic examination of the spinal cords showed edema, focal calcification, fibrosis, and capillary cell proliferation in group A, but in group B such changes were not seen. The number of ganglion cells was significantly higher in group B compared with group A (P<0.0005). **CONCLUSION:** Exposure of spinal cord to amniotic fluid causes structural neural tissue damage that can be prevented by fetal surgery through repairs of myelomeningocele.

[Prenatal surgery to close meningomyeloceles was inspired by research in sheep suggesting that amniotic fluid damages nerves in the open lesions. This recent study confirms that theory, and suggests that prenatal surgery could prevent some of this damage and perhaps improve neurological outcomes. The MOMS study has yet to be published. Stay tuned.]

Cecconi F, Piacentini M, Fimia GM. The involvement of cell death and survival in neural tube defects: a distinct role for apoptosis and autophagy? *Cell Death.Differ.* 2008 Jul;15(7):1170-7.

Abstract: Neural tube defects (NTDs), such as spina bifida (SB) or exencephaly, are common congenital malformations leading to infant mortality or severe disability. The etiology of NTDs is multifactorial with a strong genetic component. More than 70 NTD mouse models have been reported, suggesting the involvement of distinct pathogenetic mechanisms, including faulty cell death regulation. In this review, we focus on the contribution of functional genomics in elucidating the role of apoptosis and autophagy genes in neurodevelopment. On the basis of compared phenotypical analysis, here we discuss the relative importance of a tuned control of both apoptosome-mediated cell death and basal autophagy for regulating the correct morphogenesis and cell number in developing central nervous system (CNS). The pharmacological modulation of genes involved in these processes may thus represent a novel strategy for interfering with the occurrence of NTDs.

[Spina bifida is the most complex birth defect compatible with life. It appears that the mechanisms that cause it are equally complex. This study examines the role of cell death, which is a normal biological process during development, but seems to be out of control in animal models of spina bifida.]

Encinas Hernandez JL, Soto C, Garcia-Cabezas MA, Pederiva F, Garriboli M, Rodriguez R, Peiro JL, Carceller F, Lopez-Santamaria M, Tovar JA. Brain malformations in the sheep model

of myelomeningocele are similar to those found in human disease: preliminary report. *Pediatr.Surg.Int.* 2008 Dec;24(12):1335-40.

Abstract: **PURPOSE:** To examine if brain malformations, similar to those which account for cognitive disorders seen in human disease, are present in an ovine model of myelomeningocele (MMC). **METHODS:** An MMC-like lesion was surgically created in 16 fetal lambs between 60 and 80 days of gestation. Ten did not undergo fetal repair (group A), 2 were repaired with an open two-layer closure (group B), 2 with open biogluce coverage (group C) and 2 with fetoscopic coverage (group D). Lambs were killed and their brains were examined. Two brains from normal unoperated lambs served as controls. **RESULTS:** Thirteen lambs died in utero (81%). Two lambs in group A and 1 in group B were delivered at term. Group A brains showed hydrocephalus and extensive areas of polymicrogyria. There was also an extensive denudation of the ependymal lining under the polymicrogyric areas and the corpus callosum was thinner than normal. No hindbrain herniation was observed. Brains from group B and the control did not show any of these abnormalities. **CONCLUSIONS:** Some of the central nervous system abnormalities associated to MMC in human patients are also found in the uncorrected fetal lamb model of MMC but not in the only survivor to intrauterine coverage. Further studies are necessary to ascertain if these abnormalities can be prevented by coverage of the defect.

[In this study creating a “hole” in the prenatal spine like that seen with spina bifida leads to changes in the brain including hydrocephalus, abnormalities of the folds in the brain (gyri) and even changes in the corpus callosum. This suggests that prenatal surgery can prevent some of these changes. The MOMS study has yet to be published. Stay tuned.]

Fauza DO, Jennings RW, Teng YD, Snyder EY. Neural stem cell delivery to the spinal cord in an ovine model of fetal surgery for spina bifida. *Surgery* 2008 Sep;144(3):367-73.

Abstract: **BACKGROUND:** We introduce the notion of prenatal neural stem cell (NSC) delivery to the spinal cord as an adjuvant to fetal repair of spina bifida. **METHODS:** Fetal lambs with experimental myelomeningocele (MMC; n = 25) were divided in 3 groups: group I, no repair; group II, standard surgical MMC coverage; and group III, MMC coverage plus delivery of a murine NSCs clone into the spinal cord defect. Donor cells constitutively expressed lacZ encoding for Escherichia coli beta-galactosidase, yet they were further labeled by exposure to either BrdU and/or to the fluorescent membrane dye PKH-26. Blinded initial clinical evaluations and multiple spinal cord analyses were undertaken soon after birth. **RESULTS:** Both survival and the incidence of major paraparesis were significantly worse in group I compared with groups II and III. In group III, NSC density was highest within the most damaged areas of the spinal cord, with selective engraftment within those regions. Donor NSCs retained an undifferentiated state in vivo, producing neurotrophic factors within the defect. No animals in group III had a worsened condition following this intervention. **CONCLUSIONS:** Neural stem cells retain an undifferentiated state and produce neurotrophic factors in the short term after delivery to the fetal spinal cord, in the setting of experimental MMC. Further scrutiny of NSC delivery to the spinal cord as a therapeutic strategy against spina bifida is warranted.

[This is an exciting study in which stem cells were placed prenatally into the spines of unborn lambs who had experimental spina bifida. The stem cells collected in the area of the defect and produced chemicals that allow nerve cells to grow. Whether this treatment will lead to better neurological outcomes—even in sheep—remains to be seen.]

Gastroenterology

Bani-Hani AH, Cain MP, King S, Rink RC. Tap water irrigation and additives to optimize success with the Malone antegrade continence enema: the Indiana University algorithm. *J.Urol.* 2008 Oct;180(4 Suppl):1757-60.

Abstract: **PURPOSE:** We report the long-term effectiveness of standard tap water for Malone antegrade continence enema irrigation as well as our algorithm for managing refractory constipation/fecal incontinence in a large single institution experience. **MATERIALS AND METHODS:** We retrospectively reviewed the charts of 256 Malone antegrade continence enema procedures performed for chronic constipation and/or incontinence due to neuropathic bowel. Continence, type of fluid used to irrigate the colon, volume of flushes and the need for additives were recorded and a database was created. All patients were initially treated with tap water irrigation. Those in whom tap water irrigation failed underwent complete bowel cleanout with enemas and GoLYTELY via the Malone antegrade continence enema, followed by a gradual increase in irrigation volume. If this was unsuccessful, additives of mineral oil, MiraLAX or glycerin were added to the irrigant daily. **RESULTS:** A total of 236 patients with at least 6 months of followup were included in this study. Mean age at surgery was 10.2 years (range 2 to 36) and mean followup in the entire cohort was 50 months (range 6 to 115). Mean volume of colonic flushes was 642 ml (range 100 to 1,000). Of the patients 196 (83.1%) achieved total fecal continence with tap water flushes alone. Using additives increased the overall continence rate to 93.6% ($p < 0.0001$). **CONCLUSIONS:** The Malone antegrade continence enema procedure has proved invaluable for treating children with refractory constipation. When additives are used in conjunction with water flushes, they can significantly improve the overall fecal continence rate in partially continent children.

[The antegrade continence enema (ACE) procedure has been shown to help maintain continence in children with spina bifida. In this study, 83% of the children who had the Malone ACE were able to maintain continence by using tap water to flush out their bowels through the opening in the abdominal wall. An additional 11% needed another substance (like MiraLax or mineral oil) to maintain continence.]

Becmeur F, Demarche M, Lacreuse I, Molinaro F, Kauffmann I, Moog R, Donnars F, Rebeuh J. Cecostomy button for antegrade enemas: survey of 29 patients. *J.Pediatr.Surg.* 2008 Oct;43(10):1853-7.

Abstract: **OBJECTIVE:** This study evaluated the Trap-door button use (Cook Medical, Bloomington, IL) for antegrade enemas in children. **METHODS:** Since 2002, patients with fecal incontinence or encopresis and constipation underwent percutaneous cecostomy under laparoscopy using a button. Technical details are described. Age at surgery, operative time, hospital stay, diagnosis, indications for cecostomy, and duration of follow-up were recorded. A survey was proposed via a questionnaire that was sent to the patients. Patients wearing the button for less than 1 month were excluded from this evaluation. The survey concerned volume and frequency of enemas, difficulties encountered, benefits and disadvantages of this method, and assessment of the antegrade enemas in continence. **RESULTS:** Twenty-nine patients, 18 males and 11 females, aged 3 to 21 years (mean, 8.5 years) underwent laparoscopic Trap-door button placement. The indications for all the patients were intractable fecal incontinence in 24 cases and constipation with encopresis in 5 cases. Incontinence was because of myelomeningocele

(n = 10), anorectal malformations (n = 11), caudal regression syndrome (n = 1), 22q11 syndrome (n = 1), and Hirschsprung disease with encephalopathy with convulsions (n = 1). Constipation with encopresis was because of sacrococcygeal teratoma (n = 1), cerebral palsy (n = 1), and acquired megarectum with psychiatric and social disorders (n = 3). A total of 26 cecostomy button placements and 3 sigmoidostomy button placements were successful with no intraoperative complication. The mean operative time was 25 minutes (10-40 minutes), and the hospital stay was 2.5 days (1-4 days). Twenty-two parents or patients answered the questionnaire. At the time of this survey, 2 patients had improved their fecal continence and had had the button removed. A mean of 4 weekly enemas was enough to improve fecal continence troubles (range, 1 daily to 1 for 2 weeks). The volume for enemas was 250 to 1000 mL (mean, 700 mL). The time required for the irrigation of the bowel by gravity took from 5 to 60 minutes (mean, 25 minutes) for 20 patients. Before surgery, 14 patients needed a diaper, day and night, and 6 needed sanitary protection. Soiling was a very significant inconvenience for all the patients. After surgery, only 5 patients needed a diaper (cerebral palsy, 22q11, cloacal malformation, myelomeningocele, bladder exstrophy) because of moderate results or urinary incontinence and continued soiling. Patients were asked to give an assessment (null = 0, bad = 1, fair = 2, good = 3, very good = 4). None of the patients felt there had been no changes or a bad result. There were 5 patients who felt they had an average result, 5 a good result, and 12 a very good result. The mean grade was 3.44 (17.2/20). A total of 3 patients had hypertrophic granulation tissue formation around the cecostomy button, and 12 had tiny leakage. **CONCLUSION:** Percutaneous placement of a cecostomy button under laparoscopic control is an easy and major complication-free procedure. The use of the Trap-door device by the patients or with the help of the parents for antegrade enemas is effective and satisfactory. It improves the quality of life and is reversible.

[A number of procedures are used to place the antegrade continence enema. In this study, laparoscopic placement of the Cook Medical Trap-door button was used in 10 children with meningomyelocele. The procedure was well tolerated and the results were comparable to the success rates documented in other studies.]

Del PG, Mosiello G, Pilati C, Lamartina M, Battaglino F, Buffa P, Redaelli T, Lamberti G, Menarini M, Di BP, et al. Treatment of neurogenic bowel dysfunction using transanal irrigation: a multicenter Italian study. *Spinal Cord*. 2008 Jul;46(7):517-22.

Abstract: **STUDY DESIGN:** Thirty-six patients with unsatisfactory treatment of neurogenic bowel dysfunctions (NBD) were enrolled from Spinal Units and Rehabilitation Centers in Italy. Treatment was for 3 weeks using a newly developed integrated system with an enema continence catheter for transanal irrigation (Peristeen, Coloplast A/S Kokkedal Denmark). **OBJECTIVES:** To evaluate the effects of Peristeen Anal Irrigation on NBD and patient quality of life (QoL). **SETTING:** Italy. **METHODS:** Lesion level, ambulatory status and hand functionality were determined in all patients. NBD symptoms and QoL were evaluated before and after treatment, using a specific questionnaire. Statistical analysis was performed using McNemar Test and Sign Test. **RESULTS:** Thirty-six patients were enrolled, and 32 patients completed the study. At the end of the treatment, 28.6% of patients reduced or eliminated their use of pharmaceuticals. Twenty-four patients became less dependent on their caregiver. There was a significant increase in patients' opinion of their intestinal functionality (P=0.001), QoL score (P=0.001) and their answers regarding their degree of satisfaction (P=0.001). A successful outcome was recorded for 68% of patients with fecal incontinence, and for 63% of patients with constipation. **CONCLUSION:** Peristeen Anal Irrigation is a simple

therapeutic method for managing NBD and improving QoL. It should be considered as the treatment of choice for NBD, playing a role in the neurogenic bowel analogous to that of intermittent clean catheterization in bladder treatment.

[This study describes the use of an old technique in which a catheter with an attached balloon is inserted into the rectum. The balloon is blown up, which keeps the catheter in place, and an enema is given through the catheter. The 12 children with spina bifida enrolled in this study generally tolerated this procedure and had better continence afterwards. It is one of many methods that can be helpful improve fecal continence.]

Montes-Tapia F, Cura-Esquivel I, Garza-Luna U, Martinez-Flores G, Munoz-Maldonado G, brego-Moya V. Sigmoid fixation associated with rectopexy using a laparoscopic approach could prevent relapse of rectal prolapse in pediatric patients with spinal dysraphia. *J.Pediatr.Surg.* 2008 Aug;43(8):1551-3.

Abstract: Recurrent rectal prolapse, resistant to medical treatment, is an indication for surgical treatment. Patients with spinal dysraphia frequently have already been treated by sclerotherapy or other surgical techniques, but unsuccessfully. **METHODS:** We present 2 patients, who underwent laparoscopic rectopexy, with spinal dysraphia and complete rectal prolapse relapse after conservative treatment. In these patients, we performed, as an additional technique, fixation of the rectosigmoid to avoid recurrence by invagination or prolapse of the anterior wall. **RESULTS:** Follow-up at 14 and 11 months, respectively, did not find any recurrence. **CONCLUSION:** We suggest that laparoscopic rectopexy with sigmoid fixation should be considered as an alternative for the treatment for patients with spinal dysraphia and rectal prolapse to avoid recurrence.

[Recurrent rectal prolapse—in which part of the rectum protrudes through the anus—is not an uncommon problem in children with spina bifida. This study describes a surgical technique used in two children who had intractable prolapse.]

Nanigian DK, Nguyen T, Tanaka ST, Cambio A, DiGrande A, Kurzrock EA. Development and validation of the fecal incontinence and constipation quality of life measure in children with spina bifida. *J.Urol.* 2008 Oct;180(4 Suppl):1770-3.

Abstract: **PURPOSE:** Fecal incontinence and constipation in children with spina bifida are recognized to impact quality of life. Most disease specific quality of life instruments on fecal incontinence target adults and/or children without neuropathic bowel. We developed an instrument to evaluate bowel function and its impact on quality of life in children with spina bifida and their caregivers. **MATERIALS AND METHODS:** A 51-item questionnaire termed the FIC QOL (Fecal Incontinence and Constipation Quality of Life) survey was developed from expert opinion, patient interviews, and modification of previously published adult and pediatric studies for nonneuropathic bowel dysfunction. The items are divided into 7 quality of life factor groupings, including bowel program, dietary management, symptoms, travel and socialization, family relationships, caregiver emotional impact and financial impact. The questionnaire was given to caregivers of children with and without spina bifida. Discriminant validity was evaluated by comparing the spina bifida and control groups. Test-retest reliability was evaluated by having 41 patients complete 2 surveys within 4 to 6 weeks. **RESULTS:** Comparing questionnaires from 92 index patients and 52 controls showed a statistically significant difference for all 7 quality of life factor groupings. The FIC QOL instrument objectively demonstrated the negative impact of fecal incontinence and constipation on quality of life in these families.

Comparing 82 questionnaires at 2 time points demonstrated the reliability of all FIC QOL questions. CONCLUSIONS: The FIC QOL instrument provides a valid and reliable measure of the effect of fecal incontinence and constipation on the quality of life of caregivers and their children with spina bifida.

[This study describes a new instrument that measures the effects of constipation and fecal incontinence on the quality of life of children with spina bifida.]

Shoshan L, Ben-Zvi D, Katz-Leurer M. Use of the anal plug in the treatment of fecal incontinence in patients with meningomyelocele. *J.Pediatr.Nurs.* 2008 Oct;23(5):395-9.

Abstract: BACKGROUND: Children with meningomyelocele (MMC) commonly present with urinary and fecal incontinence. Despite bowel training and enemas, many still suffer from fecal soiling, which may cause skin irritation and malodor. Fear and embarrassment may cause some of these children to avoid partaking in social activities. The anal plug (AP), an anal tampon, has been developed to prevent fecal soiling. The purpose of this study was to evaluate the use of the AP in children with MMC with regard to reduction of fecal soiling and impact on social functioning. METHODS: Twenty children and young adults with MMC, neurological level L(5) or proximal, participated in the study. MAIN OUTCOME MEASURES: Number of incidents of fecal soiling, degree of comfort of the accessory of choice, and impact of fecal soiling on social functioning were used as the main outcome measures for this study. INTERVENTION: Each participant kept a daily diary reporting on each of the outcome measures over the course of 5 weeks. The first week, prior to intervention with the AP, was used as the base control for each patient. Over three sessions, the use of the AP was explained and demonstrated, after which it was given to the participants to be used. RESULTS: Seventy-five percent of the participants completed the study. There was a significant improvement in all outcome parameters following the intervention. The median number of weekly incidents of soiling dropped from 4 to 0 ($p = .002$). The median reported effect of fecal soiling on social integration before the use of the AP was found to be very bothersome, whereas during the use of the AP, it was barely bothersome ($p = .004$). CONCLUSION: Use of the AP in children and young adults with MMC can prevent fecal soiling and promote "social continence."

[The rectal sphincters are really "smart" because they allow people to release gas while retaining solid and semi-liquid stool. In this study, an old treatment, the anal plug, was used in children and young adults with spina bifida to help maintain fecal continence. The plug literally plugs the anus and does not allow anything—even gas—to escape. While this intervention can be very effective for the short term—for example going out to a social event—it does not allow the escape of gas and can lead to abdominal cramping if used for an extended time. It should be considered as part of an overall program of bowel management.]

General

Baradaran N, Ahmadi H, Nejat F, El KM, Mahdavi A. Nonneural congenital abnormalities concurring with myelomeningocele: report of 17 cases and review of current theories. *Pediatr.Neurosurg.* 2008;44(5):353-9.

