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Chapter 1 : Anencephalus in Scotland - Europe PMC Article - Europe PMC

Title: Anencephalus, spina bifida and congenital hydrocephalus, England and Wales Issue 32 of Studies on medical and population subjects.

Data are based on 16 state-based birth defects surveillance systems. View Large Folate and non-NTD birth defects While the relationship between folate and NTD is well established, folate deficiency may also be related to other serious birth defects. A randomized control trial of periconception folic acid-containing multivitamin supplementation demonstrated a reduced occurrence of urinary tract and cardiovascular congenital abnormalities and congenital limb deficiencies 7. The occurrence of orofacial cleftings, cleft lip and cleft palate, may also be reduced by a high dose of folic acid. This preventive effect may be the result of other mechanisms of action. One investigator has suggested that cleftings may be related to the compensation of impaired mitosis caused by a folate deficiency 7. A large-population CDC study demonstrating protection against NTDs by folic acid 11 also analyzed other birth defects. To further explore the relationship between folate and non-NTD birth defects, a large epidemiologic database of women taking medications during pregnancy was examined for the risks of non-NTD birth defects such as oral clefts and cardiovascular and urinary tract defects in babies born to mothers taking two general classes of folate antagonists 19 , The analysis revealed two important findings: This latter group presumably exert their antifolate activity through some other mechanism s. The former group of medications includes relatively commonly prescribed medications trimethoprim, triamterene, methotrexate, aminopterin and sulfasalazine. The anticonvulsants studied were carbamazepine, phenytoin, phenobarbital and primidone. The striking protective effects of folic acid on birth defects for mothers taking DHFR inhibitors help confirm the importance of folic acid, and its deficiencies, in the development of non-NTDs as well as NTDs 19 , Equally striking was the finding that folic acid had no appreciable protective effect in mothers taking antiepileptics. These data suggest that antiepileptics interfere at other levels of folate metabolism. Perhaps larger doses or a longer duration of folic acid treatment prior to conception would reduce the risk of birth defects in their offspring. Of course, drug dosing may be affected by folic acid, so drug levels would need to be studied in experimental and clinical settings. The mothers in the study with affected babies may have had increased susceptibilities to folic acid deficiencies that were unmasked by drug provocation. Clearly, folic acid supplementation must be given to all women of childbearing age taking antifolate medications. Moreover, the risks of birth defects must be weighed for women needing to take anticonvulsants. Modified pharmacologic approaches to prevention may be effective, such as use of other forms or higher doses of folic acid. Folate deficiency is associated with chromosomal breakage in vitro This raises the possibility that at least some of the chromosomal abnormalities and their attendant birth defects may be traced to folate deficiencies. A recent case report speculates that abnormal maternal folate metabolism a mutation in the MTHFR gene , combined with folate deficiency, may have been predisposing factors for a child born with both Down syndrome and spina bifida The trisomy 21 may have been promoted through folate-dependent mechanisms of chromosomal instability and meiotic nondisjunction. This case may catalyze research on the potential link between disturbed folate metabolism and meiotic and possibly mitotic chromosomal errors Folic acid protection As described above, a number of studies have shown an association between increased periconceptional intake of folic acid and a reduced incidence of structural developmental anomalies. Increased folic acid intake is associated with significantly fewer NTDs in combination with another major birth defect, particularly orofacial clefts, cardiac and limb defects and omphalocele 7. The combined data, placed on a background of strong biologic plausibility, make it highly likely that periconceptional intake of folic acid can reduce the risk of a variety of major birth defects. Folic acid from vitamin supplements and fortified foods is more readily absorbed than is folate naturally contained in food. There are two general forms of folic acid used in biological studies: These data led the U. Public Health Service in to recommend that all fertile women of child-bearing age consume 0. Folate levels

