Branchio-oculo-facial Syndrome with Valvular Pulmonic Stenosis

Branchio-oculo-facial syndrome (BOFS) is a rare autosomal dominant disorder with highly variable expression. It is characterized by craniofacial, cervical, auricular, ophthalmologic and oral anomalies. Extracraniofacial malformations are uncommon. Atrial Septal defect and Tetralogy of Fallot's have been described in association with BOFS(1,2). Neuro-development is normal; cases of moderate to severe retardation with hypotonia, visual, hearing and speech problems have been reported. The molecular basis of the disorder has not been elucidated.

This term neonate was born to parents with one previously normal child. The father and three paternal uncles had premature graying of hair at about 21 years and two had preauricular pits. At one year of age she was noticed to have an unusual facial appearance with a sloping forehead, midfacial clefting, left upslanted palpebral fissure, hypertelorism, microphthalmia (Left >Right), nose with broad bridge, posteriorly angulated and low set ears with preauricular pits. Discharge was present in the left eye with hypertrophy of the skin over the ducts suggesting chronic dacrocystitis. There was a strip of hemangiomatous skin in the posterior auricular area. The palate was high arched.

The nipples were normally spaced. There was a grade II/VI systolic murmur heard at the pulmonic area. An umbilical hernia was present. Left postaxial polydactyly, bilateral single transverse palmar creases were present with absent interphalangeal creases on both thumbs. She had syndactyly of the left 4th and 5th toes. Dentition was normal and hair were sparse.

Ultrasound abdomen revealed normal kidneys. Echocardiography demonstrated valvular pulmonic stenosis with no evidence of chamber hypertrophy. Puretone audiometry revealed severe sensorineural hearing loss. Karyotype was normal.

BOFS is recognized to be a neurocristopathy. Aberrant development in the area of the nasal maxillary groove leads to atresia or hypoplasia of the nasolacrimal duct(3). The craniofacial appearance consists of dolicocephaly, sparse hair, high forehead, ocular asymmetry, upslanting palpebral fissures, hypertelorism and a wide nasal bridge with a flat tip and midfacial clefting. The auricles are low set, posteriorly rotated with a thin helix. Distortion of the upper lip due to hypertrophy of the lateral pillars of the philthrum named as 'pseudocleft' is seen in about 54% cases(4).

The post auricular area had an overlying hemangiomatous skin, which had subsided since birth. This abnormal skin with or without sinus or fistulae is the pathognomonic malformation of the BOF syndrome. Other features are oligodontia, sparse hair and premature graying of hair. Skeletal defects are rare, those described being fifth finger clinodactyly, polydactyly and single palmar creases.

Severe brain malformations are rare, but cerebellar agenesis and mild hydrocephalous have been described. Pulmonic stenosis has not been described so far. Branchio-oto-renal syndrome has a considerable overlap with BOFS. The BOR gene has been mapped and named EYA 1 gene. The known genes of the EYA family are not involved in BOFS(5). Pediatricians must be aware of this syndrome as this is an autosomal dominant disorder and carries a 50% risk of recurrence, hence genetic counseling is important.

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Mumps-Need for Urgent Action

The epidemiology of mumps in India and the magnitude of the problem are still not fully appreciated, as pointed out by Dr. Jacob John(l). Mumps continues to occur in epidemic proportions, despite the availability of an effective vaccine. We present the data on mumps cases admitted to the Institute of Maternal and Child Health, Calicut, (North Kerala) during the five-year period from 1999 to 2003. Compilation of data was done as part of monitoring of epidemic and infectious diseases. Data sheets were filled every week and records maintained. This was done as part of the Prevention of Epidemic and Infectious Disease initiative. Children with mumps are usually admitted only when complications occur. Although the data does not give exact estimate of the disease burden in the community, it is a reflection of the magnitude of the problem. The diagnosis of mumps was made clinically on the basis of the presence of acute parotitis, unilateral or bilateral(l) .

There were 301 children admitted with mumps between 1999 and 2003. After a declining trend from 1999 to 2001 the number of cases increased. There were 92 admissions in 1999, 79 in 2000, 20 in 2001 and 55 in both 2002 and 2003.

The male female ratio was 2.2:1, with 208 boys (69%) and 93 (31%) girls. Majority of cases (58%) were in the 5-9 year age group. 85 children (28%) were in the 1-4 year age group and 39 (13%) were in the 10-12 year age group. There were two children below the age