



Dandy-Walker-Like Malformation with Atrioventricular Septal Defect

Alternative Names

Craniocerebellocardiac Dysplasia
3@C Syndrome
Ritscher-Schinzel Syndrome
CCC Dysplasia

WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

OMIM Number

220210

Mode of Inheritance

Autosomal recessive

Description

Craniocerebellocardiac syndrome (3C syndrome) was first reported in 1987 by Ritscher and Schinzel. It is characterized by the association of cranial dysmorphism, cerebellar hypoplasia and cardiac malformation. Cranial dysmorphism features include relative macrocephaly, bulging forehead, prominent occiput, large anterior fontanel, ocular hypertelorism, depressed nasal bridge, down slanting palpebral fissures, cleft palate and bifid uvula. Cardiac malformations include defects of the endocardial cushion, and conotruncal defects. Additional anomalies include colobomas, hand abnormalities, and mental retardation. Hypotonia, retarded psychomotor and speech should be improved by special help. Almost, cardiac defects are responsible for the prognosis of 3C syndrome.

Molecular Genetics

The craniocerebellocardiac syndrome is inherited as an autosomal recessive pattern. The associated gene is unknown to date.

Epidemiology in the Arab World

Lebanon

Megarbane and Haddad (1999) reported two Lebanese siblings with cranio-cerebello-cardiac syndrome (3C syndrome) born to non-consanguineous parents. One of the sibs was 5-year-6-month old boy and the other was 2-year-3-month old girl. The boy was the product of an uneventful pregnancy. His psychomotor development was delayed and he could not talk. He had prominent forehead, a flat occiput, hypertelorism, slightly low posteriorly rotated ears, a wide flat nasal bridge, a high-arched palate, hypoplastic nipples, a left undescended testicle, and muscular hypotonia. MRI of the boy's brain showed bilateral small cerebral hemispheres, vermis hypoplasia, a large cisterna magna, and slight enlargement of the lateral ventricles. His chromosome karyotype was normal (46, XY). The girl was operated for a duodenal stenosis and for a secundum atrial septal defect when she was a neonate. Her clinical course and physical features were quite similar to that of her brother, except that she had a mild ptosis of the eyelids. She had the same MRI brain scan of her brother, but without ventricles enlargement. Her karyotype was normal (46, XX). Both sibs were mentally retarded. In a global review of 23 patients with 3C syndrome who were studied previously, Megarbane and Haddad (1999) only noted five patients with abdominal malformation. However, the two patients from Lebanon were the first to have duodenal stenosis associated with the 3C syndrome which might extend the wide range of phenotypic variability of this syndrome.

References

Megarbane A, Haddad J. Does the cranio-cerebello-cardiac syndrome (3C syndrome)



include abdominal malformations? Clin
Dysmorphol. 1999; 8(4):305-7.

Contributors
Abeer Fareed: 19.6.2006