can be determined in several ways for individual and population studies. RBC folate levels generally reflect longer-term tissue stores, whereas serum or plasma levels are more indicative of short-term dietary intake 23 , 24 , 7. Thus, serum folate concentration may be a valid diagnostic test, particularly if done in conjunction with RBC folate values. Serum studies are technically less demanding and less dependent on sample handling and thus may be at times more appropriate for population field testing. As of January , the U. The effects of fortification on folate levels among nonpregnant women of childbearing age aged 15â€”44 y were assessed. The findings indicate substantial increases in serum and RBC folate concentrations among women of childbearing age. Similarly, the roughly bell-shaped distribution of folate levels in this population has shifted overall to higher levels. Findings by other recent studies support the CDC data documenting increased folate levels in women of childbearing age Impact of folic acid on risk for NTDs Since the late s, the reported prevalence of anencephaly and spina bifida in the U. These declines likely represent two factors: Similar trends have been reported in the United Kingdom To ascertain the true impacts from folic acid, birth certificate surveillance data for these defects may need to include data from pregnancies that are prenatally diagnosed and then lost or terminated 2. The rate of spina bifida has declined significantly between and 3 , The rate of spina bifida in was In contrast, after a decline in the early part of the decade, the anencephalus rate has been stable since The rate of anencephalus in was While this goal appears to have been nearly achieved, there are two approaches that provide the prospect of further reductions: Using data from a case-control study, the risk of an NTD has been found to be inversely proportional to early pregnancy maternal RBC folate levels in a continuous dose-response relationship 9. The particular susceptibility to the more severe NTD, anencephaly, is unknown. This defect may correlate with a more severe folate deficiency. These factors may eventually be discerned through animal and genetic epidemiologic research.

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Chapter 2 : - NLM Catalog Result

Get this from a library! Anencephalus, spina bifida and congenital hydrocephalus, England and Wales [Stephen Clifford Rogers; Josephine A C Weatherall; Great Britain.

Single umbilical artery [4] Causes Patau syndrome is the result of trisomy 13, meaning each cell in the body has three copies of chromosome 13 instead of the usual two. Patau syndrome can also occur when part of chromosome 13 becomes attached to another chromosome translocated before or at conception in a Robertsonian translocation. Affected people have two copies of chromosome 13, plus extra material from chromosome 13 attached to another chromosome. With a translocation, the person has a partial trisomy for chromosome 13 and often the physical signs of the syndrome differ from the typical Patau syndrome. Most cases of Patau syndrome are not inherited, but occur as random events during the formation of reproductive cells eggs and sperm. An error in cell division called non-disjunction can result in reproductive cells with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of the chromosome. Mosaic Patau syndrome is also not inherited. It occurs as a random error during cell division early in fetal development. Patau syndrome due to a translocation can be inherited. An unaffected person can carry a rearrangement of genetic material between chromosome 13 and another chromosome. This rearrangement is called a balanced translocation because there is no extra material from chromosome Although they do not have signs of Patau syndrome, people who carry this type of balanced translocation are at an increased risk of having children with the condition. The most common characteristics of this syndrome are problems such as late development, mental disability, multiple malformations, cardiomyopathy, and kidney abnormalities. The most common physical signs for Patau Syndrome are the decreasing of muscle tone, small hands, small ears, small head and mouth, as well as wide and short hands with short fingers. Physical development for children affected by Patau Syndrome occurs more slowly than children without Patau syndrome. However, children affected by Patau Syndrome should still undergo regular physical activity, even though muscle development may occur more slowly. Diagnosis Diagnosis is usually based on clinical findings, although fetal chromosome testing will show trisomy While many of the physical findings are similar to Edwards syndrome there are a few unique traits, such as polydactyly. However, unlike Edwards syndrome and Down syndrome , the quad screen does not provide a reliable means of screening for this disorder. This is due to the variability of the results seen in fetuses with Patau. Treatment of Patau syndrome focuses on the particular physical problems with which each child is born. Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help individuals with Patau syndrome reach their full developmental potential. Surviving children are described as happy and parents report that they enrich their lives. One and ten year survival was Klaus Patau in

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Rogers SC, Morris M. Infant mortality from spina bifida, congenitalhydrocephalus, monstrosity, and congenital dseases of the cardiovascular system in England and Wales. Ann Hum Genet. Feb; 34 (3)