Abstract: OBJECTIVE: Meningomyelocele (MMC) is a common central nervous system birth defect. Various congenital and acquired abnormalities have been reported with MMC, some of which are secondary to the pathophysiology and some are morbidities of

the underlying disease. The aim of this study was to discuss current possible theories explaining diverse anomalies/abnormalities seen in a series of 390 patients with MMC. METHODS: A retrospective study was performed using the records of 390 patients with MMC at Children's Hospital Medical Center in Tehran, Iran, from January 2001 to January 2007. A series of 17 cases of MMC with attributed organ anomalies, not explained by a causal effect of the underlying disorder, were compiled. There were 3 cardiac anomalies including ventricular septal defect, pulmonary artery atresia and tetralogy of Fallot, 4 musculoskeletal malformations, consisting of missing rib, polydactyly and complex distal limb anomaly, 4 urological anomalies such as bladder exstrophy, horseshoe kidney and dysplastic kidneys, 2 occipital encephaloceles, 2 congenital adrenal hyperplasia patients with ambiguous genitalia, 1 omphalocele, 1 albinism and 1 Klippel-Feil syndrome. A review of the literature and discussion explaining each of these observations, have been performed and some possible theories have been proposed. CONCLUSIONS: Although various organ anomalies with different embryological origin had been observed and reported with MMC, it is difficult to explain their development using one of the current theories of MMC formation. It could be attributed to a possible genetic defect or merely an incidental finding. A teratological insult during the embryogenic phase would be an alternative assumption.

[This study from Iran documents what previous studies have shown: that many children with meningomyelocele also have other birth defects. A unifying theory to explain these findings has not been found.]

Cabrera RM, Shaw GM, Ballard JL, Carmichael SL, Yang W, Lammer EJ, Finnell RH. Autoantibodies to folate receptor during pregnancy and neural tube defect risk. *J.Reprod.Immunol.* 2008 Oct;79(1):85-92.

Abstract: Periconceptional folic acid can reduce the occurrence of neural tube defects (NTDs) by up to 70%, and autoantibodies for folate receptors (FRs) have been observed in serum from women with a pregnancy complicated by an NTD. This population-based cohort study has examined serum from pregnant mothers for autoantibodies to FRs, antibodies to bovine folate binding protein (FBP), and inhibition of folic acid binding to FR and FBP in association with NTD risk. The mid-gestational maternal serum specimens used for this study were collected during the 15-18th week of pregnancy. Samples were obtained from the California Birth Defects Monitoring Program; 29 mothers had a pregnancy complicated by spina bifida and 76 mothers had unaffected children. The presence of IgG and IgM antibodies to human FR, bovine FBP, and inhibition of folic acid binding to FR and FBP was determined. Higher activity of IgM to FBP in cases versus controls was observed ($P=0.04$). Higher activity of IgM and IgG autoantibodies to FR was observed ($P<0.001$ and $P=0.04$, respectively). Risk estimates at two standard deviations above average control antibody concentrations were $OR=2.07$ ($CI=1.02, 4.06$) for anti-FBP IgM, $OR=2.15$ ($CI=1.02, 4.69$) for anti-FR IgG and $OR=3.19$ ($CI=1.47, 6.92$) for anti-FR IgM. These data support the hypothesis that high titers of antibodies and blocking of folic acid binding to FRs by maternal serum should be regarded as risk factors for NTDs.

[Some mothers who deliver children with spina bifida have elevated levels of antibodies that partially block the effects of folic acid. This study provides additional data to support this association. Whether screening women for antibodies to folate receptors will decrease the occurrence of having a child with a neural tube defect remains to be seen.]

Chang CK, Wong TT, Huang BS, Chan RC, Yang TF. Spinal dysraphism: a cross-sectional and retrospective multidisciplinary clinic-based study. *J.Chin Med.Assoc.* 2008 Oct;71(10):502-8.

Abstract: **BACKGROUND:** Spinal dysraphism is a common birth defect that causes different kinds of secondary impairments, including joint deformities, reduced mobility, and bowel/bladder dysfunction. Due to the diversity in terminology, cultural/ethnic differences, and medical policies, prior study results cannot be generalized to all populations. Therefore, we performed this study to define the characteristics of patients in Taiwan with spinal dysraphism. **METHODS:** Patients diagnosed with a myelomeningocele or lipomyelomeningocele were identified from the database of our spinal dysraphism multidisciplinary clinic. A cross-sectional study was conducted by telephone interview and retrospective chart review. Clinical characteristics, such as neurologic level, orthopedic deformities, assistive device use, and level of ambulation, were collected. Spearman's correlation (r) tests were performed between ambulation or neurologic level and other variables. **RESULTS:** Seventy-eight subjects were included in the current study. Subjects with myelomeningoceles had more severe neurologic involvement, poorer ambulation outcome, and higher rates of orthopedic deformities, assistive device use, lower hand function, and bowel/bladder dysfunction. The correlation test revealed that the level of ambulation was negatively influenced by a higher neurologic level, a history of shunt placement, and various orthopedic deformities. Neurologic level also had widespread influence on history of shunt placement, orthopedic deformities, assistive device use, the need for additional assistive devices, aggressiveness of assistive devices, and bowel/bladder dysfunction. **CONCLUSION:** For patients with spinal dysraphism, the neurologic level is the most important prognostic factor for many other clinical characteristics, including ambulation status.

[This study describes a spina bifida program in Taiwan. Although the findings are similar to those published from other programs, including those in the USA, it is important to have data from all around the world.]

Dosa NP, Foley JT, Eckrich M, Woodall-Ruff D, Liptak GS. Obesity across the lifespan among persons with spina bifida. *Disabil.Rehabil.* 2008 Nov 26;1-7.

Abstract: **Purpose.** Identify risk factors for obesity across the lifespan for individuals with spina bifida. **Methods.** Cross sectional chart review study of 203 patients aged 6-58 years. Obesity was based on body mass index. Rates were calculated for children aged 6-11 years; adolescents aged 12-19 years and adults aged > 20 years. Chi-square analyses were used to determine differences in obesity rates among subgroups. An ordered logistic regression model was developed for the three age groups to estimate the probability of a change in BMI classification from normal weight to overweight or overweight to obese, controlling for sex, functional motor level, shunt status and insurance status. **Results.** Obesity rates for children, adolescents and adults were 18, 8 and 37%, respectively. Obesity rates were higher among adults ($\chi^2 = 27, p < 0.01$) and for individuals who were publicly insured ($\chi^2 = 7.2, p < 0.03$). The ordered regression model for children demonstrated no independent association between sex, shunt status, functional motor level or insurance status and change in BMI category. For adolescents, lower functional motor level (i.e. sacral) increased the risk of becoming obese (Odds Ratio: 2.13; 95% CI: 1.12-4.06; $p < 0.02$). Among adults, female sex increased risk (OR = 2.28; 95% CI: 1.03-5.04; $p < 0.04$). **Conclusions.** Obesity rates for children and adolescents with spina bifida are similar to the general population; however, obesity rates are higher among adults,

particularly women. Risk factors are similar to those observed in the general population. [This study found that the prevalence of obesity among adolescents and adults with spina bifida was similar to that found in the general population (and higher than it should be). Adult women were especially at risk. Programs to address obesity that include increased physical activity and nutritional interventions should be studied in these individuals.]

Idowu OE, Apemiye RA. Outcome of myelomeningocele repair in sub-Saharan Africa: the Nigerian experience. *Acta Neurochir.(Wien.)* 2008 Sep;150(9):911-3.

Abstract: **OBJECTIVE:** Early repair of myelomeningocele (MM) is associated with decreased morbidity and mortality. In sub-Saharan Africa, the peculiar harsh economic and social realities make late presentation, malnutrition and sepsis at presentation prevalent. As these factors may affect surgical repair, the aim of this study was to review the outcome of repair of MM in this peculiar patient population. **MATERIALS AND METHODS:** A prospective study of patients with MM who presented at the Lagos State University Teaching Hospital, Lagos, Nigeria over a 12 month period was studied. Follow up ranged from 8 weeks to 1 year. **RESULTS:** Five patients died while awaiting surgery. Thirty-six patients, 24 males and 12 female infants with MM were operated on during the study period. Majority of the patients were operated on or after the seventh day of life. The median age at surgery was 11th day of life (range second day-2 years). The mean surface area of the myelomeningocele was 26.4 cm² (range 12.6-62.9 cm²). The post-operative morbidity and mortality was five and one, respectively. **CONCLUSIONS:** Intracranial sepsis is the main cause of death in MM patients who are not operated on time. The patients we are seeing are probably those that survived to present at the hospital; by that time primary closure is usual without the need for complex plastic procedures. Delay in presentation even with associated malnutrition and local sepsis seems not to significantly affect the outcome of MM repair.

[This study describes the treatment of 41 selected children with meningocele in Nigeria. Five infants (12%) died while awaiting surgery; 26 (72%) had closure of their lesions after 7 days of age. Hydrocephalus was treated with endoscopic third ventriculostomy. These data are very important in providing a perspective of what is happening around the world with these children.]

Kulkarni AV, Cochrane DD, McNeely PD, Shams I. Medical, social, and economic factors associated with health-related quality of life in Canadian children with hydrocephalus. *J.Pediatr.* 2008 Nov;153(5):689-95.

Abstract: **OBJECTIVE:** To study the factors associated with health-related quality of life (HRQL) in Canadian children with hydrocephalus, using a comprehensive model of determinants of child health, including socioeconomic factors. **STUDY DESIGN:** A cross-sectional study was performed between November 2005 and November 2006 at 3 Canadian pediatric hospitals. Parents of children with hydrocephalus age 5 to 18 years completed the Hydrocephalus Outcome Questionnaire (HOQ) and the Health Utilities Index Mark 3 (HUI-3). **RESULTS:** A consecutive sample of 340 subjects participated from a total of 366 eligible children (mean age, 11.6 +/- 3.6 years; mean time from the diagnosis of hydrocephalus, 10.0 +/- 4.6 years). Adjusted multivariate linear regression models demonstrated that the most important determinants of poorer HRQL included lower family income, lower parental education, worse family functioning, seizures, myelomeningocele, and prolonged treatment for cerebrospinal fluid shunt obstruction.

CONCLUSIONS: Despite a national universal health care system, socioeconomic disparities remain important as determinants of HRQL. Given the absence of a parallel private health care system in Canada, this suggests that the impact of socioeconomic factors is related to issues other than access to care.

[This study evaluated 340 children with hydrocephalus, including 112 who had spina bifida. The primary outcome was health-related quality of life. The following factors predicted a worse outcome: lower income, lower parental education, worse family functioning, seizures, spina bifida, and prolonged treatment for cerebrospinal fluid shunt obstruction. Thus, as has been shown in the USA, lower socioeconomic status, as well as having spina bifida, have a negative impact on quality of life in children with hydrocephalus. Poverty and poor education remain major determinants of health and well-being all over the world.]

Peixinho C, Tavares-Ratado P, Tomas MR, Taborda-Barata L, Tomaz CT. Latex allergy: new insights to explain different sensitization profiles in different risk groups. *Br.J.Dermatol.* 2008 Jul;159(1):132-6.

Abstract: **BACKGROUND:** Differences in latex allergen sensitization profiles have been described between children subjected to repetitive surgical interventions and health care workers (HCW). 'Major' allergens for patients with spina bifida are Hev b 1, 3 and 7, while for HCW, 'major' allergens are Hev b 2, 5, 6.01 and 13. The reason for these differential sensitization profiles is currently unknown. **OBJECTIVES:** To investigate latex allergen profiles on internal and external surfaces of natural rubber latex gloves. **METHODS:** Eighty-two samples of commonly used surgical gloves (41 glove brands) were used for analysis. Specific allergen levels of Hev b 1, 3, 5 and 6.02 on both surfaces of the gloves were quantified using an enzyme immunoassay, a FITkit (FIT Biotech, Tampere, Finland). **RESULTS:** Differences in allergen levels were observed between internal and external surfaces of all glove types. Concentrations of Hev b 1 and Hev b 3 were significantly higher on external surfaces, while internal surfaces had higher allergen levels of Hev b 5 and Hev b 6.02. Analysis of surgical and examination gloves, powdered and nonpowdered gloves also showed that the content of Hev b 5 and Hev b 6.02 was significantly higher on internal surfaces while that of Hev b 1 and Hev b 3 was higher on external surfaces. **CONCLUSIONS:** Our study showed different allergen profiles on internal and external surfaces of natural rubber latex gloves. These results may suggest a relationship between latex allergen localization and sensitization routes in different risk groups.

[People with spina bifida have an increased occurrence of latex allergies. So do health care workers. This study evaluated the internal and external surfaces of latex gloves and found that they had different allergen profiles. The authors hypothesize that health care workers become sensitized by the allergens on the inside of gloves while people with spina bifida are sensitized to allergens on the external surface of latex gloves, e.g., during surgical and other procedures.]

Rademacher N, Black DP, Ulrich BD. Early spontaneous leg movements in infants born with and without myelomeningocele. *Pediatr.Phys.Ther.* 2008;20(2):137-45.

Abstract: **PURPOSE:** To compare quantity and quality of spontaneous leg movements during early infancy in babies with myelomeningocele (MMC) and babies with typical development (TD). **METHODS:** Nine infants with MMC and 12 with TD moved spontaneously while supine for 5 minutes at ages 1, 3, and 6 months. We used a 6-camera system to monitor leg movements. Resultant leg displacement and velocity were used to

determine movement frequency and, for each movement, duration, distance, peak velocity, jerk, and number of acceleration peaks. RESULTS: Movements of infants with MMC were shorter in duration with fewer acceleration peaks than their peers. Asymmetrical interlimb frequencies in infants with TD resulted in one leg moving more than the other, which was similar to the lower symmetrical interlimb frequencies of infants with MMC. CONCLUSIONS: Infants with MMC show depressed movement activity. Further research is needed to determine if therapy can facilitate spontaneous activity and leg control.

[This project studied the leg movements of infants born with meningomyelocele and found qualitative differences compared with the movements of otherwise healthy babies.]

Rivera AF, vila Torres RR, Parrilla Rodriguez AM, de L, I, Gorrin Peralta JJ. Exploratory study: knowledge about the benefits of breastfeeding and barriers for initiation in mothers of children with spina bifida. *Matern.Child Health J.* 2008 Nov;12(6):734-8.

Abstract: The aim of the study is to identify the level of knowledge on breastfeeding by mothers of infants with spina bifida and the barriers encountered in initiating breastfeeding. A non-probabilistic sample (n=30) of mothers was used in this study. The sample was obtained in two institutions specializing in care for infants with congenital anomalies in Puerto Rico. A self-administered questionnaire was used consisting of six sections. Descriptive statistics were used for data analysis. Among participants, 56.7% had adequate knowledge on the benefits of breastfeeding. The most frequently encountered barriers in initiating breastfeeding were related to the neonatal intensive care unit (NICU). Problems with the medical and nursing procedures, routines, support from personnel, and adequacy of the environment were the most frequent. It is important to develop education to encourage nurses and other health care professionals need suggest, support and encourage breastfeeding to mothers of infants with neural tube defects.

[The birth of a baby with meningomyelocele causes a great deal of stress. Medical personnel in the intensive care nursery perform a number of activities to prevent the baby from developing an infection while surgeons close the back and, if necessary, treat hydrocephalus. After back surgery, babies are placed in ways that minimizes pressure over the surgical area. These activities can be very disruptive to breast feeding while the stress can decrease the amount of breast milk that the new mother produces. On the other hand, overwhelming evidence supports breastfeeding in infants. Hospital staff should engage whatever means possible to implement breast feeding for these infants.]

Rowe DE, Jadhav AL. Care of the adolescent with spina bifida. *Pediatr.Clin.North Am.* 2008 Dec;55(6):1359-74, ix.

Abstract: Myelodysplasia with attendant spinal dysraphic disorders can be and often is progressive. In addition to the neurologic deficit present since the prenatal period, hydrocephalus, Chiari malformations with hydromyelia, tethered cord syndrome, or combinations of one or all lead to changing functional ability and progressive deformities. With recent advances in neurologic monitoring and treatment, many of these deformities can be prevented from appearing or becoming destabilizing if the subtle signs are picked up and early intervention is provided. Advances in urologic surgery have also provided better control of continence in addition to reduced infections and renal destruction. This article reviews the monitoring and treatment considerations that have led to such a significant improvement in outcomes in patients who have myelodysplasia.

[This article provides a sound review of the issues facing teens with spina bifida. Health care providers tend to focus on medical issues like bowel, bladder, mobility and skin, while “typical adolescent” needs and behaviors tend to be ignored.]

Teulier C, Smith BA, Kubo M, Chang CL, Moerchen V, Murazko K, Ulrich BD. Stepping responses of infants with myelomeningocele when supported on a motorized treadmill. *Phys. Ther.* 2009 Jan;89(1):60-72.

Abstract: BACKGROUND AND PURPOSE: Infants with myelomeningocele (MMC) have difficulty with, and show delays in, acquiring functional skills, such as walking. This study examined whether infants with MMC will respond to treadmill practice by producing stepping patterns or at least motor activity during the first year after birth. This study also compared the stepping trajectories of infants with MMC across age with those of infants with typical development (TD) to analyze the characteristics of the development of stepping patterns in infants with MMC early in life. **PARTICIPANTS:** Twelve infants with MMC (lumbar and sacral lesions) and 12 infants with TD were the participants in this study. **METHODS:** The infants were tested on a treadmill at ages 1, 3, 6, 9, and 12 months, with no treadmill practice between test sessions. Infants were supported on the treadmill for twelve 20-second trials. A digital camera and behavior coding were used to determine step rate, interlimb stepping patterns, step parameters, and motor activity level. **RESULTS:** Treadmill practice elicited steps in infants with MMC (14.4 steps/minute during the year) but less so than in infants with TD (40.8 steps/minute). Responsiveness was affected by lesion level but varied markedly among infants. Interlimb stepping was less readily alternating, but step parameters were similar to those produced by their peers with TD. Finally, holding infants with MMC on a moving treadmill resulted in greater motor activity (17% during the year) than holding infants on a nonmoving treadmill. **DISCUSSION AND CONCLUSION:** Infants with MMC responded to the treadmill by stepping (but less so than infants with TD) and showing increased motor activity, but they demonstrated a different developmental trajectory. Future studies are needed to explore the impact of enhancing sensory input during treadmill practice to optimize responses in infants with MMC.

[The authors of this study evaluated leg responses of infants with meningocele while they were supported on a treadmill. This type of evaluation may be a useful baseline to assessing motor function—and perhaps may be useful after prenatal interventions like surgery or stem cell transplantation into the spinal cord.]

Tilford JM, Grosse SD, Goodman AC, Li K. Labor Market Productivity Costs for Caregivers of Children with Spina Bifida: A Population-Based Analysis. *Med. Decis. Making* 2008 Aug 25.

