Median Cleft Face Syndrome

MEDIAN CLEFT FACE SENDROMU

Mehmet Y. MECDEL*, Bekir Sıtkı ŞAYLI, Peruze S. UÇAN*****

* Ankara University, School of Medicine, Department of Pediatric Surgery,
** Ankara University, School of Medicine, Department of Medical Biology and Genetics,
*** Ankara University, School of Medicine (Student)

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 suggestes autosomal recessive inheritance.

Key Words: Median cleft, face syndrome

ÖZET

Bu makale ile, 1978 doğumlu bir kızda "median cleft face" (orta çizgi yarıklı yüz veya orta çizgi yüz yarığı) sendromu sunulmaktadır. Üç yaşlarında geçirilen reperatif operasyon ve 7 yaşlarında geçirilen akciğer tüberkülozu dışında hem gelişme öyküsü hem öteki fizik muayene bulguları normal değerlendirilmiştir. Genetik analiz otozomal resesif etiyolojiyi düşündürür nitelikte bulunmuştur.

Anahtar Kelimeler: Median cleft, face sendromu

Turkiye Klinikleri J Med Sci 1996, 16:245-247

Median cleft face syndrome (1967) (1) is an interesting example of dysmorphic entities involving the face (Fronto-nasal dysplasia, 1970) (2). It is characterized by cranium bifidum. Mental retardation especially in thos with other system disorders may be apparent as well. Clinical presentation varies from a simple notch to the full cleft. No genetic basis has yet been proved, and almost all cases reported so far are sporadic. We suggest autosomal recessive inheritance while presenting an example with parental consanguinity, although this pattern has already been proposed.

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 healty 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 epicanthal 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

Geliş Tarihi: 14.12.1995

Yazışma Adresi: Dr. Mehmet Y. MECDEL
Ankara University, School of Medicine,
Department of Pediatric Surgery

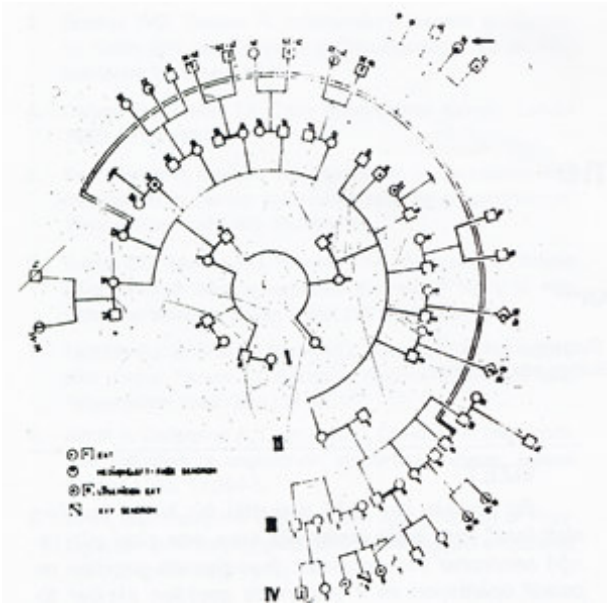

Figure 1. Pedigree chart of B family.

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 cafeau-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).