A family study of major central nervous system malformations in South Wales. A study of major congenital defects in Japanese infants. An epidemiological study of congenital malformations in New York State. Changes in the incidence of anencephalus. Completeness and accuracy of reporting malformations on birth certificates. Congenital anomalies in the newborn, including minor variations. Congenital malformations of the central nervous system in Scotland. Spray adhesives, birth defects, and chromosomal damage. Environment and birth defects. Epidemiologic aspects of the problem of congenital malformations. Etiology and pathogenesis of congenital cleft lip and cleft palate, an NIDR state of the art report. Fetal malformations due to thalidomide. Hormonal pregnancy tests and congenital malformation. Hormonal pregnancy tests and congenital malformations. How can the teratogenic action of a factor be established in Man? Infant mortality from spina bifida, congenital hydrocephalus, monstrosity, and congenital diseases of the cardiovascular system in England and Wales. Major central nervous system malformations in South Wales. Incidence, local variations and geographical factors. Pregnancy factors, seasonal variation and social class effect. Malformations in a population observed for five years after birth. Metropolitan Atlanta congenital defects program. Monitoring, Birth Defects and Environment: The Problem of Surveillance. No association of emotional stress or vitamin supplement during pregnancy to cleft lip or palate in man. Prospective versus retrospective approach in the search for environmental causes of malformations. Recognizable Patterns of Human Malformation. Major Problems in The further epidemiological differentiation of cleft lip and palate: The incidence of congenital malformations: A study of 5, pregnancies. Trends in surveillance of congenital malformations. New Directions in Research. Unrecognized epidemic of anencephaly and spina bifida. Validity of characterization of urban places according to reported congenital malformation rates.

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Chapter 4 : Q&A: Abortions for fetal abnormality

ANENCEPHALUS, SPINA BIFIDA, AND HYDROCEPHALUS incidence of hydramnios with birth order, which suggests that hydramnios is not the explanation of theearlier.

What is the law regarding abortions for fetal abnormality? Antenatal recognition of fetal malformations relies on accurate detection from screening programmes using either maternal serum screening, routine ultrasound scanning or a combination of both. The early pregnancy scan between 10 and 14 weeks is undertaken to determine the gestational age of the fetus, if the mother is expecting a multiple birth and the risk of Down syndrome. Structural abnormalities are picked up during the second ultrasound scan which occurs between 18 and 20 weeks. Some fetal malformations become clearer after 24 weeks. Examples of these are the Hypoplastic Left Heart Syndrome and cerebral ventriculomegaly. As noted in a Europe-wide study, the antenatal detection rate was highest for anencephalus. Work on this will commence shortly. For this reason, it is important that doctors provide a good assessment and a decision is made based on the consultation with the doctor. A study conducted in Pakistan revealed that the most common malformation for such children was congenital heart disease. It is important for parents to be provided with good information on the future quality of life for their babies and family. Richmond S, Atkins J. A population-based study of the prenatal diagnosis of congenital malformations over 16 years. Garne E, et al. Prenatal diagnosis of severe structural congenital malformations in Europe. Birth prevalence of cleft lip and palate in Northern Ireland to Cleft Palate-Craniofacial Journal ; Congenital anomaly notifications , England and Wales. Health Statistics Quarterly Spring ; Congenital heart disease and associated malformations in children with cleft lip and palate in Pakistan. British Journal of Plastic Surgery ; Central nervous malformations in presence of clefts reflect developmental interplay. Associated malformations in cases with oral clefts. Epidemiology of cleft palate alone and cleft palate with accompanying defects. European Journal of Epidemiology ; An epidemiologic study of orofacial clefts with other birth defects in Victoria, Australia. Associated malformations in infants with cleft lip and palate: Elsewhere on the site.

Chapter 5 : Techniques for assessing teratogenic effects: epidemiology. - CORE

1. Author(s): Rogers,S C; Weatherall,Josephine A C Title(s): Anencephalus, spina bifida and congenital hydrocephalus, England and Wales / S. C. Rogers, J. A.

Chapter 6 : Patau syndrome | Revolvy

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ANENCEPHALUS, SPINA BIFIDA, AND HYDROCEPHALUS For the congenital malformations, as for many these authors' results to areas other than England and Wales.

Chapter 9 : Epidemiology of stillbirths from congenital abnormalities in England and Wales,

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