Abstract: BACKGROUND: Caregiver productivity costs are an important component of the overall cost of care for individuals with birth defects and developmental disabilities, yet few studies provide estimates for use in economic evaluations. **OBJECTIVE:** This study estimates labor market productivity costs for caregivers of children and adolescents with spina bifida. **METHODS:** Case families were recruited from a state birth defects registry in Arkansas. Primary caregivers of children with spina bifida (N = 98) reported their employment status in the past year and demographic characteristics. Controls were abstracted from the Current Population Survey covering the state of Arkansas for the same time period (N = 416). Estimates from regression analyses of labor market outcomes were used to calculate differences in hours worked per week and lifetime costs.

RESULTS: Caregivers of children with spina bifida worked an annual average of 7.5 to 11.3 hours less per week depending on the disability severity. Differences in work hours by caregivers of children with spina bifida translated into lifetime costs of \$133,755 in 2002 dollars using a 3% discount rate and an age-and sex-adjusted earnings profile. Including caregivers' labor market productivity costs in prevention effectiveness estimates raises the net cost savings per averted case of spina bifida by 48% over the medical care costs alone. CONCLUSIONS: Information on labor market productivity costs for caregivers can be used to better inform economic evaluations of prevention and treatment strategies for spina bifida. Cost-effectiveness calculations that omit caregiver productivity costs substantially overstate the net costs of the intervention and underestimate societal value.

[The health-related care of children with spina bifida—including care in the intensive care nursery, other hospitalizations, surgical treatments, therapy services, equipment, medication and supplies—is very costly. This analysis evaluated the cost of care in terms of labor market productivity and concluded that lifetime costs are \$133,755—in addition to the health-related costs. It does not include the “cost” of lost opportunities, such as the parent who cannot return to school or the family who cannot go on vacation.]

Genetics

Au KS, Tran PX, Tsai CC, O'Byrne MR, Lin JI, Morrison AC, Hampson AW, Cirino P, Fletcher JM, Ostermaier KK, et al. Characteristics of a spina bifida population including North American Caucasian and Hispanic individuals. *Birth Defects Res. A Clin. Mol. Teratol.* 2008 Oct;82(10):692-700.

Abstract: BACKGROUND: Meningomyelocele (MM) is a common human birth defect. MM is a disorder of neural development caused by contributions from genes and environmental factors that result in the NTD and lead to a spectrum of physical and neurocognitive phenotypes. METHODS: A multidisciplinary approach has been taken to develop a comprehensive understanding of MM through collaborative efforts from investigators specializing in genetics, development, brain imaging, and neurocognitive outcome. Patients have been recruited from five different sites: Houston and the Texas-Mexico border area; Toronto, Canada; Los Angeles, California; and Lexington, Kentucky. Genetic risk factors for MM have been assessed by genotyping and association testing using the transmission disequilibrium test. RESULTS: A total of 509 affected child/parent trios and 309 affected child/parent duos have been enrolled to date for genetic association studies. Subsets of the patients have also been enrolled for studies assessing development, brain imaging, and neurocognitive outcomes. The study recruited two major ethnic groups, with 45.9% Hispanics of Mexican descent and 36.2% North American Caucasians of European descent. The remaining patients are African-American, South and Central American, Native American, and Asian. Studies of this group of patients have already discovered distinct corpus callosum morphology and neurocognitive deficits that associate with MM. We have identified maternal MTHFR 667T allele as a risk factor for MM. In addition, we also found that several genes for glucose transport and metabolism are potential risk factors for MM. CONCLUSIONS: The enrolled patient population provides a valuable resource for elucidating the disease characteristics and mechanisms for MM development.

[This paper describes the sample of children with meningomyelocele who have been evaluated

by the well-known collaborative study that includes centers in Texas, Ontario, California, and Kentucky. More studies that involve large collaboratives can enhance research and our current understanding of people with spina bifida.]

Deak KL, Siegel DG, George TM, Gregory S, Shley-Koch A, Speer MC. Further evidence for a maternal genetic effect and a sex-influenced effect contributing to risk for human neural tube defects. *Birth Defects Res. A Clin. Mol. Teratol.* 2008 Oct;82(10):662-9.

Abstract: **BACKGROUND:** Neural tube defects (NTDs), including spina bifida and anencephaly, are the second most common birth defect with an incidence of 1/1000. Genetic factors are believed to contribute to NTD risk and family-based studies can be useful for identifying such risk factors. **METHODS:** We ascertained 1066 NTD families (1467 affected patients), including 307 multiplex NTD families. We performed pedigree analysis to describe the inheritance patterns, pregnancy outcomes, and recurrence risks to relatives of various types. **RESULTS:** Myelomeningocele or spina bifida (66.9%) and cranial defects (17.7%) were the most common NTD subtypes observed. The overall male:female ratio for affected individuals was 0.82, and there were even fewer males among individuals with an upper level NTD (0.62). Among twins, 2 of the 5 monozygotic twins and only 3 of 35 dizygotic twins were concordant, while 27% of the same sex twins were concordant, but none of the different sex twins. The estimated 6.3% recurrence risk to siblings (CI 0.04-0.08) is consistent with previous reports. Families with two or more affected individuals show a higher proportion of female transmitters ($p = 0.0002$). Additionally, the number of affected relatives in maternal compared to paternal lineages was more than double ($p = 0.006$). There were significantly more miscarriages, infant deaths, and stillborn pregnancies of the maternal aunts and uncles ($p < 0.0001$) and of first cousins ($p = 0.04$). **CONCLUSIONS:** Our data provide several lines of evidence consistent with a maternal effect, as well as a sex-influenced effect, in the etiology of NTDs.

[More than 1400 individuals with neural tube defects were evaluated in this study to gain insights into the modes of inheritance for these disorders. They found that NTDs, especially the more severe ones, were more common in females. They also found a high recurrence risk for maternal half siblings, and increased frequency of affected relatives on the maternal side of the family, suggesting maternal and sex-influenced effects as well as environmental ones (including folic acid). The inheritance of NTDs, including spina bifida, seems to be as complex as the condition itself.)

Goetzinger KR, Stamilio DM, Dicke JM, Macones GA, Odibo AO. Evaluating the incidence and likelihood ratios for chromosomal abnormalities in fetuses with common central nervous system malformations. *Am.J.Obstet.Gynecol.* 2008 Sep;199(3):285-6.

Abstract: **OBJECTIVE:** To determine the incidence and likelihood ratios for chromosomal abnormalities in fetuses with common central nervous system malformations on ultrasound. **STUDY DESIGN:** A database derived retrospective cohort study of all patients referred for ultrasound and genetic evaluation from 1990-2006. Fetal karyotypes were delineated by prenatal diagnosis or postnatal examination. The incidence and likelihood ratios were calculated for the association of each central nervous system abnormality with trisomy 13, 18, and 21. **RESULTS:** Of 62,111 women included, 587 (0.9%) had major fetal central nervous system abnormalities. The only central nervous

system anomalies associated with trisomy 21 were ventriculomegaly and choroid plexus cysts. When isolated, only spina bifida, holoprosencephaly, and agenesis of the corpus callosum were significantly associated with trisomy 13, anencephaly with trisomy 18, and ventriculomegaly with trisomy 21. Likelihood ratios positive range from 2-20 depending on the central nervous system malformation and aneuploidy type.

CONCLUSION: Central nervous system malformations detected on ultrasound are strongly associated with and predictive of chromosomal abnormalities, especially trisomy 13 and 18.

[This study reminds us that meningomyelocele can be found as part of several chromosome anomalies, especially trisomy 13.]

Lu W, Zhu H, Wen S, Yang W, Shaw GM, Lammer EJ, Finnell RH. Nicotinamide N-methyl transferase (NNMT) gene polymorphisms and risk for spina bifida. *Birth Defects Res.A Clin.Mol.Teratol.* 2008 Oct;82(10):670-5.

Abstract: BACKGROUND: Moderate hyperhomocysteinemia is a known risk factor for NTDs in a variety of experimental model systems and is believed to be important in humans as well. The enzyme nicotinamide N-methyl transferase (NNMT) was identified in a genome-wide linkage scan as being an important regulator of homocysteine homeostasis in a Spanish population, making it an interesting candidate gene for NTDs. METHODS: We evaluated 11 SNPs (single nucleotide polymorphism) of the NNMT gene in our study population. In this study, 252 cases (infants with spina bifida) and 335 controls (nonmalformed infants), born during the period 1983-1986 in selected counties in California, were genotyped for variants of the NNMT gene. Allelic, genotype, and haplotype associations with spina bifida risk were evaluated and analyzed. RESULTS: None of the SNPs studied alone showed allelic or genotypic associations with spina bifida. However, the TCAG haplotype for block 3 (rs2852447, rs2852425, rs4646337, and rs11569688) showed a decreased risk for spina bifida among non-Hispanic Whites (OR 0.4; 95%CI: 0.1-1.0). CONCLUSIONS: No association was found between infant NNMT gene variants and risk for spina bifida in our study population. However, small sample sizes for most variant groups and for phase-unknown haplotype data limited the power of the study.

[The genetic modes of inheritance for neural tube defects remain mysterious. In this study, a search for defects in specific genes related to homocysteine was inconclusive. The mystery continues.]

Mitchell LE. Spina Bifida Research Resource: study design and participant characteristics. *Birth Defects Res.A Clin.Mol.Teratol.* 2008 Oct;82(10):684-91.

Abstract: BACKGROUND: Myelomeningocele is a common serious malformation. In the majority of affected individuals, it is believed to be nonsyndromic and determined by the effects of multiple genetic and nongenetic factors. METHODS: The Spina Bifida Research Resource (SBRR) is an ongoing, family-based study, designed to identify maternal and embryonic genes related to myelomeningocele. Families that include at least one individual with myelomeningocele are eligible to participate in the SBRR. Recruitment into the SBRR has occurred in two phases. Descriptive analyses were undertaken to characterize the case individuals enrolled in Phase 1. In addition, the characteristics of subgroups of case individuals, defined by lesion level, were compared. RESULTS: During Phase 1, 671 families including 683 case individuals were enrolled.

Families in which the case individual(s) were known or suspected to have a recognized pattern of malformations and families that did not complete the study interview were excluded from the present analyses. The case individuals in the remaining families (n = 534) were predominantly female (53%) and non-Hispanic Caucasian (87%), and in the majority (74%) the highest level of the lesion was in the lumbar region. Differences in the characteristics of the case individuals with lumbar and thoracic level lesions were detected. CONCLUSIONS: This article provides details regarding study design, recruitment, and data collection, and the characteristics of the SBRR Phase 1 study population. The data available from the SBRR, in combination with a rapidly evolving understanding of the variation within the human genome, provide an unprecedented opportunity to explore the genetic contribution to myelomeningocele.

[This paper describes the sample of 683 children with meningomyelocele who have been enrolled in the genetic studies at Texas A&M. Studies of genetic risk factors for spina bifida using family-based study designs continue with the hope of identifying genetic mechanisms for this condition.]

Pangilinan F, Geiler K, Dolle J, Troendle J, Swanson DA, Molloy AM, Sutton M, Conley M, Kirke PN, Scott JM, et al. Construction of a high resolution linkage disequilibrium map to evaluate common genetic variation in TP53 and neural tube defect risk in an Irish population. *Am.J.Med.Genet.A* 2008 Oct 15;146A(20):2617-25.

Abstract: Genetic and environmental factors contribute to the etiology of neural tube defects (NTDs). While periconceptional folic acid supplementation is known to significantly reduce the risk of NTDs, folate metabolic pathway related factors do not account for all NTDs. Evidence from mouse models indicates that the tumor protein p53 (TP53) is involved in implantation and normal neural tube development. To determine whether genetic variation in TP53 might contribute to NTD risk in humans, we constructed a high resolution linkage disequilibrium (LD) map of the TP53 genomic region based on genotyping 21 markers in an Irish population. We found that nine of these variants can be used to capture the majority of common variation in the TP53 genomic region. In contrast, the 3-marker haplotype commonly reported in the TP53 literature offers limited coverage of the variation in the gene. We used the expanded set of polymorphisms to measure the influence of TP53 on NTDs using both case-control and family based tests of association. We also assayed a functional variant in the p53 regulator MDM2 (rs2279744). Alleles of three noncoding TP53 markers were associated with NTD risk. A case effect was seen with the GG genotype of rs1625895 in intron 6 (OR = 1.37 [1.04-1.79], P = 0.02). A maternal effect was seen with the 135/135 genotype of the intron 1 VNTR (OR = 1.86 [1.16-2.96], P = 0.01) and the TT genotype of rs1614984 (RR = 0.58 [0.37-0.91], P = 0.02). As multiple comparisons were made, these cannot be considered definitive positive findings and additional investigation is required.

[The genetic modes of inheritance for neural tube defects remain mysterious. In this study, a search for defects in specific genes related to TP53 (a protein related to tumors that can affect mice with neural tube defects) was inconclusive. The mystery continues.]

Learning

Hannay HJ, Walker A, Dennis M, Kramer L, Blaser S, Fletcher JM. Auditory interhemispheric transfer in relation to patterns of partial agenesis and hypoplasia of the corpus callosum in spina bifida meningomyelocele. *J.Int.Neuropsychol.Soc.* 2008 Sep;14(5):771-81.

Abstract: Spina bifida meningocele with hydrocephalus (SBM) is commonly associated with anomalies of the corpus callosum (CC). We describe MRI patterns of regional CC agenesis and relate CC anomalies to functional laterality based on a dichotic listening test in 90 children with SBM and 27 typically developing controls. Many children with SBM (n = 40) showed regional CC anomalies in the form of agenesis of the rostrum and/or splenium, and a smaller number (n = 20) showed hypoplasia (thinning) of all CC regions (rostrum, genu, body, and splenium). The expected right ear advantage (REA) was exhibited by normal controls and children with SBM having a normal or hypoplastic splenium. It was not shown by children with SBM who were left handed, missing a splenium, or had a higher level spinal cord lesion. Perhaps the right hemisphere of these children is more involved in processing some aspects of linguistic stimuli.

[The corpus callosum, which serves as a bridge between the right and left sides of the brain, is known to be abnormal in many children who have meningocele with hydrocephalus. The authors of this study document the effects of changes in the corpus callosum on a test of listening. This has implications for the learning disabilities often found in these children.]

Hannay HJ, Dennis M, Kramer L, Blaser S, Fletcher JM. Partial agenesis of the corpus callosum in spina bifida meningocele and potential compensatory mechanisms. *J.Clin.Exp.Neuropsychol.* 2009 Feb;31(2):180-94.

Abstract: After a review of Arthur Benton's conceptual and methodological contributions to the understanding of normal and pathological development, we discuss agenesis of the corpus callosum (CC), criteria for potential neuroanatomical compensatory mechanisms in CC agenesis, and the results of an examination of magnetic resonance imaging (MRI) data of the CC in 193 children with spina bifida meningocele (SBM). There were 26 CC regional patterns. Although complete agenesis did not occur, partial agenesis was observed in 102 children and within 15 CC regional patterns. Only 4.1% had a normal CC. Quantitative assessment of the area of the CC in 26 NC children and 68 children with SBM revealed that all subgroups with CC anomalies had smaller areas than did a subgroup with a normal CC. Areas were especially small in rostral/splenial agenesis and splenial agenesis but larger with rostral agenesis. Subgroups with normal/hypoplastic regions or complete hypoplasia also had CC areas that were smaller than normal but larger than the areas for the splenial agenesis groups. The relative rarity of anterior commissure enlargement (3.1%) and longitudinal bundles of Probst (0.1%) suggest that these particular fiber tract anomalies are unlikely candidates for structural compensatory mechanisms. The hippocampal commissure, enlarged in 13%, may be a more promising candidate. Overall, however, the functionality of anomalous fiber tracts and commissures in SBM is yet to be determined.

[The corpus callosum, which serves as a bridge between the right and left sides of the brain, is known to be abnormal in many children who have meningocele with hydrocephalus. In this study, only 4% of the children evaluated had a completely normal corpus callosum. This has implications for the learning disabilities often found in these children.]

Hasan KM, Sankar A, Halphen C, Kramer LA, Ewing-Cobbs L, Dennis M, Fletcher JM. Quantitative diffusion tensor imaging and intellectual outcomes in spina bifida: laboratory investigation. *J.Neurosurg.Pediatrics* 2008 Jul;2(1):75-82.

Abstract: OBJECT: Patients with spina bifida (SB) have variable intellectual outcomes.

The authors used diffusion tensor (DT) imaging to quantify whole-brain volumes of gray matter, white matter, and cerebrospinal fluid (CSF), and perform regional quantitative microstructural assessments of gray matter nuclei and white matter tracts in relation to intellectual outcomes in patients with SB. **METHODS:** Twenty-nine children with myelomeningoceles and 20 age- and sex-matched children with normal neural tube development underwent MR imaging with DT image acquisition and assessments of intelligence. The DT imaging-derived metrics were the fractional anisotropy (FA), axial (parallel), and transverse (perpendicular) diffusivities. These metrics were also used to segment the brain into white matter, gray matter, and CSF. A region-of-interest analysis was conducted of the white and gray matter structures implicated in hydrocephalus. **RESULTS:** The amount of whole-brain gray matter was decreased in patients with SB, with a corresponding increase in CSF ($p < 0.0001$). Regional transverse diffusivity in the caudate nucleus was decreased ($p < 0.0001$), and the corresponding FA was increased ($p < 0.0001$), suggesting reduced dendritic branching and connectivity. Fractional anisotropy in the posterior limb of the internal capsule increased in the myelomeningocele group ($p = 0.02$), suggesting elimination of some divergent fascicles; in contrast, the FA in several white matter structures (such as the corpus callosum genu [$p < 0.001$] and arcuate fasciculus) was reduced, suggesting disruption of myelination. Diffusion tensor imaging-metrics involving gray matter volume and the caudate nucleus, but not other structures, predicted variations in IQ ($r = 0.37-0.50$; $p < 0.05$). **CONCLUSIONS:** Diffusion tensor imaging-derived metrics provide noninvasive neuronal surrogate markers of the pathogenesis of SB and predict variations in general intellectual outcomes in children with this condition.

[Spina bifida (meningomyelocele) is more than a disorder of the spine. Many differences are found in the brains of these children including polymicrogyria and disorders of the corpus callosum. In this study, diffusion tensor imaging was used to compare the brains of children with meningomyelocele to otherwise healthy children. They report that children with MM had smaller brains with disruptions to white matter and to branching of the nerves. More studies using this methodology need to be performed before we can link anatomical differences to actual performance.]

Swartwout MD, Cirino PT, Hampson AW, Fletcher JM, Brandt ME, Dennis M. Sustained attention in children with two etiologies of early hydrocephalus. *Neuropsychology* 2008 Nov;22(6):765-75.