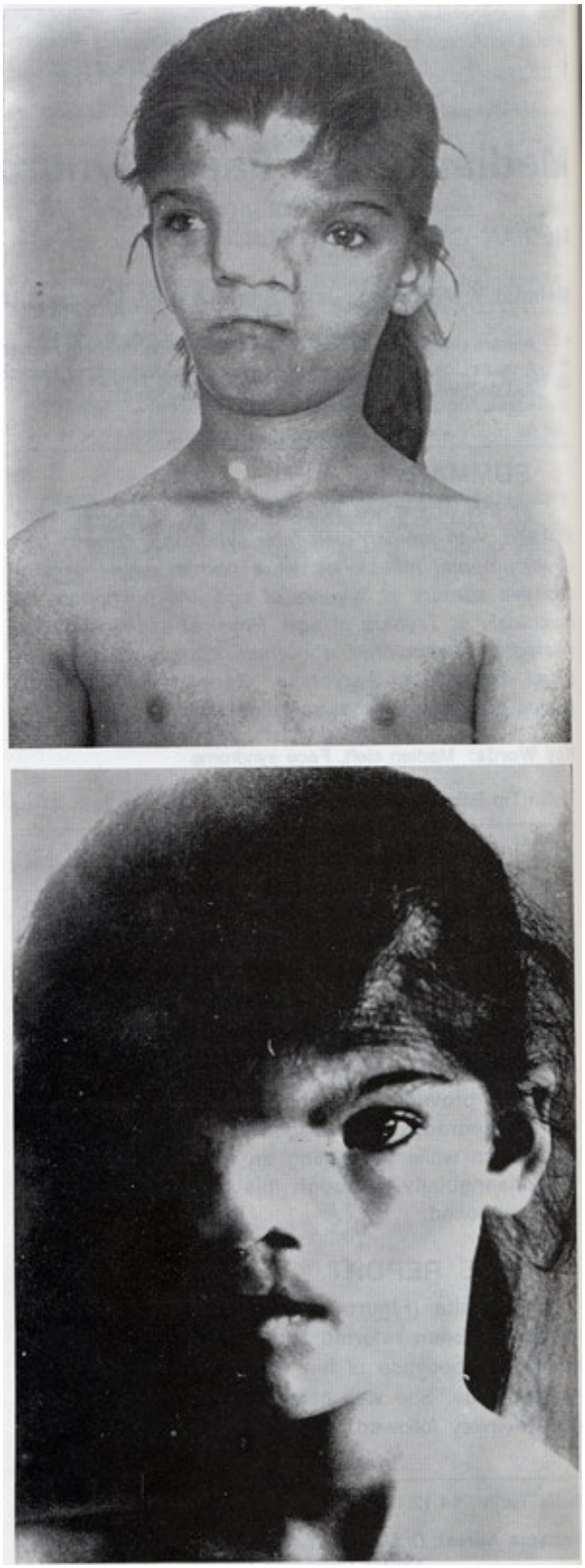

Figure 2-3. The proband at 12 years showing the present physical findings and repair of cleft.

Genetic heterogenity 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

1. De Myer W. The median cleft face syndrome. Neurology 1967;17:961-71.
2. Sedano HO, et al. Frontonasal dysplasia. J Pediapr 1970;76:906-13.
3. Midion M, Chidzonga. AND Joseph K Shija. Congenital median cleft of the lower lip, bifid tongue with ankyloglossia, cleft plate, and submental epidermoid cyst: Report of a case. J Oral Maxillotac Surg 1988;6:809-12.
4. Baraister ML. The hypertelorism microtia clefting syndrome. Med Genet 1982;19:387-8.
5. Braith Waite F, Watson J. A report of three unusual clefts lips. Br J Plast Surg 1949;2:38.
6. Fogh Anderson P, ed. Inheritance of harelip and cleft palate, Copenhagen: Busck, 1942.
7. Fraser FC. Hereditary disorders of the nose and mouth. In Proc 2nd Int Cong Hum Genet 1963;3:1852-5.
8. Fraser FC. The genetics of cleft lip and cleft palate. Am J Hum Genet 1970;22:336-52.
9. Shields ED, Bixler D, Fogh Anderson P. Cleft palate: A genetic and epidemiologic investigation. Clin Genet 1981;20:13-24.
10. Demenais F, Bonaiti Pellie C, Braid ML, Feingold J. An epidemiological and genetic study of facial clefting in France. II Segregation analysis. J Medical Genetics 1984;21:436-40.
11. Mc Kusick VA, ed. Mendelian inheritance in man. Catalogs of autosomal dominant, autosomal recessive, and X-linked pheno types, 7th ed. Baltimore: Johns Hopkins University Press, 1986.
12. Eiberg H, Bixler D, Neilsen LS, Conneally PM, Mohr J. Suggestion of linkage of a major locus for nonsyndromic orofacial cleft with FI3A and tentative assignment to chromosome 6. Clinical Genetics 1987;32:129-32.