

Oculo-facio-cardio Dental Syndrome

Oculofaciocardiodental syndrome (OFCD) is a rare genetic disorder affecting ocular, facial, dental and cardiac systems [1]. The genetic analysis of OFCD patients revealed mutations in the *BCOR* (BCL-6 interacting co repressor) gene on chromosome Xp11.4 [1]. It has been suggested that OFCD is an X-linked dominant trait [2], and embryonic lethality for males [1]. Till date, very few cases have been reported worldwide [3]. We present a female child diagnosed with minor features of OFCD syndrome, suggesting that OFCD syndrome may be mild or underdiagnosed.

A 3-yr-old girl presented with complaints of fever, cough and cold with severe anemia. She was born out of a non-consanguineous marriage with no history of abortions or stillbirths in the mother. Facial features suggested a long and narrow face, micrognathia, long philtrum, retrognathia, and a pointed nose. On oral examination, she had a high arched palate, maloccluded and malaligned teeth, radiculomegaly and delayed dentition. Ophthalmic examination was suggestive of microphthalmia and a congenital cataract in right eye. Chest X-ray was suggestive of cardiac enlargement and 2-D echocardiography confirmed patent ductus arteriosus. The girl had a nasal twang of voice suggesting possibility of velopharyngeal insufficiency. She had a short stature (weight-for-height $<-3SD$), but the developmental milestones were appropriate for age with no intellectual disability. None of the family member had any similar faciocardial features.

Although, these features (in isolation) may be seen in a variety of other conditions, OFCD is one of the few rare craniofacial or dental disorders where the genetic cause is known and the mode of action of the mutant gene is fairly well studied [1]. The phenotypic and genotypic



Fig. 1: Long and narrow face, cataract and microphthalmia in right eye, long philtrum, pointed nose and retrognathia in oculo-facio-cardio dental syndrome.

confirmation may lead to early identification, and may thus prevent fatal outcomes.

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