Abstract: Several studies have shown that children with spina bifida meningomyelocele (SBM) and hydrocephalus have attention problems on parent ratings and difficulties in stimulus orienting associated with a posterior brain attention system. Less is known about response control and inhibition associated with an anterior brain attention system. Using the Gordon Vigilance Task (Gordon, 1983), we studied error rate, reaction time, and performance over time for sustained attention, a key anterior attention function, in 101 children with SBM, 17 with aqueductal stenosis (AS; another condition involving congenital hydrocephalus), and 40 typically developing controls (NC). In SBM, we investigated the relation between cognitive attention and parent ratings of inattention and hyperactivity and explored the impact of medical variables. Children with SBM did not differ from AS or NC groups on measures of sustained attention, but they committed more errors and responded more slowly. Approximately one-third of the SBM group had attention symptoms, although parent attention ratings were not associated with task performance. Hydrocephalus does not account for the attention profile of children with SBM, which also reflects the distinctive brain dysmorphologies associated with this

condition.

[Children with meningomyelocele (MM) and hydrocephalus often are diagnosed as having ADHD (attention disorder with hyperactivity), although they often do not respond well to stimulants and other medications used to treat ADHD. This study compared children with MM to those with hydrocephalus from another cause and to children who were otherwise healthy. The children with MM were comparable on tasks of sustained attention but committed more errors and responded more slowly. The hydrocephalus per se was not sufficient to explain these differences. This study confirms the findings of other studies that meningomyelocele leads to diffuse changes in the brain that affect cognition.]

Tarazi RA, Zabel TA, Mahone EM. Age-related differences in executive function among children with spina bifida/hydrocephalus based on parent behavior ratings. *Clin.Neuropsychol.* 2008 Jul;22(4):585-602.

Abstract: Previous research has suggested that adolescents with myelomeningocele and shunted hydrocephalus (MMH) have difficulties with aspects of executive functioning and, in turn, with functional independence. There is little research, however, examining patterns of executive functioning across adolescence in this population. The goal of this cross-sectional study was to examine parent ratings of executive function in children with MMH and in typically developing peers across late childhood and adolescence. Parents of 36 individuals with MMH and 35 typically developing peers, ages 10 to 18 years, completed the Behavior Rating Inventory of Executive Function (BRIEF). The BRIEF is organized into eight scales and two primary indices—Metacognition (MCI) and Behavioral Regulation (BRI). As a whole, the children with MMH had significantly higher BRIEF T-scores, as well as a higher prevalence of clinically significant T-scores across subscales, particularly those representing cognitive control. Effects of group, age, and age-by-group interactions on the mean raw scores of the MCI and BRI were examined using regression analyses. There were significant group effects ($p < .05$) for both the BRI and MCI, with the controls having significantly lower mean ratings than the MMH group. There was also a significant contribution of age-by-group interaction on the BRI ($p < .05$). Although mean raw scores on the BRI for the MMH group remained stable across ages, mean raw scores in the control group decreased as age increased. Thus, healthy children have age-related improvements in executive control behaviors across adolescence, particularly behavioral control, while children with MMH demonstrate no age-related improvements in parent reported executive behaviors across adolescence. Therefore, children with MMH may continue to require targeted interventions and modifications to address executive dysfunction into young adulthood in order to promote functional independence.

[Many children with meningomyelocele and hydrocephalus (MMH) have been shown to have problems with executive function—the ability to initiate an activity, plan, organize, shift thought or attention, inhibit behaviors, and prepare responses. This study showed that children and teens with MMH had more problems on a test of executive function and did not improve with age as did children and teens without MMH.]

Neurosurgery

Danzer E, Adzick NS, Rintoul NE, Zarnow DM, Schwartz ES, Melchionni J, Ernst LM, Flake AW, Sutton LN, Johnson MP. Intradural inclusion cysts following in utero closure of

myelomeningocele: clinical implications and follow-up findings. J.Neurosurg.Pediatrics 2008 Dec;2(6):406-13.

Abstract: OBJECT: The goal in this study was to evaluate the incidence and clinical implications of the development of cutaneously derived intradural inclusion cysts (ICs) following fetal myelomeningocele (fMMC) closure. METHODS: Retrospective databases and responses to a parental questionnaire were reviewed to determine the incidence, clinical presentation, and outcomes of fMMCs in children in whom ICs developed at follow-up. RESULTS: Prior to the National Institutes of Health (NIH)-sponsored Management of Myelomeningocele Study (MOMS), 54 patients underwent fMMC closure at the authors' institution. Sixteen (30%) presented with symptomatic tethered cord syndrome (TCS) at a median age of 27 months (range 4-93 months). Ten (63%) of the 16 (19% of the total) developed TCS in association with an intradural IC. In 9 (90%) of 10 patients, the IC was seen on preoperative MR imaging, and in 1 it was found during surgery. Four additional children (7% of the total) with evidence of an IC on surveillance MR imaging are currently asymptomatic at 94, 84, 60, and 60 months of age, respectively. All but 1 (an L-3 level lesion) IC developed in infants with L-4 and L-5 defects. After cyst removal, 6 children are asymptomatic at a median follow-up of 36 months (range 12-63 months). Following IC removal, 4 children lost normal bladder function and now require clean intermittent catheterization, and 1 lost normal leg function and now requires a walking aid for ambulation. Histologically, 8 lesions were dermoid, 1 was an epidermoid, and 1 was a mixed dermoid-epidermoid IC. Three patients developed another IC and required its removal at 24, 39, and 51 months, respectively. One required another tethered cord release within 57 months after IC removal. CONCLUSIONS: Cutaneously derived intradural ICs can develop following fMMC surgery. Deterioration of bladder function, risk of recurrence, and loss of lower-extremity function appear to be the most important long-term complications of IC in children with fMMCs. The ongoing NIH-sponsored MOMS may help determine whether children with fMMC are at increased risk of IC development compared with children treated with postnatal MMC closure. Parents seeking fMMC closure should be informed about the possibility of IC formation and the potential clinical consequences.

[As more children have *in utero* surgery, more complications of that surgery will be found. This study documents the occurrence of tethered spinal cord in 30% of children who had prenatal surgery, with a significant number having intradural inclusion cysts. It is not clear whether this complication occurs at a different rate in those who have prenatal versus postnatal surgery.]

Kohl T, Tchatcheva K, Merz W, Wartenberg HC, Heep A, Muller A, Franz A, Stressig R, Willinek W, Gembruch U. Percutaneous fetoscopic patch closure of human spina bifida aperta: advances in fetal surgical techniques may obviate the need for early postnatal neurosurgical intervention. Surg.Endosc. 2008 Sep 26.

Abstract: BACKGROUND: A percutaneous minimally invasive fetoscopic approach was attempted for closure of a spina bifida aperta in two fetuses with L5 lesions. The goal was to obviate the need for postnatal neurosurgery to manage this condition. METHODS AND RESULTS: The percutaneous fetoscopic procedures were performed by a two-layer approach at respectively 22 +/- 2 and 22 +/- 4 weeks of gestation. The fetuses were delivered respectively at 32 +/- 6 and 32 + 3 weeks of gestation. Their neural cords were completely covered although in small areas skin closure was incomplete. Postnatally, complete skin closure occurred beneath an occlusive draping within 2 to 3 weeks such that neurosurgical intervention was not required. Both neonates showed reversal of

hindbrain herniation, near-normal leg function, and satisfactory bladder and bowel function. For one of the two fetuses, ventriculoperitoneal shunt insertion was not required. CONCLUSIONS: Percutaneous minimally invasive fetoscopic patch closure of spina bifida aperta offers a substantially less maternal trauma than open fetal surgical repair and currently may even obviate the need for postnatal neurosurgical repair. With a little further improvement in surgical techniques and a better understanding of incorporating surgical patches into the fetus, complete skin closure seems possible in the near future.

[The authors of this German study used a fetal endoscope prenatally to close the backs of two fetuses who had spina bifida. They argue that this procedure, which is less invasive than the prenatal surgery being done in the MOMS study, may be a future consideration when closing the lesions prenatally. Fetoscopy has been done in lambs, with outcomes that have improved over time. As technical advances in surgery like robotics and endoscopes improve, less invasive techniques for doing prenatal surgery likely will be used in humans.]

Geerdink N, Pasma JW, Rotteveel JJ, Roeleveld N, Mullaart RA. Compound muscle action potentials in newborn infants with spina bifida. *Dev. Med. Child Neurol.* 2008 Sep;50(9):706-11.

Abstract: The aim of this study was to investigate the relationship between compound muscle action potentials (CMAPs) and neurological impairment in newborn infants with spina bifida. Thirty-one newborn infants (17 males, 14 females, mean gestational age 39 wks [SD 2]; mean birthweight 3336 g [SD 496]) with spina bifida were investigated at a median age of 2 days (range 1-18 d). Motor and sensory impairment and muscle stretch reflexes were assessed and neuroimaging was performed. CMAPs were recorded from the tibialis anterior muscle and the gastrocnemius muscle after percutaneous electrical nerve stimulation. CMAPs were obtained in almost all infants. The area under the curve of the CMAP (CMAP-area) was associated with motor and sensory impairment and with the presence of muscle stretch reflexes, but not with the morphological level of the spinal anomaly. These associations were stronger for the gastrocnemius muscle than for the tibialis anterior muscle. In conclusion, the CMAP-area correlates with neurological impairment in neonatal spina bifida and provides an estimate of residual motor neuron function in affected spinal segments. The assessment of CMAPs after percutaneous electrical nerve stimulation is recommended as an additional instrument to the clinical neurological examination and imaging studies.

[This study evaluated the electrical activity of leg muscles in newborns with spina bifida, and found that the compound muscle action potentials (CMAPs) were associated with motor and sensory function. They recommend adding this evaluation to our current evaluation of babies. There are several problems with that recommendation, however. First, the neurological function of babies change in the first several weeks of life (sometimes from spinal shock following surgery), and the neurological function of the legs does not stabilize until they are several weeks of age. Second there is no evidence that adding such an evaluation alters care. The cost of medical care continues rising. Any increases in cost should be justified before being implemented.]

Gumus N. A new approach to closure of myelomeningocele defect: z advancement-rotation flap. *Ann. Plast. Surg.* 2008 Dec;61(6):640-5.

Abstract: Because myelomeningocele defects vary in size, shape, and location, no single procedure applies to all. A large number of techniques have been described for closing the back defects occurring after excision of the meningocele sac, but new studies focus more on simple, reliable, and versatile alternatives because large defect is a challenging problem for reconstruction. This study presented a new fasciocutaneous flap, called z advancement-rotation flap for the coverage of meningomyelocele defects with the successful outcomes of 11 patients. Meningocele defect is imagined as a rhombus shape placed vertically over the midline and accepted to have corners and angles of a rhombus, but any skin excision from the margins of the wound is not made for creating a defect, which will be completely similar to a rhombus so that its angles are obtuse and real appearance is elliptic or round. Flaps are elevated from both sides of the wound and can easily be advanced, then rotated to the midline, and the donor area can be primarily sutured with no complications. Defect sizes ranged from 7.5 x 6 to 12 x 9.5 cm, and mean operation time for flap elevation and closure was 29 minutes. Presented technique is not only simple, safe, and stable but also has got some more advantages such as short operation time because of easy dissection, minimal blood loss, and primary closure of the whole wound as well. It seems to be an alternative for safe, rapid, less bleeding, and easy surgery resulting in a solution for the closure of large meningomyelocele defects.

Turhan HN, Eser O, Demir Y, Aslan A, Koken R, Melek H. Repair of wide myelomeningocele defects with the bilateral fasciocutaneous flap method. *Turk.Neurosurg.* 2008 Jul;18(3):311-5.

Abstract: OBJECTIVE: Five large myelomeningocele defects repaired by bilateral fasciocutaneous flaps are presented in this article. PATIENTS and METHODS: After the neurological repair, bilateral fasciocutaneous flaps were raised, transposed over the midline, and sutured to each other. RESULTS: All operations lasted shorter than one hour and no major complication was noted in the follow-up. Three of the defects were in the thoracolumbar and two in the lumbosacral regions. The areas of the defects were between 5x6 cm (30 cm²) and 8x10 cm (80 cm²). CONCLUSION: The bilateral fasciocutaneous flap technique offers a shorter operation duration and lower bleeding rate; it is simple to learn and practice and does not abolish alternative operation techniques.

[These studies describe outcomes after closure of large defects in meningomyelocele. Many techniques are used because the “optimal” one has not yet been found.]

Jans L, Vlummens P, Van DS, Verstraete K, Abernethy L. Hemimyelomeningocele: a rare and complex spinal dysraphism. *JBR.-BTR.* 2008 Sep;91(5):198-9.

Abstract: We report a case of hemimyelomeningocele. This is a rare spinal dysraphism, consisting of the association of a split cord malformation with myelomeningocele. MRI demonstrates division of the spinal cord into two hemicords which are separated by a bony spur. The left hemicord forms a myelomeningocele, thus resulting in a hemimyelomeningocele. Accurate and early diagnosis of spinal dysraphisms is essential for optimal, individualised neurosurgical treatment.

Ozturk E, Sonmez G, Mutlu H, Sildiroglu HO, Velioglu M, Basekim CC, Kizilkaya E. Split-cord malformation and accompanying anomalies. *J.Neuroradiol.* 2008 Jul;35(3):150-6.

Abstract: OBJECTIVE: To present the magnetic resonance imaging (MRI) appearances of spinal split-cord malformation (SCM) and to investigate the various types of congenital spinal disorders associated with SCM. MATERIALS AND METHODS: MR examinations of 23 patients with SCM were carried out in our hospital between June 2002 and May 2007 and retrospectively analysed. RESULTS: Nineteen (82.6%) patients were diagnosed as type I SCM, while four (17.4%) were diagnosed as type II SCM. The most commonly involved site of SCM was the dorsolumbar area (47.8%) while cervical involvement was the least common (4.3%). No accompanying congenital spinal disorders were detected in four patients (17.4%). In 19 patients (82.6%), congenital spinal disorders accompanying SCM were detected, the most common of which was a low-lying cord, found in 14 patients (60.9%). Other anomalies included hydromyelia in seven patients (30.4%), lipoma in six (26%), meningomyelocele in four (17.4%), thick filum in three (13%) and dermoid cyst in three (13%). CONCLUSION: In preoperative planning for SCM, its characteristics and those of the accompanying anomalies should be determined. MRI is a valuable tool for making such determinations.

[These two studies describe split cord malformations in people with neural tube defects. In these conditions the spinal cord is divided for a variable length. A bony spur may be present in the middle of the division, or two separate spinal cords may occur. Associated problems like fluid in the central canal of the cord (syrinx or hydromyelia), and progressive neurological damage may occur with these malformations.]

Morota N, Ihara S. Postnatal ascent of the cerebellar tonsils in Chiari malformation Type II following surgical repair of myelomeningocele. *J.Neurosurg.Pediatrics* 2008 Sep;2(3):188-93.

Abstract: OBJECT: Postnatal improvement in Chiari malformation type II (CM-II) following surgical repair of myelomeningocele was evaluated. METHODS: The authors reviewed data obtained in 20 cases in which patients underwent postnatal myelomeningocele repair within the first 48 hours after birth between October 2002 and September 2006. In 14 patients (Group 1), myelomeningocele was diagnosed in utero and the infants were delivered by cesarean section at 35-39 weeks' gestation (mean 36.4). The 6 infants in Group 2 were born after full-term gestation (39-41 weeks), and their myelomeningoceles were diagnosed postnatally. In all 20 patients, the myelomeningoceles were surgically repaired postnatally. Dynamic change of the herniated cerebellar tonsils in CM-II before and after the myelomeningocele repair, associated hydrocephalus, and symptomatic CM-II were analyzed. RESULTS: In Group 1, the CM-II was confirmed before myelomeningocele repair in 13 cases (93%). The spinal level of the caudal end of the cerebellar tonsils ranged from C-2 to C-7. Ascent of the cerebellar tonsils was observed in 11 patients (range 1-4 spinal levels, mean 2 levels) and continued even after ventriculoperitoneal (VP) shunt placement in most patients. A VP shunt was required for the treatment of hydrocephalus in 12 patients (86%). Symptomatic CM-II developed in 8 of 13 patients (61%), 3 of whom required surgical decompression. In Group 2, CM-II was confirmed in 5 infants (83%), with the cerebellar tonsils at a spinal level of C-2 to C-4 or C-5. Ascent of the cerebellar tonsils was observed in 4 patients (range 1-1.5 spinal levels, average 1.1 levels), and no patient had symptomatic CM-II. A VP shunt was placed in 5 patients (83%). No patient was lost to

follow-up during the 18-month follow-up period. The only statistically significant difference between the 2 groups was the presence of symptomatic CM-II in Group 2 ($p = 0.02$). CONCLUSIONS: Patients showed ascent of the cerebellar tonsils after postnatal myelomeningocele repair. Placement of a VP shunt helped promote the ascent. However, postnatal myelomeningocele repair in the patients in Group 1 failed to consistently prevent development of symptomatic CM-II. This limited experience suggests that postnatal repair of myelomeningocele can partially reverse the anatomical CM-II, but symptomatic CM-II cannot be prevented in some patients when the repair is performed after 36 weeks' gestation.

[In a Chiari type II malformation the lower cerebellum and brainstem appear as if someone had pulled down on the spinal cord before the child was born. This paper documents that following back closure and ventricular shunt insertion, the lower cerebellum actually moves up (towards the top of the head). However, despite this movement, signs and symptoms of the Chiari malformation still occurred. This is consistent with the observation that the severity of appearance of this malformation does not correlate well with the development or severity of symptoms.]

Ramos E, Marlin AE, Gaskill SJ. Congenital dermoid tumor in a child at initial myelomeningocele closure: an etiological discussion. *J.Neurosurg.Pediatrics* 2008 Dec;2(6):414-5.

Abstract: The authors report on a case of a full-term infant with lumbar myelomeningocele who was found to have an intramedullary mass at the time of surgical repair of the defect. The intramedullary mass was consistent with a dermoid tumor both macroscopically and microscopically. This case provides evidence that dermoid tumors occurring at the site of previous surgical myelomeningocele repair are not always a consequence of incomplete excision of the dermal elements.

[Benign dermoid tumors may appear after the lesion in meningomyelocele is closed. This paper argues that sometimes this may be due to incomplete excision; however, on other occasions it may be due to a tumor that already was developing in the area.]

Sinha AK, Lall R, Benson R, O'Brien DF, Buxton N. Intraparenchymal pericatheter cyst following ventriculoperitoneal shunt insertion: does it always merit shunt revision? *Zentralbl.Neurochir.* 2008 Aug;69(3):152-4.

Abstract: Intraparenchymal pericatheter cyst is a rare complication of shunt surgery. It occurs as a consequence of increased CSF outflow resistance distal to a patent ventricular catheter. Treatment has traditionally consisted of shunt revision. We report on two such cases managed conservatively. The role of shunt revisions in cases with asymptomatic intraparenchymal pericatheter cyst is debatable.

