LETTERS TO THE EDITOR

Drug induced VATER association: is dibenzepin a possible cause?

The VATER complex is a non-random combination of three or more vertebral, anorectal, tracheo-oesophageal, radial, and renal anomalies of a co-occurrence nature. In cardiac abnormalities are sometimes also involved (VACTERL). It has not been recognised as a specific syndrome and its components are viewed repeated etiologies in their population based study, emphasised the aetiological heterogeneity of this entity. Exposure to proges- togen or oestrogen or both in the first trimester of pregnancy has been suggested as a possible cause and, recently, lead intoxication and lovastatin administration have also been implicated. We describe the first case of VACTERL association in a neonate whose mother had been treated with dibenzepin.

The proband is the second child of a 25 year old woman who suffered from depression and had been treated with dibenzepin (Victoriil) (80 mg x 3/day) throughout her pregnancy. The mother did not smoke or drink alcohol or coffee, and had not suffered from any infection during pregnancy. The patients are not related, and the father is healthy. The family history was unremarkable. The mother had not taken any medica- tion during her first pregnancy and gave birth to a normal live male child. During the second pregnancy (1992), ultra- sound and zetoprotein measurements were not performed. The fetal heart rate near term showed normal values.

The 40 week term infant, born in February 1993, with a weight appropriate for gesta- tional age (3260 g), presented with oesogas- phageal atresia,1,3 karyotypic anomalies,2 rhizomelic limb shortening with short, broad, and bowed long bones, the femora being most severely affected. There is meta- physeal flaring and irregularity in infancy, causing prominent joints with restricted mobility. The ribs are short and flared. Variable features include micrognathia, midface hypoplasia, long philtrum, facial haemangioma, abnormal rib number, mild platyspondyly, increased acetalbar angles, and skin dimples over bony prominences.

To date no cases of kypohomal dysplasia have been reported.

The subject, a male child, remained mildly dysmorphic at the age of 3 years 3 months. Rhizomelic shortening of the limbs with anterior bowing of the thighs was still evident, but had improved since infancy. His gait was abnormal with a tendency to toe walk on the left and he began to complain of intermittent lower back pain. A radiograph showed flattening and fragmentation of the right capital femoral epiphysis (figure) consistent with Perthes disease.

Chances in the hip which are similar to those in Perthes disease but always symmet- ric are found in certain skeletal dys- plasias and malformation syndromes, such as multiple epiphyseal dysplasia, mucopolysac-

4 Levine F, Muenke M. VACTERL association with high previous lead exposure; similarity to animal models of lead teratogenicity. Pedia- trics 1991;87:590-2.

Kypohomal dysplasia

A six year follow up of a child with kypohomal dysplasia has shown some interesting and previously undescribed features. This case was originally described by Temple et al.

Kypohomal dysplasia is a rare condition with a generalised abnormality of skeletal growth. It is characterised by rhizomelic and mesomelic limb shortening with short, broad, and bowed long bones, the femora being most severely affected. There is meta- physeal flaring and irregularity in infancy, causing prominent joints with restricted mobility. The ribs are short and flared. Variable features include micrognathia, midface hypoplasia, long philtrum, facial haemangioma, abnormal rib number, mild platyspondyly, increased acetalbar angles, and skin dimples over bony prominences.

Velocardiofacial syndrome and DiGeorge sequence

I was interested to read the series of articles published in the Journal of Medical Genetics3-7 regarding phenotypic overlap of the velocar- diofacial syndrome and DiGeorge sequence, preceded by Judith Hall's Editorial advocating the use of the acronym CATCH 22, as suggested by Wilson et al.8 The attention to velocardiofacial syndrome and 22q11 is very exciting and the authors are to be congratu- lated on this collective body of material.

I would like to respond to a number of points which, in my opinion, point out the important role of clinical diagnosis, even in conditions where molecular analysis has yielded positive results. The articles of Greenberg,2 Goldmuntz et al,9 Driscoll et al,4

The patient aged 3 years 6 months. There is bowing of both femoral shafts with flexion deformities at the knees. The capital femoral epiphysis on the right appears collapsed and fragmented. The metaphyses are flared.

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DiGeorge syndrome is a genetic condition characterized by a deletion of genetic material from chromosome 22. This deletion can lead to various physical and developmental abnormalities. Patients with DiGeorge syndrome may have a range of symptoms, including cardiovascular defects, immune deficiencies, and learning disabilities. The syndrome is often diagnosed through a combination of clinical symptoms and genetic testing.

The prevalence of DiGeorge syndrome is estimated to be around 1 in 10,000 births, although this number can vary depending on the specific diagnostic criteria used. The deletion is usually inherited in an autosomal dominant manner, meaning that if one parent has the condition, there is a 50% chance that a child will inherit it.

DiGeorge syndrome is often associated with other genetic disorders, such as velocardiofacial syndrome (VCFS) and chromosome 18 deletion syndrome. These conditions share some common features, such as heart defects and immune deficiencies. Research suggests that there may be some overlap in the clinical manifestations of these conditions, although the underlying genetic mechanisms differ.

The syndrome is typically diagnosed in infancy or early childhood, and treatment may involve a multidisciplinary approach, including cardiology, genetics, and immunology. Early intervention can help to mitigate some of the complications associated with the condition.

Multiple origins of X chromosome tetrasomy

Multiple origins of X chromosome tetrasomy refer to the presence of an extra X chromosome in some individuals, which can lead to various physical and developmental abnormalities. The condition is usually inherited in an autosomal dominant manner, and the presence of the extra chromosome can be due to different mechanisms, such as errors during meiosis or mitosis.

The extra X chromosome is often associated with learning disabilities, behavioral problems, and other neurological issues. The genetic complexity of X chromosome tetrasomy can make it challenging to understand the full spectrum of symptoms and to develop effective treatment strategies.

Researchers continue to investigate the biological and molecular mechanisms underlying X chromosome tetrasomy. Understanding these mechanisms is crucial for improving diagnostic tools and developing targeted interventions for affected individuals.