Median Cleft Face Syndrome

MEDIAN CLEFT FACE SENDROMU

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**SUMMARY**

In this article we report a female individual, born in 1978, with median cleft face syndrome. Growth and developmental milestones were normal except reconstructive surgery at 3 years of age and pulmonary tuberculosis at 7 years of age. Physical examination revealed no associated abnormality as laboratory findings and 46, xx karyotype. Genetic analysis suggests autosomal recessive inheritance.

**Key Words:** Median cleft, face syndrome

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**CASE REPORT**

Proposita (Figure 1-3) GB, born in Ankara in 1978, has been referred to our services for genetic counseling because of her facial dysmorphism at 12 years of age. She was born through spontaneous vaginal delivery followed a normal pregnancy and enlarged forehead, elongated nasal bridge together with cleft lip and palate have been noted. She was otherwise healthy and her development has almost been normal with some problems of nursing. Reconstructive surgery has been applied at 3 years of age. No significant interference has been noticed during her growth and development, except long-lasting drug treatment for pulmonary tuberculosis at 7 years of age. She attends a normal preliminary school with a good achievement and adoption.

The height was 137 cm and weight 32.5 kg. The forehead was enlarged. The frontal hairline course was clockwise on the left and counter-clockwise on the right. Eyebrows were bowed and a mild right strabismus was noted. The distance between the median canthi was very long and asymmetrical (more pronounced to the left), forming a mild epicanthial fold on the left. The broad nasal bridge proceeds down till the nostrils. Nostrils were asymmetric too, the left one being larger. The filtrum was long and large. There was a surgical scar on the left side. Her mouth resumes a fishlike appearance when the lips are apposed, with a hypoplastic chin. Beans and another surgical scar from hard palate to the nose were observed inside the mouth. Ears being normal were

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slightly asymmetric in localization. Still another scar was seen anterior to the right external acoustic meatus probably after a dermoid cyst has been removed. While the neck appeared normal, inferior to her left breast was a large (3 x 6 cm) hyperpigmented area, and a few cafe-au-lait spots on the back. Slight joint laxity was the only systemic finding. All laboratory tests, including karyotyping, were reported to be within normal limits. Her mother was 30, father 32 years of age and both were normal and healthy, and first-cousins. The first child of the family was male, born at 8th months of gestation, deceased within few hours of birth. No autopsy has been performed but he was described normal in appearance. G was the second child and after her, mother has given birth to another healthy baby now 8 years old. Family history includes a first trimester abortion. As it can be seen on the pedigree chart no any other members of the family has facial cleft.

With these findings, the case presented here was accepted an example of median cleft face syndrome, autosomal recessive in nature.

**DISCUSSION**

Different types of abnormalities are given under the topic of median cleft. Clinical presentation, as mentioned earlier, may vary from a simple notch on lip and/or palate and chin to the much larger and deep clefts and in simple cases only soft tissues are involved whereas bones are intact (Midion M. Child Zonga, ANP Joseph K. Shija, Baraister Mi, Breath, Waite and Watson 1949) (3-5).

**Figure 1.** Pedigree chart of B family.

**Figure 2-3.** The proband at 12 years showing the present physical findings and repair of cleft.
Genetic heterogeneity may also be present. De Myer (1967) (11) called the entity median cleft face syndrome whereas Sedano (1970) (2) frontonasal dysplasia. Despite monogenic transmission models, some proposed multifactorial transmission theory (Fogh, Anderson, 1942, Fraser 1963 and 1970, Shields et al. 1981, Demenais et al. 1984, Mc Kusik 1988) (6-11). This case is apparently of rare occurrence contrary to the high frequency of all cases of clefting, if it is considered a single entity. We couldn’t find any report from our country. Although this case is sporadic, parental consanguinity and the absence of anomalies among other relatives suggests autosomal recessive etiology. Eiberg et al (12) suggested in 1987 a major locus on 6th chromosome with multiple alleles playing role in the formation of orofacial clefts.

REFERENCES