[Many bad things can happen to ventriculo-peritoneal shunts like infection or migration of the catheter into the bowel. Sometimes a cyst will form at the end of the catheter. In this study 2 children who had this complication were followed without surgery and did well.]

Sival DA, Verbeek RJ, Brouwer OF, Sollie KM, Bos AF, den Dunnen WF. Spinal hemorrhages are associated with early neonatal motor function loss in human spina bifida aperta. *Early Hum.Dev.* 2008 Jul;84(7):423-31.

Abstract: BACKGROUND: In spina bifida aperta (SBA), leg movements caudal to the

meningomyelocele are present in utero, but they disappear shortly after birth. It is unclear whether leg movements disappear by impact of the neuro-developmental malformation or by superimposed traumatic damage. If superimposed traumatic damage is involved, targeted fetal intervention could improve motor outcome. AIM: To characterize neuromuscular pathology in association with perinatal motor function loss in SBA. PATIENTS/METHODS: In fetal SBA (n=8; 16-40 weeks GA), the median time interval between ultrasound registrations of fetal motor behavior and post-mortem histology was 1 week. Histology was assessed cranial, at and caudal to the meningomyelocele and compared with findings in fetal controls (n=4). RESULTS: Despite fetal movements caudal to the meningomyelocele (5/6), histology indicated muscle fiber alterations (6/6) that concurred with neuro-developmental and traumatic spinal defects [Neuro-developmental defects: spinal ependymal denudation (3/8), reduced amount of (caspase3-negative) lower motor neurons (LMNs; 8/8), aberrant spinal vascularization (8/8). Traumatic defects: gliosis (7/8), acute/fresh spinal hemorrhages near LMNs (8/8)]. CONCLUSION: In all delivered SBA patients, recent spinal hemorrhages were superimposed upon pre-existing defects. If early therapeutic strategies can prevent these superimposed secondary spinal hemorrhages, motor outcome may improve.

[This study documented “recent” spinal hemorrhages in babies born with meningomyelocele. These hemorrhages may be indications of ongoing damage to the spinal cord and nerves. The authors suggest that preventing them prenatally may improve ultimate neurological function in these children. Whether or not this theory is true remains to be seen.]

Tseng JH, Kuo MF, Kwang TY, Tseng MY. Outcome of untethering for symptomatic spina bifida occulta with lumbosacral spinal cord tethering in 31 patients: analysis of preoperative prognostic factors. *Spine J.* 2008 Jul;8(4):630-8.

Abstract: BACKGROUND CONTEXT: The most important goal for treating symptomatic lumbosacral spinal cord tethering is early untethering. PURPOSE: To investigate preoperative symptoms that may have affected the outcome. STUDY DESIGN: Patients with or without improvement and with or without favorable outcome after untethering were compared retrospectively by chart and image review. PATIENT SAMPLE: Thirty-one patients (age between 2 days to 25 years) with spina bifida occulta and symptomatic cord tethering were analyzed. Presenting symptoms (neurological deficits, urological dysfunction, and lower limb deformities) were assessed before and after untethering. OUTCOME MEASURES: Favorable outcome was defined as complete relief of symptoms or mild symptoms whereby patients are able to look after their own personal care without assistance. Unfavorable outcome was defined as moderate or severe disability whereby patients are unable to attend to their own bodily needs without assistance, are bedridden, or require constant nursing attention. METHODS: Differences in patient characteristics and presenting symptoms were compared between those with and without clinical improvement and favorable outcome. Multivariate logistic regression was used to identify prognostic factors affecting the outcome. RESULTS: The average age at surgery was 7.2 years, with a male-to-female ratio of 1.2. The average follow-up time was 4 years. At least one of the following symptoms was present in all patients: neurological deficits (83.9%), urological dysfunction (77.4%), or limb deformities (38.7%). After untethering, all patients had either symptoms stabilized (14 patients, 45.2%) or improved (17 patients, 54.8%), and 14 patients (45.2%) achieved total resolving of symptoms. Logistic regression confirmed that younger age (< or =2 years, odds ratio [OR] 22.0, p=.026), lipomas of filum terminale (OR 25.6, p=.042), and a poor anal tone (OR 10.4, p=.061) were positive prognostic factors for the improvement in

symptoms. The functional outcome was determined by the age at surgery (OR 0.9 per year since 1 year old, $p=.04$) and the presence of limb deformities (OR 0.06, $p=.017$).
CONCLUSIONS: In conclusion, our study suggests that untethering should be performed immediately once the patient shows evidence of symptomatic lumbosacral cord tethering, irrespective of age. Untethering can interrupt progression of symptoms, but sphincter dysfunction and muscle weakness are more likely to improve or resolve. Benefits can be seen in all patients, but young children (before 2 years old) have a higher chance to gain favorable outcome. Retethering is a main concern during follow-up, particularly for the more complicated lipomyelomeningoceles. Investigations using electrophysiologic and urodynamic studies are helpful for early detection of subtle symptomatic cord tethering or retethering.

[Tethering of the spinal cord may lead to neurological signs and symptoms that progress over time. This article argues that once a diagnosis of tethered spinal cord is made, early surgery can improve the long-term outlook: signs and symptoms stop getting worse and some actually improve. Younger children seem to do better following surgery than do older children.]

Warf BC, Campbell JW. Combined endoscopic third ventriculostomy and choroid plexus cauterization as primary treatment of hydrocephalus for infants with myelomeningocele: long-term results of a prospective intent-to-treat study in 115 East African infants. *J.Neurosurg.Pediatrics* 2008 Nov;2(5):310-6.

Abstract: **OBJECT:** Shunt dependence is more dangerous for children in less developed countries. Combining endoscopic third ventriculostomy (ETV) with choroid plexus cauterization (CPC) was previously shown to treat hydrocephalus more effectively than ETV alone in infants < 1 year of age. The goal of this prospective study was to evaluate the effectiveness of ETV-CPC as primary treatment of hydrocephalus in infants with myelomeningocele. **METHODS:** One hundred fifteen consecutive East African infants with myelomeningocele requiring treatment for hydrocephalus were intended for primary management using ETV-CPC. Patient information was prospectively entered into a database. Outcomes were evaluated by life table analysis. Potential predictors for treatment failure were evaluated using multivariate logistic regression. **RESULTS:** Ninety-three patients had a completed ETV-CPC with > 1 month of follow-up. The ETV-CPC procedure was successful in 71 patients (76%), with a mean and median follow-up of 19.0 months. Treatment failures occurred before 6 months in 86% of the patients, and none occurred after 10 months. The operative mortality rate was 1.1%, and there were no infections. Life table analysis suggested that 72% of the patients would be successfully treated using a single ETV-CPC and 78% would remain shunt-independent with reopening of a closed ETV stoma. Multivariate logistic regression showed scarring of the cistern ($p = 0.021$) or choroid plexus ($p = 0.026$) as predictors of failure, but age at the time of surgery was not a significant predictor. **CONCLUSIONS:** Using ETV-CPC appears to successfully provide a more durable primary treatment of hydrocephalus for infants with spina bifida than does shunt placement. These results support ETV-CPC as the better treatment option for these children in developing countries.

[Ventricular shunts often fail, sometimes requiring emergency revision. In poor countries, routine monitoring and emergency services may be unavailable making typical shunt treatment impractical. In this study 93 children in Uganda had endoscopic third ventriculostomy with choroid plexus cauterization. (In 9 patients the procedure had to be abandoned.) The authors calculated that shunt dependency would be avoided in 78% of all children who had the procedure.]

Occult Spinal Dysraphism

Chang ST, Ku CH, Hsieh MF, Chen LC, Chu HY, Chang CC, Tsai KC. Contribution of the multifidus muscle for control of upright posture in subjects with spina bifida occulta. *J.Sport Rehabil.* 2008 Aug;17(3):283-99.

Abstract: **CONTEXT:** The multifidus muscle plays a role in controlling lumbosacral position and postural sway. One of its attachment sites is the exact site of spina bifida occulta (SBO). **OBJECTIVE:** To investigate the role of the muscle for postural control in SBO. **DESIGN:** Cross-sectional cohort. **PARTICIPANTS:** Eighty subjects with SBO (38 in minor; 42 in major) and controls. **INTERVENTIONS:** N/A. **MAIN OUTCOME MEASURES:** Subjects stood upright on a platform at 0 degrees and on an inclined surface (10 degrees and 20 degrees) with feet in plantarflexion/dorsiflexion, together with eyes open and closed. The platform system measured posture to obtain sway area and sway mean for statistics. **RESULTS:** Upon sway area/mean, the group differences of major/minor SBO vs. control were all significant. Sway means of minor SBO were lower than those of major SBO at corresponding inclined degrees. **CONCLUSIONS:** Subjects with SBO demonstrated increased sway as compared to controls.

[In adults with spina bifida occulta, impaired control of postural function in both the resting upright and stressful postures was found. The greater the defect, the worse the control. The clinical implications of this finding remain to be seen.]

Sasani M, Asghari B, Asghari Y, Afsharian R, Ozer AF. Correlation of cutaneous lesions with clinical radiological and urodynamic findings in the prognosis of underlying spinal dysraphism disorders. *Pediatr.Neurosurg.* 2008;44(5):360-70.

Abstract: Spinal dysraphism is characterized by a lack of fusion of the vertebral arches that occurs in the absence of spinous processes with variable amounts of lamina. Here, we retrospectively present the importance of cutaneous lesions and their correlation with clinical presentation, radiological examination and urodynamic assessment. We retrospectively reviewed 612 (6.12%) cases with skin lesions from 10,000 consecutive live-born children seen at two institutions between January 1998 and March 2005. We divided all children into a control group and three groups based on clinical assessment, radiological examination and urodynamic evaluation results. Neurological deficits were identified in 113 (18.46%) children, while spinal dysraphism disorders, tethered cord syndrome and associated orthopedic malformations were found in 171 (27.94%), 119 (19.45%) and 28 (4.57%) patients, respectively. The incidence of tethered cord with lumbosacral dimple lesions over the sacrum (32/119, 26.65% of patients) was 3.5-fold higher than that of lesions found over the coccyx (9/119, 7.5% of patients). Uro-neurological symptoms were found in 207/612 (33.82%) children. Urodynamic assessment revealed decreased bladder capacity in 10% of patients, detrusor hyperflexia during filling in 47% and a low-compliance detrusor in 71%. Discordance between ultrasonography and MRI was found in 16.58% of patients. Spinal cord untethering was performed for 109 patients. Nearly all children with resolution were at the end of their follow-up period (24 months). Retethering occurred in 21 (19.26%) patients, and a second untethering surgery was performed in 12 patients. When spina bifida was associated with lumbar skin lesions, there may have been an increased incidence of

tethered cord and other spinal cord disorders. MRI scans are more reliable and give an exact diagnosis of tethered cord. Neurological and urological instability are ultimately a clinical diagnosis, and there is controversy about their indications for surgery. However, the correlation between urodynamic assessment and cutaneous lesions with a tethered cord found by MRI examination allow for an early diagnosis and the possibility of prompt treatment.

[The findings from this study argue that when a baby is found to have skin lesions—like increased or decrease pigment, a dimple, or a patch of hair—over the spine imaging studies of the spine (like an MRI scan) and neurological (including urological) evaluations should be undertaken to determine if surgical intervention could be helpful in decreasing signs and symptoms or preventing their development.]

Spacca B, Buxton N. Spina bifida occulta and monozygotic twins. *J.Neurosurg.Pediatrics* 2008 Oct;2(4):258-60.

Abstract: Central nervous system maldevelopment can have different presentations in twins. We report on a case of different presentations of spina bifida occulta in monozygotic twins. The first twin presented at birth with a lipomyelomeningocele; a tethered cord was diagnosed in the second twin at 2 years of age. Neural tube defects (NTDs) are a group of common congenital malformations of the brain and spine generated during neurulation. The genetic basis of this process is still not well known. Whenever an NTD is diagnosed in one of a pair of twins, the other twin should also be evaluated for NTDs.

[Since neural tube defects have a genetic component, if one twin has an NTD, the second one should be evaluated for an NTD, including a hidden one like an occult spinal dysraphism. Shared NTDs are more likely in identical twins and in same-sex fraternal twins.]

Yuan Z, Cheng W, Hou A, Wang W, Zhang S, Liu D, Gao F, Li H, Wang W. Constipation is associated with spina bifida occulta in children. *Clin.Gastroenterol.Hepatol.* 2008 Dec;6(12):1348-53.

Abstract: **BACKGROUND & AIMS:** Spina bifida occulta (SBO) is a common developmental variant. The aim of this study was to re-examine the possible association between SBO and constipation in children. **METHODS:** A total of 113 children with constipation underwent plain abdominal radiography, anorectal manometry, neurophysiologic study, electromyography testing, and colonic transit study. Eighty-six were diagnosed with functional constipation (FC) and 27 were diagnosed with nonretentive fecal incontinence (NRFI). The incidence of SBO in these children was compared with 226 sex- and age-matched controls. Twenty-four SBO children with either FC or NRFI also underwent individualized biofeedback training and electric stimulation therapy based on the investigation results. **RESULTS:** The incidence of SBO in the FC and NRFI groups was 47.7% and 77.8%, respectively. Statistically, this is significantly higher than that of the control group (chi-square, 23.9%; $P < .05$). Compared with the FC or NRFI children without SBO, the FC and NRFI children with SBO had decreased vector volumes and electromyography amplitudes, increased rectal sensory thresholds, and prolonged latency of pudendo-anal reflex. All 24 children who underwent individualized biofeedback training and electrical stimulation treatment had sustained symptomatic improvement with less straining, fewer incomplete bowel movements, and less abdominal pain. The recovery rate was 79.2% (19 of 24). **CONCLUSIONS:**

Constipation in children is associated with increased incidence of SBO. Individualized biofeedback combined with electrical stimulation improves both the symptoms and the objective anorectal function measurements.

[Constipation is a well known condition in children with meningomyelocele. This study suggests that it also commonly occurs in children who have occult spinal dysraphism.]

Zambito A, Dall'oca C, Polo A, Bianchini D, Aldegheri R. Spina bifida occulta. Foot deformities, enuresis and vertebral cleft: clinical picture and neurophysiological assessment. *Eur.J.Phys.Rehabil.Med.* 2008 Dec;44(4):437-40.

Abstract: AIM: The aim of the study was to investigate the relationship between the clinical evidence of foot deformities in spina bifida occulta and the associated neurophysiological damage. METHODS: The authors studied 47 patients with foot deformities (37 flat foot, 10 pes cavus) and vertebral cleft, variably associated with enuresis, midline cutaneous lesions, and further orthopaedic deformities. An electrophysiological evaluation was performed in an attempt to investigate the peripheral nervous system in greater detail, including conventional motor and sensory nerve conduction, F-wave recording and electromyogram (EMG) testing. RESULTS: The peroneal nerve F wave latency was longer in patients with pes cavus than in those with flat foot ($P<0.04$). Conversely, the posterior tibial nerve F-wave latency was longer in patients with flat foot than in those with pes cavus ($P<0.02$). Needle EMG showed large amplitude motor unit potentials during voluntary recruitment in all patients, suggesting a neurogenic origin of these EMG changes. Neurophysiological study makes it possible to distinguish between myogenic and lower motor neuron involvement. The existence of some degree of spinal cord dysraphism may be pathophysiologically associated with foot deformities. CONCLUSION: Children with foot deformities and clinical evidence of occult spinal dysraphism should have a neuro-physiological assessment in order to obtain an early diagnosis and avoid ineffective foot surgery.

[Occult spinal dysraphism may lead to progressive signs and symptoms, including deformity of the foot. In an ideal world spinal surgery to prevent complications should be done before orthopaedic deformities appear.]

Orthopaedics

Apkon SD, Fenton L, Coll JR. Bone mineral density in children with myelomeningocele. *Dev.Med.Child Neurol.* 2009 Jan;51(1):63-7.

Abstract: The aim of the present study was to document bone mineral density (BMD) in children with myelomeningocele and to identify variables that contribute to reduced BMD. The study included 24 children with myelomeningocele (nine males, 15 females; age range 4-18y), who had varied levels of neurological impairment (thoracic/high-lumbar, n=6; mid-lumbar, n=9; sacral, n=9) and ambulatory status (non-ambulators, n=12; part-time ambulators n=2; full-time ambulators, n=10). BMD measurements of the femoral neck and whole body using dual energy X-ray absorptiometry assessments of dietary calcium intake, and serum markers of bone metabolism were obtained. BMD is presented as standardized scores (z-scores) which are age- and sex-matched to normally developing children. The mean femoral-neck z-score was -2.41. Femoral-neck z-scores differed significantly according to ambulatory status, with lower z-scores in children who were wheelchair-dependent ($p=0.03$). The mean z-score at the femoral neck demonstrated

a trend toward lower z-scores in children with higher levels of lesions. Almost all children met their recommended daily intake of calcium. Markers of bone metabolism were normal in all patients. This study demonstrates that reduced BMD is a major complication in children with myelomeningocele. There is a significant relationship with low BMD in children who are wheelchair-dependent, a trend in those with higher neurological levels, and no relationship between fractures and reduced BMD.

Ausili E, Focarelli B, Tabacco F, Fortunelli G, Caradonna P, Massimi L, Sigismondi M, Salvaggio E, Rendeli C. Bone mineral density and body composition in a myelomeningocele children population: effects of walking ability and sport activity. *Eur.Rev.Med.Pharmacol.Sci.* 2008 Nov;12(6):349-54.

Abstract: Myelomeningocele causes serious locomotor disability, osteoporosis and pathologic fractures. The aim of this study was to investigate the relationship between body composition, bone mineral density, walking ability and sport activity in myelomeningocele children. 60 patients aged between 5 and 14 yrs with myelomeningocele (22 ambulatory and 38 non-ambulatory), were studied. Fat mass and fat-free-mass were calculated by anthropometry. The bone mineral density at lumbar and femoral neck were evaluated. Bone mineral density at the lumbar and femoral neck was lower than in the normal population. In the non-ambulatory group, bone mineral density was approximately 1 SD lower than in the ambulatory one ($p < 0.01$). Fat mass was greater than expected but without significant differences between walking group (mean 26%) and wheel-chair users (25%). Patients practised sport activity had a better bone mineral density and body fat compared with other patients with the same disability. Patients with myelomeningocele have decreased bone mineral density and are at higher risk of pathologic bone fractures. All subjects showed an excess of fat as percentage of body weight and are shorter than normal children. The measurement of bone mineral density may help to identify those patients at greatest risk of suffering of multiple fractures. Walk ability and sport activity, associated with the development of muscle mass, are important factors in promoting bone and body growth, to reduce the risk of obesity and of pathological fractures.

[These two studies document the increased prevalence of osteoporosis in children and teens who have meningomyelocele. Using a wheelchair increases the risk while participating in sports decreases the risk. Interestingly a one-to-one correspondence between the degree of osteoporosis and the occurrence of fractures does not occur. Other factors (like immobility following surgery) seem to contribute to fractures in addition to the osteoporosis.]

Park KB, Park HW, Joo SY, Kim HW. Surgical treatment of calcaneal deformity in a select group of patients with myelomeningocele. *J.Bone Joint Surg.Am.* 2008 Oct;90(10):2149-59.

Abstract: BACKGROUND: The surgical treatment of calcaneal deformity in patients with myelomeningocele has not been uniformly successful in correcting the deformity and preventing recurrence. The purpose of the present study was to examine the results of posterior transfer of the anterior tibial tendon with concurrent procedures in an attempt to balance the muscular forces on the foot and ankle and to obtain a plantigrade foot. We investigated whether surgery improved pressure distribution over the plantar surface of the foot and whether concurrent abnormal movements observed at the knee, hip, and pelvis influenced the surgical outcome. **METHODS:** Thirty-one feet in eighteen patients

who were able to walk were included in the study. The mean age at the time of surgery was seven years and four months, and the mean duration of follow-up was forty-seven months. Eight patients were classified as having an L5-level myelomeningocele, and ten patients were classified as having a sacral level myelomeningocele. A tibialis anterior tendon transfer was performed in all patients, and accompanying osseous deformities were also corrected in twelve feet. Measurements on plain radiographs, the results of gait analyses, and dynamic foot pressures that were determined before surgery and at the time of the final follow-up were compared. **RESULTS:** No recurrence or worsening of the deformity was observed in any of the patients, and no other types of foot deformity developed after surgery. Postoperative kinematic studies showed a significant ($p < 0.0001$) increase in peak plantar flexion and a significant decrease in peak dorsiflexion force of the ankle in the stance phase of gait. Peak pressures under the forefoot and midfoot were increased after surgery, and the relative amount of weight-bearing on the heel as compared with the forefoot was shifted toward more equal weight-bearing. However, less improvement in foot-pressure distribution was observed in patients with increased pelvic rotation before surgery. Those patients also had decreased knee extension in stance phase and increased hip abduction and pelvic obliquity both before and after surgery in comparison with patients who had normal pelvic rotation. **CONCLUSIONS:** Appropriately combined corrective surgical procedures for the treatment of calcaneal deformity in patients with myelomeningocele can effectively reduce the pressure placed on the calcaneus, increase pressures in the forefoot and midfoot, and prevent recurrence of the calcaneal deformity. However, in the presence of excessive pelvic movement in the coronal and transverse planes and decreased knee extension in stance phase, adequate improvement in pressure distribution over the plantar surface of the foot is not likely to occur after this type of foot surgery. **LEVEL OF EVIDENCE:** Therapeutic Level IV. See Instructions to Authors for a complete description of levels of evidence.

[This study describes a surgical technique for repairing heel deformities in children with meningomyelocele. However, the heel or foot cannot be viewed in isolation. Abnormalities at the knee and/or hip can affect the foot and can have a major effect on surgical outcomes.]

Roehrig S, Like G. Factors affecting shoulder pain in adolescents and young adults with spina bifida. *Pediatr.Phys.Ther.* 2008;20(3):224-32.

Abstract: **PURPOSE:** This study was designed to determine whether factors affecting shoulder pain in adults with spinal cord injury also affected adolescent and young adult manual wheelchair users with spina bifida. **METHODS:** Forty-one participants with spina bifida rated their pain using the Wheelchair User's Shoulder Pain Index. **RESULTS:** Results showed no significant relationship between shoulder pain and age or duration of wheelchair use but significant differences in pain between age groups. Participation in a sports program and level of lesion were not factors in pain intensity. Propelling up an incline was the activity for which the highest intensity of pain was reported. **CONCLUSION:** Shoulder pain in adolescents and young adults with spina bifida is not as great as previously reported in adults with spinal cord injuries but older subjects had greater pain than younger subjects.

[Shoulder pain in people with spina bifida who use wheelchairs is not uncommon and is felt to be due to "overuse." This study documented the frequency of pain and noted that the pain was unrelated to level of lesion; it was more common in older individuals, however.]

Wenz W, Bruckner T, Akbar M. Complete tendon transfer and inverse Lambrinudi arthrodesis:

preliminary results of a new technique for the treatment of paralytic pes calcaneus. *Foot Ankle Int.* 2008 Jul;29(7):683-9.

Abstract: **BACKGROUND:** Paralytic pes calcaneus is commonly associated with myelomeningocele and continues to be one of the most difficult deformities to treat. The purpose of the present study is to describe and report the preliminary results of a new procedure to dynamically correct paralytic pes calcaneus. **MATERIALS AND METHODS:** Since 2002, the senior surgeon operated on six patients with myelomeningocele and developed a new procedure to dynamically correct paralytic pes calcaneus. This new procedure combines a complete tendon transfer (tibialis anterior and posterior, peroneus brevis and longus, and extensor digitorum and hallucis longus) to the Achilles tendon and an additive triple arthrodesis. The extent of the calcaneus deformity was evaluated by measuring the talocalcaneal angle. The mean followup was 32 months. **RESULTS:** Excellent results were achieved in five of the six patients with a mean age of 17.5 years. Using the American Orthopaedic Foot and Ankle Society (AOFAS) ankle and hindfoot scale, pain and function improved from an average of 41.6 preoperatively to 72.0 postoperatively. The deformity as assessed by the talocalcaneal angle, the calcaneal pitch, and the talometatarsal I angle improved after surgery significantly ($p < 0.004$). After 12 months, an additional surgery was required in two patients due to disabling drop-foot. **CONCLUSION:** This new surgical technique to correct paralytic pes calcaneus including complete tendon transfer and triple arthrodesis is promising. Although this is a technically demanding procedure, it provides an excellent functional result without the residual complications associated with ankle arthrodesis.

[This paper describes the outcome of surgical correction for pes calcaneus (in which the heel is displaced downward) in 6 teens with meningomyelocele. The study is small and the surgery extensive. More studies should be done on this technique before it becomes widely accepted.]

Wolf SI, Alimusaj M, Rettig O, Doderlein L. Dynamic assist by carbon fiber spring AFOs for patients with myelomeningocele. *Gait.Posture.* 2008 Jul;28(1):175-7.

Abstract: Patients with calf muscle insufficiency and a calcaneus gait are often dependent on ankle-foot orthoses (AFO). The orthosis is intended to improve walking and posture and should prevent structural deformities. AFOs are often manufactured with a dorsiflexion stop. The design of this type of orthosis has been investigated in several previous studies. In the current study, orthoses with a dorsal carbon fiber spring were compared with the classic design. Five patients with Spina Bifida took part in the current study. All participants underwent a 3D gait analysis including kinematic (VICON infrared cameras) and kinetic (Kistler force plates) data collection. The measurements showed that the carbon spring was able to support the patient during the complete stance phase. It was found that the use of a carbon fiber spring significantly increases the energy return during the 3rd rocker, simulating the natural push-off action ($p < 0.05$). Via a simple mechanical test, the contribution of the carbon spring to the overall kinetics could be estimated proving that the spring does assist the patient for push-off. The more physiological ankle and knee kinematics implies a functional improvement from the carbon springs compared to classic orthosis. This investigation showed, further, that in the fitting process a neutral alignment with the shoe wear has to be carefully checked since the spring kinematics and kinetics during stance phase were influenced significantly by the alignment. Further studies are needed to assess the clinical outcome and to prove the functional benefit of this kind of orthosis.

[Although the AFO (ankle foot orthosis) seems to be a simple device, it comes in a mind-

boggling array of varieties. This study describes an AFO with a carbon fiber spring that was tested on 5 individuals with meningocele. The study is small and the braces required a lot of adjustment. More studies should be done on this bracing technique before it becomes widely accepted.]

Prenatal Diagnosis

Chen CP. Prenatal diagnosis, fetal surgery, recurrence risk and differential diagnosis of neural tube defects. *Taiwan.J.Obstet.Gynecol.* 2008 Sep;47(3):283-90.

Abstract: Prenatal screening with alpha-fetoprotein (AFP) and ultrasonography have allowed the prenatal diagnosis of neural tube defects (NTDs) in current obstetric care, and open spina bifida has been considered a potential candidate for in utero treatment in modern pediatric surgery. This article provides an overview of maternal serum AFP screening, amniotic fluid AFP assays, amniotic fluid acetylcholinesterase immunoassays and level II ultrasound for NTDs, prenatal repair of fetal myelomeningocele, recurrence risk of NTDs, and differential diagnosis of NTDs on prenatal ultrasound.

[This article provides a nice review of prenatal screening for NTDs.]

D'Addario V, Rossi AC, Pinto V, Pintucci A, Di CL. Comparison of six sonographic signs in the prenatal diagnosis of spina bifida. *J.Perinat.Med.* 2008;36(4):330-4.

Abstract: AIMS: To compare the diagnostic accuracy of sonographic signs that may be looked for in fetuses with spina bifida. METHODS: Forty-nine fetuses affected by spina bifida were enrolled, at a gestational age of 18-28 weeks. The following sonographic signs were looked for: "lemon" sign, small cerebellum, effaced cisterna magna, small posterior fossa, ventriculomegaly and direct visualization of a spinal defect. RESULTS: The "lemon" sign was present in 53%, a small cerebellum in 96%, an effaced cisterna magna in 93%, a small posterior fossa in 96%. Ventriculomegaly was present in 40/49 (81%) cases and was severe in 20 fetuses and borderline in the remaining 20. The spinal defect was missed in one fetus presenting the cerebellar and posterior fossa signs. In two fetuses, the myelomeningocele was present without cranial signs of Chiari II malformation and in both cases the defect was covered by intact skin. CONCLUSIONS: Our results confirm the usefulness of evaluation of the posterior fossa in the diagnosis of spina bifida, particularly in cases of small spinal defects that may be missed at ultrasound. Conversely, myelomeningocele covered by intact skin was not associated with the cranial signs of Chiari II malformation.

[This article describes the frequency with which various signs, observed on prenatal ultrasounds, occur in babies born with meningocele.]

Grosse SD, Ouyang L, Collins JS, Green D, Dean JH, Stevenson RE. Economic evaluation of a neural tube defect recurrence-prevention program. *Am.J.Prev.Med.* 2008 Dec;35(6):572-7.

Abstract: BACKGROUND: Women with a pregnancy affected by a neural tube defect (NTD) are encouraged to take folic acid prior to a subsequent pregnancy, but it is unknown whether organized attempts to identify and counsel such women to prevent recurrent NTDs are cost effective. METHODS: Data from the South Carolina recurrence-prevention program for October 2001-September 2002 were analyzed between October 2002 and December 2003 to calculate costs. Cost-effectiveness modeling of the program

during 1992-2006 was conducted during 2007. Results were calculated for three scenarios based on recurrence risk, supplement use, and the effectiveness of folic acid in preventing recurrences. For each scenario, quality-adjusted life years (QALYs) were calculated separately using prevented NTD-affected live births; prevented NTD-affected births (including fetal deaths); and all prevented NTD-affected pregnancies. RESULTS: The prevention program cost approximately \$155,000 per year in 2003 dollars to protect 35 pregnancies and prevent approximately one NTD. The direct costs associated with an NTD depend on type and outcome, but are approximately \$560,000 in 2003 dollars for a live birth with spina bifida. The base-case cost-effectiveness ratio was \$39,600 per QALY gained from avoided NTD-affected live births and stillbirths, and \$14,700 per QALY gained from the avoidance of all NTD-affected pregnancies. The baseline NTD recurrence risk and the use of folic acid supplements by women who are at high risk for an NTD-affected pregnancy were influential parameters. CONCLUSIONS: The South Carolina NTD recurrence-prevention program appears comparable in cost effectiveness to other preventive services. Other states might consider including NTD recurrence prevention in birth defect-prevention programs.

[The authors evaluated the cost effectiveness of a folic acid program in South Carolina. In this program, women were phoned on a regular basis; women who reported trying to get pregnant were reimbursed for prenatal vitamins and were provided with supplements containing folic acid. The program was calculated to cost \$39,600 per quality-adjusted life year, which is comparable to other prevention programs. These analyses tend to be somewhat arbitrary, but it is reassuring to know that a program like this has value similar to many other prevention programs.]

Koszutski T, Kawalski H, Kudela G, Wroblewska J, Byrka-Owczarek K, Bohosiewicz J. Babies with myelomeningocele in Poland: parents' attitudes on fetal surgery versus termination of pregnancy. *Childs Nerv.Syst.* 2009 Feb;25(2):207-10.

Abstract: BACKGROUND: Recent surgical advances have resulted in fetal surgery becoming an option for children with myelomeningocele (MMC). However, there is little information about the parents' attitudes towards such therapy. METHODS: Following a lecture on the current status and problems of fetal surgery for MMC, a 12-item questionnaire was administered to 58 parents of children with MMC. Questionnaire topics included knowledge of the disease and treatment options, as well as attitudes and concerns towards fetal surgery or termination of pregnancy. RESULTS: Following the lecture, 14 (out of 58) parents felt that knowledge of the disease would allow for abortion to be an option, while 18 were uncertain. Once informed of potential risks and benefits of fetal surgery, 34 parents had a positive or rather positive attitude towards this procedure. CONCLUSIONS: On the basis of these results, it was concluded that this population is potentially interested in the use of fetal surgery.

[This was a survey of parents attending a presentation on spina bifida. After the lecture more than half had a positive attitude about prenatal surgery.]

Winqvist B, Ogle K, Muhajarine N. Exploring physicians' views and values in relation to maternal serum screening. *J.Obstet.Gynaecol.Can.* 2008 Jul;30(7):564-72.

Abstract: OBJECTIVE: Little is known about physician support for prenatal screening. We sought a better understanding of physicians' values and opinions as they relate to prenatal screening, termination of affected pregnancies, and disability. METHODS:

Surveys were sent to all family physicians/general practitioners and obstetricians in Saskatchewan during May and June 2005. RESULTS: Of those physicians who responded, all obstetricians and 91% of family physicians reported offering maternal serum screening (MSS) to pregnant women in their practices. Of respondents who offered MSS to their patients, 87% of obstetricians and 72% of family physicians reported offering MSS to all pregnant women. Approximately one half of respondents agreed that they had enough knowledge to counsel a pregnant woman with a fetus affected by Down syndrome or spina bifida; 40% said the same about a fetus with trisomy 18. Twenty-six percent of physicians agreed that offering MSS was in conflict with their culture, religion, or personal value systems if it led to termination of pregnancy. One third of physicians reported having concerns about the increasing capacity for genetic testing of fetuses and the social, ethical, and clinical implications of such testing. CONCLUSION: Physicians held diverse views regarding prenatal screening, selective termination, and disability. Personal views and biases, in either direction, are relevant to our understanding of the clinical encounter and the ethical quandaries faced by practitioners. These value differences also may explain at least some of the variation in the use of MSS observed across the country, although the current study was not designed to make a causal link. There is a need to better understand how value differences affect the uptake of new reproductive technologies and the implications for health care policy and medical practice.

[This was a survey to ascertain whether physicians routinely ordered maternal prenatal serum screening (including screening MSAFP for neural tube defects). The majority of physicians offered screening, even if the consequences of the screening (termination of pregnancy) were in conflict with their values.]

Psychosocial Studies

Bellin MH, Kovacs PJ, Sawin KJ. Risk and protective influences in the lives of siblings of youths with spina bifida. *Health Soc. Work* 2008 Aug;33(3):199-209.

Abstract: The impact of childhood chronic health conditions like spina bifida (SB) is a shared family experience. However, the lived experience of siblings is not well known. One hundred and fifty-five brothers and sisters of a child with SB responded to an open-ended question included in an anonymous self-administered mail questionnaire designed to enhance awareness of how adolescent siblings experience this chronic condition. Content analysis performed by interdisciplinary authors identified four domains-- Rewards and Consequences of Spina Bifida, Journey Toward Acceptance of Spina Bifida, Emotional Climate of Siblings, and Qualities of the Social Environment--that depict emotional complexity in response to the diverse risk and protective influences present in the lives of siblings. By bolstering the siblings' strengths and resources found in their adaptive coping mechanisms, spirituality, cohesive family ties, and supportive peer friendships, social workers and other health care professionals may help them to successfully navigate the intense and confusing emotions that may naturally spring from the challenges and opportunities associated with SB.

[Siblings of children with spina bifida face many challenges. This paper reports on a survey of adolescent siblings. Common themes included the effect of the spina bifida on daily life and the omnipresence of the condition. Having a sibling with spina bifida also led to some unique opportunities. The authors encourage all of us to pay attention to siblings of children who have spina bifida, and to address their needs as well.]

Heffelfinger AK, Koop JI, Fastenau PS, Brei TJ, Conant L, Katzenstein J, Cashin SE, Sawin KJ. The relationship of neuropsychological functioning to adaptation outcome in adolescents with spina bifida. *J.Int.Neuropsychol.Soc.* 2008 Sep;14(5):793-804.

Abstract: Adolescents with spina bifida (SB) vary in their ability to adapt to the disease, and it is likely that numerous risk and protective factors affect adaptation outcomes. The primary aim was to test neuropsychological impairment, exemplified herein by executive dysfunction, as a risk factor in the Ecological Model of Adaptation for Adolescents with SB. Specific hypotheses were that: (1) executive functioning predicts the adaptation outcome of functional independence in adolescents with SB; (2) executive functioning mediates the impact of neurological severity on functional independence; and (3) family and adolescent protective factors are related to functional independence and moderate the relationship between executive functioning and functional independence. Forty-three adolescents aged 12-21 years completed neuropsychological measures and an interview that assessed risk, adolescent and family protective factors, and functional independence. Age, level of lesion, executive functioning, and the protective factor adolescent activities were significantly correlated with the functional independence outcome. In hierarchical regression analysis, the model accounted for 61% of the variance in functional independence outcomes. Executive functioning mediated the impact of neurological severity on functional independence.

[This study evaluated the factors that affect functional independence in adolescents with spina bifida, with a focus on executive function (the ability to plan, initiate, sequence, sustain, inhibit competing responses, and pace work). They found that age, level of lesion, executive functioning, and adolescent activities were significantly correlated with functional independence. Executive function is an indicator of the severity brain injury and is linked with attention and other factors related to learning. It is unclear whether any specific interventions can improve executive function.]

Sawin KJ, Bellin MH, Roux G, Buran CF, Brei TJ. The experience of self-management in adolescent women with spina bifida. *Rehabil.Nurs.* 2009 Jan;34(1):26-38.

Abstract: It is essential for youth with chronic health conditions like spina bifida (SB) to develop self-management skills to combat vulnerability, achieve self-sufficiency, and transition to adulthood. The purpose of this qualitative study was to describe the experience of self-management in 31 adolescent women with SB. Three themes emerged from this study: (1) opportunities to engage in self-management activities--knowledge, skills, and aspirations; (2) dance of individuation--parental impact on self-management; and (3) advocacy within self-management--confronting discrimination and stigma. The findings indicate that assessment and interventions to enhance self-management in adolescent women with SB are critical for supporting the range of condition-related and life skills needed for a transition to adulthood and independent living.

[In this qualitative assessment of self-management in female adolescents with spina bifida, three themes were expressed by the participants: opportunities for self-management, separation from families to become independent, and dealing with discrimination.]

Muller-Godeffroy E, Michael T, Poster M, Seidel U, Schwarke D, Thyen U. Self-reported health-related quality of life in children and adolescents with myelomeningocele. *Dev.Med.Child Neurol.* 2008 Jun;50(6):456-61.

Abstract: The aim of the study was to investigate self-reported health-related quality of life (HRQOL) in children and adolescents with myelomeningocele (MMC) and to examine the impact of clinical impairments and limitations in activities of daily living (ADL). Fifty patients (28 females, 22 males) between 8 and 16 years of age (mean age 12y 1mo [SD 2y 4mo]) with MMC from three German paediatric centres and their mothers completed standardized measures on HRQOL (KINDL-R) and limitations in ADL (Childhood Health Assessment Questionnaire). Lesion level was thoracic in nine, sacral in 25, and lumbosacral in 11 patients. Twenty-one patients were community walkers, 17 were able to walk in the home, and seven used a wheelchair. Two-thirds had health problems related to the central nervous system causing current difficulties (eight patients had a shunt, six had hydrocephalus, and 10 had a tethered cord). Patients in the study group reported diminished overall HRQOL compared with norm data, specifically in the dimensions of emotional well-being, self-esteem, and peer relations. Adolescents reported diminished HRQOL in the dimension of peer relations. Most medical parameters as well as limitations in ADL were not significantly associated with HRQOL. Our findings confirm the results of studies which dispute a linear inverse association between condition severity and HRQOL and emphasize the importance of peer relations in young patients with MMC.

[This study of health-related quality of life found that children and teens with spina bifida expressed problems with emotional well-being, self-esteem, and peer relations. These factors were unrelated to the level of lesion, suggesting that interventions to address these issues could be successful.]

Shields N, Taylor NF, Dodd KJ. Self-concept in children with spina bifida compared with typically developing children. *Dev.Med.Child Neurol.* 2008 Oct;50(10):733-43.

Abstract: The literature was systematically reviewed to determine if children with spina bifida have lower self-concept compared with their peers with typical development. Relevant trials were identified by searching electronic databases, supplemented by citation tracking. Of 803 papers initially identified, 15 met the inclusion criteria. Meta-analysis revealed children with spina bifida scored significantly lower than children with typical development for the domains of global self-worth ($d=-0.39$, 95% confidence interval [CI] -0.65 to -0.12); physical appearance ($d=-0.26$, 95% CI -0.46 to -0.06); athletic competence ($d=-0.45$, 95% CI -0.67 to -0.22); social acceptance ($d=-0.33$, 95% CI -0.55 to -0.11); and scholastic competence ($d=-0.43$, 95% CI -0.66 to -0.21). There was no difference between the groups for the behavioural conduct domain. Children with spina bifida on average have a lower self-concept than their peers with typical development. Clinicians need to take account of this information in planning the assessment and treatment of this group.

[This is a systematic review of the literature with statistical analyses that documents that children with spina bifida have lower self-concepts than otherwise healthy children. It is unknown, however, what sorts of interventions would improve self-concept in children with spina bifida.]

van't VT, Meester H, Poenaru D, Kogei A, Augenstein K, Bransford R. Quality of life for families with spina bifida in Kenya. *Trop.Doct.* 2008 Jul;38(3):160-2.

Abstract: Spina bifida (SB) affects children worldwide. Studies from developed nations have explored the impact of SB on the quality of life of children and their parents. However, there are no such studies available from developing countries. We have

therefore undertaken to document the impact of the disability on the families of affected children in Kenya. A questionnaire was administered to 40 mothers and their children, who were receiving treatment for SB at our institution. The results of this study should indicate where community and governmental resources and educational efforts for the disabled should be directed.

[This is a survey of 40 mothers in Kenya who had children with spina bifida. Many reported receiving no support from their community. Some reported that family members, even grandparents, had not seen the child because they were afraid of a curse. Incontinence often was more stressful than lack of mobility. Studies like this are very important because they document the cultural differences that affect the lives of children with spina bifida around the world.]

Vermaes IP, Janssens JM, Mullaart RA, Vinck A, Gerris JR. Parents' personality and parenting stress in families of children with spina bifida. *Child Care Health Dev.* 2008 Sep;34(5):665-74.

Abstract: **BACKGROUND:** Spina bifida (SB) can place parents at risk for increased levels of parenting stress. Little is known, however, about the role of parents' intrapersonal resources. Therefore, based on ideas of the Disability-Stress-Coping Model, relations between the severity of SB, parents' personality traits and parenting stress were examined. **METHODS:** Forty-six mothers and 37 fathers of children with SB (6-14 years) participated. Severity of SB (physical dysfunctions and cognitive functions), parental personality (Big Five) and parenting stress (Parenting Stress Index) were measured. Multiple regression analyses were performed. **RESULTS:** The severity of the child's physical dysfunctions was positively associated with parenting stress. Extraversion (mothers only), emotional stability and agreeableness (fathers only) were negatively related to parenting stress. In the final model, 64% of the variance in mothers' and 67% of the variance in fathers' levels of parenting stress was explained. Parents' personality traits explained the largest proportions of variance in parenting stress. **CONCLUSION:** Mobility, bladder and bowel dysfunctions in school-aged children with SB represent ongoing stressors for parents. Parents' intrapersonal resources of positive affectivity, however, are more important determinants of parental adjustment to SB than the child's physical dysfunctions.

[Having a child with spina bifida can be extremely stressful on families. This study showed that, while the degree of the child's disability affected this stress, parental personality and coping skills affected the stress even more.]

Vermaes IP, Gerris JR, Mullaart RA, Geerdink N, Janssens JM. PMTS and stress response sequences in parents of children with spina bifida. *Eur.J.Paediatr.Neurol.* 2008 Nov;12(6):446-54.

Abstract: **OBJECTIVE:** To test the presence and progress of Pediatric Medical Traumatic Stress (PMTS) symptoms in parents of children with spina bifida (SB). **METHODS:** Parents of 23 newborns with SB were interviewed prospectively and parents of 58 school-aged children with SB were interviewed retrospectively. PMTS symptoms were assessed with 17 DSM-IV criteria for the clusters Intrusion, Avoidance, and Increased Arousal. **RESULTS:** Within 3 months after the SB diagnosis, 75% of the parents met diagnostic criteria for symptoms of Intrusion and Increased Arousal, but not of Avoidance. In parents of school-aged children with SB, PMTS symptoms had declined in the first 4 years of the child's life and stabilized during the school years. Approximately 30% of the mothers and 20% of the fathers still met diagnostic criteria for Intrusion, Avoidance and Increased Arousal. In mothers of children with open SB, symptoms of

Intrusion and Increased Arousal had decreased more slowly than in mothers of children with closed SB. CONCLUSIONS: An SB diagnosis initially provokes traumatic stress symptoms in three-quarters of the parents; however, in most of them, these symptoms diminish during the first 4 years of the child's life. In a minority of the parents, severe stress symptoms persist beyond middle childhood. Professional psychological help may need to be offered to this selective group of parents whose levels of stress do not decline after the child's preschool years. Longitudinal research is needed to further investigate and confirm the trends that were found in parents' psychological adjustment to SB.

[The birth of a baby with spina bifida can be extraordinarily stressful for his or her parents. In this study the authors use the Pediatric Medical Traumatic Stress instrument to evaluate parents of infants as well as parents of school aged children. They found that stress generally decreased over time, and was fairly stable during elementary school years. Remember, however, that stress may increase at any time, for instance, during an illness or when a child is hospitalized, and on special occasions like the child's birthday or the first day of school.]

Wohlfeiler MM, Macias MM, Saylor CF. Paternal correlates of cognitive and behavioral functioning in children with myelomeningocele. *Dev.Med.Child Neurol.* 2008 Nov;50(11):864-9.

Abstract: This study examined paternal correlates of the cognitive and behavioral functioning of children with myelomeningocele, when controlling for maternal and biological/child correlates as possible sources of variance. Participants were 48 parent dyads of children with myelomeningocele (21 males, 27 females) between the ages of 4 and 12 years (mean 8y, 2mo, SD 2y 3mo). Lesion levels of participants ranged from the thoracic to sacral (thoracic-L3: n=15; L4-L5: n=15; sacral or lipomeningocele: n=18), of whom 38 had been shunted for hydrocephalus. Half of the participants (n=24) were community ambulators. Potential predictors of cognitive and behavioral functioning included paternal and maternal parenting stress, as assessed by the Parenting Stress Index - Short Form paternal, and maternal perceptions of support and resources, as assessed by the Family Resource Scale and the Family Support Scale, and child medical severity. Paternal variables significantly correlated with behavioral functioning but not with cognitive functioning. Regression analyses revealed that paternal personal distress and maternal perceived adequacy of social support accounted for significant variance in overall child behavioral functioning. Only child medical severity and annual household income explained significant variance in overall child cognitive functioning. These findings add to the growing body of theory and research documenting that fathers make unique and significant contributions to child adjustment in children with myelomeningocele. Both fathers and mothers need to be considered in interventions supporting development and adjustment of children with myelomeningocele and their families.

[This study evaluated cognitive and behavioral outcomes in children with meningomyelocele, focusing on fathers. They found that paternal personal distress and maternal perceived adequacy of social support (including from the child's father) affected the child's behavior. On the other hand, the child's cognition was affected most by the severity of the child's spina bifida and the annual household income (again supporting previous studies that have documented the importance of socioeconomic status on outcomes). Although most clinical encounters—and research studies—involve mothers but not fathers, this study reminds us that fathers are a critical part of the child's life and outcomes.]

Urology

Abrahamsson K, Jodal U, Sillen U. Reference interval for serum creatinine in children and adolescents with myelomeningocele. *Scand.J.Urol.Nephrol.* 2009 Jan 13;1-3.

Abstract: Objective. In children and adolescents with myelomeningocele (MMC), the relation between muscle mass and body composition varies considerably, making it difficult to evaluate the relevance of renal function assessments done with serum (s)-creatinine. Therefore, a reference interval for enzymic s-creatinine in individuals with MMC was constructed. **Material and methods.** Patients with MMC aged 1.5-18 years who consistently had chromium-51-labelled edetic acid clearance within normal limits (88-132 ml/min/1.73 m²) were selected for the study. Included were 52 girls and 71 boys with a total of 471 s-creatinine determinations. Children with normal body composition were used as controls. **Results.** The mean s-creatinine concentration at the age of 1.5 years was 22 micromol/l in girls and 21 micromol/l in boys and increased to 38 micromol/l in girls and 58 micromol/l in boys at the age of 18 years. There was no difference between the genders until the age of 12. From the age of 3 years, there was a significantly lower median s-creatinine concentration in the MMC children compared with a healthy age-matched population. The median concentration of the MMC children was equivalent to the 2.5 percentile of the control group. **Conclusion.** From the age of 3 years, the reference interval for children and adolescents with MMC showed a lower median concentration for enzymic s-creatinine and a greater interindividual variation compared with the healthy age-matched population.

[Serum creatinine is an important marker for renal function. However, it depends in part on muscle mass, and children with meningomyelocele have altered muscle mass. This study helps establish normative values for children with meningomyelocele for this important marker of renal health.]

Dodson JL, Furth SL, Hsiao CJ, ener-West M, Levey EB, Wu AW, Gearhart JP. Health related quality of life in adolescents with abnormal bladder function: an assessment using the Child Health and Illness Profile-Adolescent Edition. *J.Urol.* 2008 Oct;180(4 Suppl):1846-51.

Abstract: PURPOSE: We studied the impact of abnormal bladder function due to congenital urological disorders on health related quality of life in children. A reliable patient based method is needed to assess the impact of these conditions in children and the interventions used to treat them. **MATERIALS AND METHODS:** Participants 11 to 17 years old with bladder exstrophy-epispadias complex, spina bifida or other causes of abnormal bladder function self-administered the Child Health and Illness Profile-Adolescent Edition, a generic health related quality of life instrument. They also responded to questions about incontinence, catheterization status and bother level. Mean scores on the profile were compared to population based norms. **RESULTS:** Mean age of the 50 participants was 14.9 years, 62% were male and 82% were white. Diagnoses included bladder exstrophy-epispadias complex in 37 patients, spina bifida in 10 and other in 3. The mean +/- SD score on the disorders domain of 14.2 +/- 6.3 was significantly worse than the population norm of 20. Mean scores on the satisfaction, discomfort, resilience, risks and achievement domains were comparable to or better than the population based norm of 20. A total of 29 participants reported incontinence and 31 performed catheterization. **CONCLUSIONS:** In this study of adolescents with congenital causes of abnormal bladder function Child Health and Illness Profile-Adolescent Edition

generic health related quality of life scores were significantly worse in the disorders domain but largely comparable to or better than those of the general population in other domains. This suggests that the profile may discern between adolescents with structural urological disease and norms but it may not be sensitive enough to fully detect the impact of the condition. Alternatively adolescents may adapt well to the challenges of urological disease.

[Many instruments to assess 'health-related quality of life' are available. This study utilized the Child Health and Illness Profile-Adolescent Edition to evaluate teens who had a variety of conditions leading to abnormal bladder function. The researchers found that quality of life was affected in the physical aspects of life related to the bladder. However, it was not adversely affected in other domains like satisfaction, discomfort, resilience, risks, and achievement. These findings are consistent with previous studies that have shown that while physical factors are affected by meningomyelocele and its severity, other domains are affected more by other characteristics of the child or teen like temperament, intelligence, and by environmental factors like socioeconomic factors and support, especially from parents.]

Flannery T, Shoakazemi A, McLaughlin B, Woodman A, Cooke S. Dialysis disequilibrium syndrome: a consideration in patients with hydrocephalus. *J.Neurosurg.Pediatrics* 2008 Aug;2(2):143-5.

Abstract: Dialysis disequilibrium syndrome is a rare neurological manifestation of intermittent hemodialysis. Urea removal occurs more slowly across the blood-brain barrier than from the plasma, generating an osmotic gradient that promotes water movement into the brain and cerebral edema. The authors report the development of dialysis disequilibrium syndrome in a patient with spina bifida and an adequately functioning shunt.

[Unfortunately, many teens and adults with spina bifida are on dialysis. This paper describes the occurrence of neurological symptoms in a person with spina bifida and shunted hydrocephalus due to dialysis. This is an unusual occurrence, but one that should be considered in any person with hydrocephalus who is being dialyzed.]

Hipp JA, Hipp JD, Yoo JJ, Atala A, Andersson KE. Microarray analysis of bladder smooth muscle from patients with myelomeningocele. *BJU.Int.* 2008 Sep;102(6):741-6.

Abstract: **OBJECTIVE:** To examine whether gene profiles can provide a molecular evaluation of the quality and therapeutic potential in patients with myelomeningocele (MM), by comparing genetic profiles of smooth muscle cells (SMCs) from healthy bladders and bladders from patients, to identify genes that are over- and under-expressed in MM bladder SMCs. **MATERIAL AND METHODS:** Bladder SM biopsies were obtained from 'healthy' subjects undergoing bladder surgery for vesico-ureteric reflux and from patients with a neurogenic bladder secondary to MM. Bladder SMCs were expanded in vitro and total RNA was isolated and hybridized to gene chips to evaluate the differential expression levels of 22 283 genes. Differentially expressed genes were identified by two methods. In the first analysis, we directly compared raw data sets of healthy SMCs to those derived from patients with MM. In the second analysis, we indirectly compared healthy SMCs and MM SMCs to a reference file, to create a genetic signature of genes that are over- and under-expressed in MM SMCs. **RESULTS:** The direct analysis identified 240 genes that were over-expressed and 104 that were under-expressed in MM SMCs. Gene ontology classifications were used to identify biological

themes and pathways. Genes that were over-expressed in MM SMCs were involved in development: mesenchyme homeobox 2 (-fold change, 9.3); bone morphogenic protein 6 (4.0); fibroblast growth factor 2 (4.8); inhibin A (4.2), cartilage oligomeric matrix protein (9.97); collagen 11A (6); collagen 5A2 (3) and collagen 1A1 (2.18). The indirect analysis identified 665 genes that were over-expressed and 1343 that were under-expressed in MM SMCs. Pathway-based analysis of these genetic signatures showed an over-expression of genes involved in muscle development and focal adhesion/extracellular matrix interactions. Genes that were under-expressed in MM SMCs were mapped to muscle contraction, transmission of nerve impulses, and cell-cell adhesion pathways. **CONCLUSION:** Our results are consistent with previous studies showing that MM bladders have an excess of extracellular matrix deposition, improper contraction, and are developmentally immature relatively to healthy SMCs. The clinical implication of microarray analysis of MM SMCs is that it provides potential targets that could induce muscle differentiation and inhibit extracellular matrix production.

[Smooth muscle cells from the bladders of persons who have meningocele are different from typical cells. They have more extracellular matrix, improper contraction, and are developmentally immature. This study evaluated the genetic factors related to these differences.]

Kajbafzadeh A, Mahboubi AH, Payabvash S. Concomitant repeated intravesical injections of botulinum toxin-type A and laparoscopic antegrade continence enema; a new solution for an old problem. *BJU.Int.* 2008 Dec 22.

Abstract: **OBJECTIVE** To report our experience of treating bladder and bowel dysfunction in children with myelomeningocele, with simultaneous laparoscopic antegrade continence enema (LACE) and repeated intravesical injection of botulinum toxin-type A (BTX-A). **PATIENTS AND METHODS** Six girls and 14 boys (mean age, 8.7 years) with myelomeningocele were included in this study. All patients had received one or two intravesical injection(s) of BTX-A, but had persistent fecal incontinence or constipation despite improved urinary symptoms. We performed a two-port laparoscopic appendicostomy, immediately after repeated intravesical injection of BTX-A, through a V-shaped skin flap at McBurney's point. The stoma was finally covered by a quadrilateral skin flap, using the 'VQ' technique. The degree of urinary incontinence and bowel dysfunction were determined in each patient, and conventional urodynamic studies were performed 4 months after each injection. **RESULTS** All patients were followed-up for a mean (range) of 19.1 (14-33) months. Urinary continence improved significantly after the first injection, and remained constant after repeat injections. The maximum detrusor pressure, bladder compliance and capacity improved significantly ($P < 0.001$) compared with baseline. Interestingly, the simultaneous intravesical BTX-A injection/LACE procedure significantly improved all urodynamic variables compared with the values obtained after the last BTX-A injection alone. The laparoscopic procedure was well tolerated, and 19 (95%) children were nappy-free at the final follow-up. Only two patients had stoma stenosis, and one patient had minor stoma leakage. **CONCLUSION** Concomitant repeat intravesical injection of BTX-A and LACE can effectively manage bladder and bowel dysfunction in children with myelomeningocele. The procedure may further contribute to improve bladder urodynamic function, as effective evacuation of the bowel provides more room for bladder distension.

[This Iranian study documents the results of treating children with detrusor (bladder muscle) hyperreflexia and fecal incontinence using the combination of the ACE procedure (done laparoscopically) and repeated injections of the detrusor muscle using botulinum toxin (Botox). The follow-up was short—only 19 months—but most of the children did well with the combination

of therapies.]

Kari JA, Safdar O, Jamjoom R, Anshasi W. Renal involvement in children with spina bifida. Saudi.J.Kidney Dis.Transpl. 2009 Jan;20(1):102-5.

Abstract: Renal scarring and renal failure remain life-threatening for children born with spinal dysraphism. An early start of therapy helps to safeguard renal function for such children and avoid end-stage renal disease. However, optimal care is not always available in developing countries. We reviewed our data on all newborns with spina bifida who were born at King Abdulaziz University Hospital between 1997 and 2006. Thirty-three children with myelomeningocele (MMC) were evaluated; MMC site was thoracolumbar in 26 patients (77.1%) and in the lumbosacral area in 7 patients (22.9%). The mean age at the time of evaluation was 5.4 +/- 2.3 years. Thirty (90%) patients presented with neurogenic bladder, and 26(78%) with vesico-uretral reflux (VUR). Only 8 patients (group A) received clean intermittent catheterization (CIC), while the rest (group B) were either non-complaint or not on any therapy. Urinary tract infections overall were 4.5 +/- 3.8 per year. Patient undergoing CIC had a lower number of UTI (mean per year) 3.3 +/- 1.2 vs 6.6 +/- 2.3. Sixty two percent of group A had VUR compared with 93% in group B. The mean creatinine was 46 +/- 39 micromol/L for the whole group. However, group A had a lower mean creatinine 38 +/- 11 compared to 50 +/- 34 in group B. In conclusion, early intervention to relieve urinary retention in children born with spina bifida resulted in preserving renal function and less incidence of VUR and UTI. There is a need of more awareness about the importance of starting proactive treatment of risks of upper urinary tract disease and development of renal failure in babies with spina bifida.

[This study from Saudi Arabia documents urological care and outcomes of 33 children with meningomyelocele. Only 24% of the children were using clean intermittent catheterization (CIC). Those who were using CIC had fewer urinary tract infections, less renal scarring, less reflux, and better kidney function (as measured with creatinine). We often take a simple procedure like CIC for granted, but it has been a life-saving development in the care of children with meningomyelocele.]

Katrancha ED. Clean intermittent catheterization in the school setting. J.Sch Nurs. 2008 Aug;24(4):197-204.

Abstract: Spina bifida (SB) is a neural tube defect that causes many physical and mental disabilities. Bowel and bladder incontinence is the disability seen most often in these students that requires the school nurse's attention. Clean intermittent catheterization (CIC) provides the student with SB a vehicle to reach a satisfactory level of continence, helps build self-esteem, and allows the student greater independence. The school nurse assists the student with SB with CIC in the school setting and reinforces the importance of correct technique with the student, family, and other personnel caring for the student. The success of a continence management program at school is dependent on all these individuals working together with the common goal of maintaining normal renal function, gaining urinary continence, and promoting independence of the student.

[We often take clean intermittent catheterization (CIC) for granted. However, it is a critical part of the care for children who have meningomyelocele. This article reviews the role of the school nurse in this process of care.]

Patel RP, Kolon TF, Huff DS, Carr MC, Zderic SA, Canning DA, Snyder HM, III. Cryptorchid

testis histopathology in myelomeningocele patients. *J.Pediatr.Urol.* 2008 Dec;4(6):434-7.

Abstract: **PURPOSE:** Cryptorchidism occurs in 25% of boys with myelomeningocele (MMC) compared to 3% of the general population. Testicular biopsy histopathology correlates with future sperm counts. We studied testicular histology in boys with cryptorchidism and MMC to investigate if the MMC influences histological findings. **MATERIALS AND METHODS:** The study group consisted of six patients with MMC and undescended testis (UDT) who underwent orchiopexy and bilateral testis biopsy. Twelve testicular biopsies from six patients were compared to 40 biopsies from 20 UDT-only controls. Total germ cell count per tubule (TGC/T) and the percentage of adult dark spermatogonia (%Ad) in undescended and contralateral descended testes from the patients were compared with controls. **RESULTS:** In the study group, two had total absence of germ cells (TGC/T=0) and three had severely reduced germ cells (TGC/T<0.2). Four had total absence of Ad spermatogonia and the remaining two had severely reduced Ad spermatogonia (%Ad=5). The mean TGC/T and %Ad in patients with UDT and MMC were conspicuously lower than controls. The differences did not reach statistical significance (P=0.09-0.29). **CONCLUSION:** These results suggest that patients with both MMC and UDT have a more severe reduction in total number and more severely delayed maturation of germ cells than do patients with UDT alone. With only six patients in this study, there was not the power to detect statistical significance. In addition to the reproductive problems due to erection and ejaculatory dysfunction in patients with MMC, this severe testicular histopathology may increase the risk of subfertility.

[Undescended testicle is somewhat more common in boys with meningomyelocele than in otherwise healthy boys. In this study the undescended testes from boys with MM showed fewer and more immature germ cells. The authors hypothesize that these differences may be present in the descended testes as well, and may account for some of the infertility seen in males with MM.]

Scales CD, Jr., Wiener JS. Evaluating outcomes of enterocystoplasty in patients with spina bifida: a review of the literature. *J.Urol.* 2008 Dec;180(6):2323-9.

Abstract: **PURPOSE:** The urological complications of spina bifida impose a significant burden of disability and disease. Therapy is focused on the bladder to achieve the primary goals of maintaining normal renal function and attaining urinary continence. When medical management fails, surgical intervention, including enterocystoplasty, is frequently performed. However, practice patterns for enterocystoplasty show significant variation. Given this context, we examined outcome measures for enterocystoplasty in patients with spina bifida. **MATERIALS AND METHODS:** A MEDLINE search was performed for articles on enterocystoplasty in patients with spina bifida. A total of 226 articles were identified and manually reviewed for relevant studies. Additional articles were selected based on a cited reference search. **RESULTS:** Almost all studies are retrospective, single institution case series of a relatively small number of patients. Few uniform or validated outcome measures for enterocystoplasty exist but reported measures typically include urodynamic, continence and satisfaction parameters. Interinstitutional variability in urodynamic measurements and in definitions of continence makes a comparison of outcomes difficult. The complication rate following enterocystoplasty is significant, well described and primarily related to the use of gastrointestinal segments for urine storage. **CONCLUSIONS:** Medical management is the mainstay of neurogenic bladder therapy in the spina bifida population. Enterocystoplasty remains an important

option to prevent or reverse upper tract deterioration, and/or improve or cure socially unacceptable incontinence despite poorly defined outcome measures. The development of appropriate and validated outcomes measures may enable more uniform, effective and safe urological care of patients with spina bifida.

[This article reviews 226 articles related to bladder augmentation (enterocystoplasty) in people with spina bifida. The authors note that most studies had weak methodological design, being retrospective with small numbers of patients and having inconsistent outcomes. They also note the high frequency of complications. This topic, like many others, cries out for collaborative research using standardized techniques and evaluations.]

Storm DW, Fulmer BR, Sumfest JM. Robotic-assisted laparoscopic approach for posterior bladder neck dissection and placement of pediatric bladder neck sling: initial experience. *Urology* 2008 Nov;72(5):1149-52.

Abstract: OBJECTIVES: Bladder neck sling cystourethropexy is a common procedure used to correct intrinsic sphincter deficiency in children with spinal dyspharism. Various modifications of the procedure have been made but all involve circumferential dissection of the bladder neck and proximal urethra. The posterior dissection can be challenging and can result in injury to the rectum, urethra, or vagina. The posterior approach to the bladder neck as reported by Lottmann and later by de Badiola addresses these potential complications. Using these principles of the posterior approach, we performed a robotic-assisted laparoscopic placement of an acellular human dermal allograft bladder neck sling in 2 patients. We present our initial experience regarding this surgical technique.

METHODS: The diagnosis of intrinsic sphincter deficiency was established in 2 female patients, aged 9 and 10 years. Both patients had a neurogenic bladder secondary to spina bifida. Video urodynamics confirmed adequate bladder compliance and intrinsic sphincter deficiency. Robotic-assisted laparoscopic placement of a bladder neck sling was performed in both patients. RESULTS: Both procedures were completed intracorporeally. The mean blood loss was 20 mL. The mean operative time was 189 minutes. No intraoperative or postoperative complications occurred. The mean hospital stay was 3 days (range 2-4). The follow-up ranged from 13 to 22 months. Postoperative studies revealed continued low-pressure, compliant bladders and stable upper tracts. At last follow-up, the 2 patients were using catheterization without difficulty and were continent. CONCLUSIONS: The robotic-assisted laparoscopic approach to performing bladder neck dissection and placement of a bladder neck sling in children is technically feasible.

[This paper describes the use of the DaVinci robotic surgical system to assist with the placement of bladder neck slings in two girls with spina bifida. Both girls have shown improvement in bladder function without major adverse effects. Robotics are being used increasingly to assist with surgical procedures. Stay tuned to see what happens in this type of delicate procedure.]

Vajda P, Kispal Z, Lenart I, Farkas A, Vastyán AM, Pinter AB. Quality of life: urinary bladder augmentation or substitution in children. *Pediatr.Surg.Int.* 2009 Feb;25(2):195-201.

Abstract: PURPOSE: Bladder augmentation and substitution has been assumed to improve health-related quality of life in patients with urinary incontinence. This study was performed to elicit an evidence base for or against the above hypothesis.

METHODS: Between 1988 and 2006, 67 bladder augmentations and 7 bladder substitutions were performed at our institute. Inclusion criteria for the cross-sectional

study were a postoperative period of more than 1 year and an age of at least 10 years at the time of operation. A multimodality treatment-specific questionnaire (comprising 38 questions) was designed and sent to 61 patients. Quality of life was investigated in all patients and between the groups of patients with meningomyelocele (Group A) versus bladder exstrophy (Group B), patients, who are catheterizing themselves via urethra (Group C) versus stoma (Group D) and patients who are using (Group E) versus not using wheelchair (Group F) following the surgery. For the statistical analysis Students t test, Wilcoxon signed rank test and correlation analysis were used. RESULTS: A significant overall improvement was found in patients quality of life following this surgery ($P < 0.05$). Ninety percent of patients would prefer again bladder augmentation or substitution to their previous state. Patients with meningomyelocele are changing pads or diapers more frequently than exstrophy patients because of their bowel problems postoperatively. Quality of life improved better in patients performing CIC via stoma than in patients who perform it via their native urethra ($P < 0.05$). Outcomes were independent of patients age and of the post-augmentation time to assessment ($P < 0.05$). CONCLUSIONS: Bladder augmentation or substitution significantly improved the health-related quality of life in children and young adolescents taking part in the study. The authors are planning a prospective long-term follow-up of the patients (longitudinal study) to validate the results.

[In this Hungarian study, 61 patients who had bladder augmentation were surveyed. Those who had the creation of a catheterizable stoma seemed to be happier than those who continued catheterizing through their own urethra. The authors plan to follow this group prospectively.]

Welk BK, Afshar K, Rapoport D, MacNeily AE. Complications of the catheterizable channel following continent urinary diversion: their nature and timing. *J.Urol.* 2008 Oct;180(4 Suppl):1856-60.

Abstract: PURPOSE: We reviewed the incidence, nature and timing of complications related to the catheterizable channel following continent urinary diversion. MATERIALS AND METHODS: We retrospectively reviewed the records of 67 patients who underwent continent urinary diversion at British Columbia Children's Hospital from 2000 to 2006. Catheterizable channels included 54 Mitrofanoff appendicovesicostomies and 13 ileovesicostomies. Medical records were reviewed for predetermined complications and their timing, that is early -12 months or less, or late - more than 12 months. RESULTS: At a median followup of 28 months (range 3 to 62) a total of 17 complications were identified in 14 patients (21%). Superficial cutaneous stenosis developed in 4 of 67 cases (6%) as an early and as a late complication. These cases were initially treated with operative dilation and surgical revision as necessary. Channel stricture, which developed in 4 of 67 patients (6%) as an early and as a late complication, was treated with operative revision in 2 and endoscopic resection in 2. Three patients (5%) had stomal prolapse, which was generally a late occurrence and required operative revision in all. Channel leakage developed in 6 of 67 patients, presenting as an early complication in 50%. Endoscopic injection of bulking agents was attempted in 4 of these patients and it was successful in 2. Overall 82% of complications were successfully managed by endoscopic or superficial procedures. CONCLUSIONS: Complications of the catheterizable channel are a frequent and challenging problem. They appear to occur throughout the life of the channel with most developing within the first 2 years. Further followup is required to assess the performance and durability of continent catheterizable channels in children as patients progress to adulthood.

[One of the urological procedures for children and teens that has increased dramatically recently

is the creation of catheterizable stomas (channels). However, strictures (constrictions), leakage and prolapse (turning inside out) are common complications that are documented in this paper.]

Woodhouse CR. Myelomeningocele: neglected aspects. *Pediatr.Nephrol.* 2008 Aug;23(8):1223-31.

Abstract: The commonest cause of neurogenic bladder in children is myelomeningocele. Survival of children is much improved in the Western world, but by 35 years old, about 50% will have died. In adults, the commonest causes of death are lung and heart diseases. All physical aspects deteriorate with age, especially in those with thoracic lesions. Those who walk in childhood have a 20-50% chance of becoming wheelchair dependent as adults. Immobility, poor respiratory reserve, obesity, latex allergy and worsening kyphoscoliosis contribute to the increased risks of surgery. It is essential that safe and manageable urine drainage is established in childhood: the bladder never improves with time, and surgical reconstruction becomes progressively more difficult. Independence in adult life will only be possible with intense preparation in childhood. Children must be allowed to join in with family chores and events. Education, both academic and practical, must be encouraged. Skills such as driving, shopping and birth control must be taught. However, even with the best support, less than 40% will have gainful employment. Children who are continent and have lesions below L2 are likely to have normal sexual function. Sexual activity in adolescents, especially in those with hydrocephalus, is limited (but not absent). However, by adult life, about two thirds will have established a regular partnership. All females and those males who are naturally potent are likely to be fertile. There is a high risk of neural tube defects in their offspring unless the female partner takes prophylactic folic acid for 3 months before pregnancy and for first trimester.

[This is a sobering review discussing the prognosis and complications that occur in people with meningomyelocele. Much of the data presented are based on older studies. Unfortunately, for many areas the only data available are older data. Although for most topics the outlook is not quite as bleak as it is presented in this article, these issues still must be addressed.]

Zegers BS, Winkler-Seinstra PL, Uiterwaal CS, de Jong TV, Kimpen JL, de Jong-de Vos van Steenwijk CC. Urinary tract infections in children with spina bifida: an inventory of 41 European centers. *Pediatr.Nephrol.* 2008 Dec 9.

Abstract: The introduction of clean intermittent catheterization (CIC) in 1972 and low-dose chemoprophylaxis (LDCP, antibiotic prophylaxis), anticholinergic medication and urological surgery in the mid-1980s has improved the long-term outcome of renal function in children with neurogenic bladder sphincter dysfunction (NBSD) due to spina bifida (SB). We have conducted a European survey of the protocols for diagnosing and treating urinary tract infections (UTIs) in these children, using a web-based questionnaire. The responses from 41 centers in 14 European countries confirm that although most centers have standardized protocols for treating UTIs, there is no consensus among European centers in terms of protocols for preventing, diagnosing and treating UTIs in children with NBSD and for CIC.

[Urinary tract infections (UTI) are much more common in people with meningomyelocele than in otherwise healthy individuals. However, controversy exists about what constitutes a UTI—is it the number of bacteria, the presence of symptoms or some combination of these? Controversy also exists about the wisdom of trying to prevent subsequent infections with prophylactic antibiotics. This study is a survey from 41 centers across Europe that treat children with spina bifida. No consensus was found with regard to diagnosis, prevention or treatment of UTIs.

Having uniform guidelines and consistent treatment has been shown in other conditions to improve outcomes of care. These should be developed for urological management of children with spina bifida.]

Wound Care

Contractor D, Amling J, Brandoli C, Tosi LL. Negative pressure wound therapy with reticulated open cell foam in children: an overview. *J.Orthop.Trauma* 2008 Nov;22(10 Suppl):S167-S176.

Abstract: This article summarizes the results of a comprehensive review of the literature on the use of negative pressure wound therapy with reticulated open cell foam (NPWT/ROCF) as delivered by V.A.C.(R) Therapy (KCI, San Antonio, TX) in pediatric patients. A review of the literature revealed 20 articles that discussed the use of NPWT/ROCF in exclusively pediatric patients. Nine articles were retrospective reviews, and 11 were case studies. This review discusses the insights from these articles. This review discusses the versatility of NPWT/ROCF for use with pediatric patients with infected wounds; full-thickness burns; open fractures; large soft tissue wounds; surgical wounds of the chest, abdomen, and spine; pilonidal disease; and pressure ulcers. NPWT/ROCF has been used in children as young as a few weeks of age, and in children with comorbidities such as congenital heart disease, immunosuppression, and spina bifida. Wound healing in children can be delayed by impaired perfusion, infection, edema, and poor nutrition. Clinical considerations for using NPWT/ROCF in children can include differences in healing due to higher granulation rates requiring more frequent dressing changes, poor nutritional status, small size, and low weight. With pediatric patients, there is no consensus on foam (white or black) selection, optimum amount of negative pressure, frequency of NPWT/ROCF dressing changes, and interposing contact layer selection. Randomized prospective studies are needed to make recommendations for safe and efficacious clinical practice. Research regarding the effects of dressing types, adjunctive treatment, and wound healing in neonates and children is needed.

[This study reviews the use of a wound care system that uses negative pressure over the wound—to increase blood flow—and a foam system that assists granulation and enhances the removal of fluid. Some of the patients in these series had spina bifida with decubitus ulcers. None of the studies were prospective controlled trials, which provide the strongest evidence of a therapy. The golden rule for the care of decubitus ulcers is that you can put anything you want on them, except the patient. Until better controlled trials are done we still do not know the optimal treatment for these lesions.]