DYSCEPHALIA MANDIBULO-OCULO-FACIALIS

Hallermann-Streiff Syndrome: A Case Report

Nilüfer Eke, M.D.*

Dyscephalia mandibulo-oculo-facialis was first described by Hallermann in 1948 and Streiff in 1950; in 1958, François collected the first 25 cases from the literature and reported them under the name of a new syndrome. The total number of cases found in the literature is 47.[1]

The syndrome comprises skull abnormality, facial malformations, dental abnormalities, localised hypotrichosis, congenital abnormalities of the eyes, dwarfism, and mental and motor retardation. Craniofacial dysplasia gives these patients a characteristic «bird face» profile. The ocular abnormalities are bilateral microphthalmia (enophthalmus), blue sclera, congenital bilateral cataract, ocular nistagmus, strabismus, glaucoma and microcornea. In these patients no abnormalities of the nails have been reported.[1] In most of the cases, family history is not contributory. The male and female ratio is equal.

The purpose of this paper is to describe such a case as a first report from Turkey.

Case Report

E.Y. (67 - 30929), a five-year-old girl, was admitted to Hacettepe Children's Hospital with growth retardation and loss of vision. Her parents stated that she had stood up when she was four years old and said her first word at the same age, but she had not yet walked or talked. Her history revealed that her mother's pregnancy and delivery were normal, that she had five healthy siblings and that her parents were first cousins.

Physical examination : The patient's weight was 8.9 kg, height 84.5 cm, head circumference 46.5 cm and chest circumference 46.5 cm. The anterior and posterior fontanels were closed. An eye examination showed blue sclera, enophthalmus and bilateral congenital cataract (Figure 1). Her nose was small, thin and beaked and she had micrognathia with a high-arched palate (Figure 2). No skin, hair, eyelash or dental abnormalities were observed. The joints were hyperextensible. Her eyes were only able to perceive light and she appeared mentally retarded.

From Hacettepe University Faculty of Medicine, Department of Pediatrics, Ankara.

* Resident in Pediatrics.

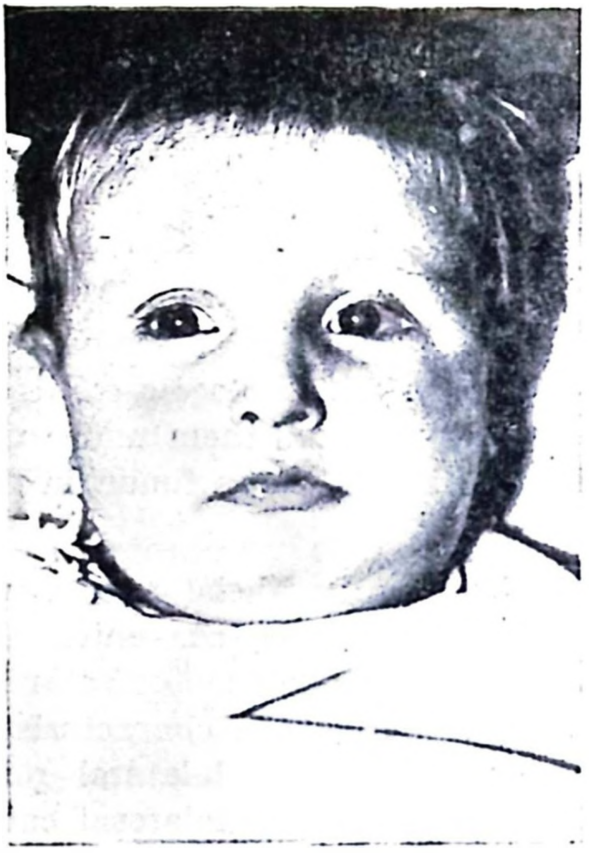

Figure 1. Bilateral congenital cataracts.

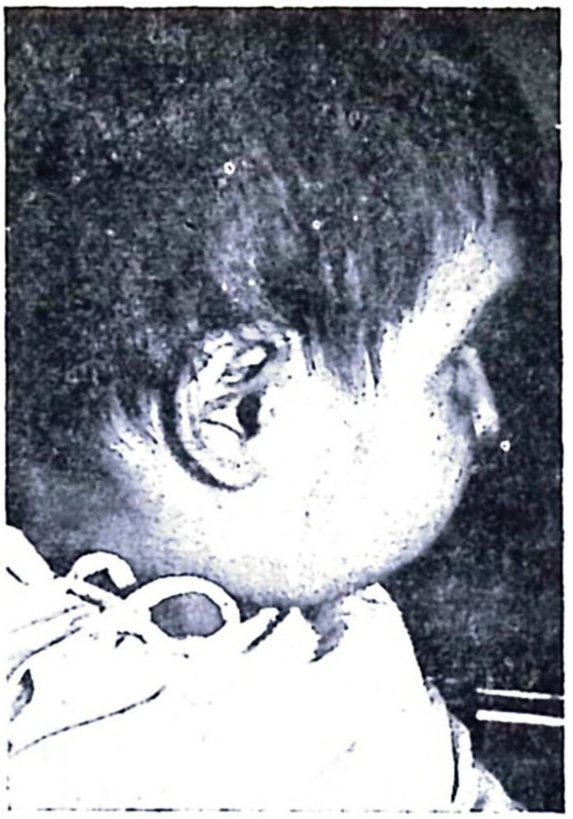

Figure 2. «Bird Face» profile.

Laboratory examination : Hb was 8.30 gm/100 ml, W.B.C. 6400 per mm^3, a blood smear showed hypochromia, anisocytosis, poikilocytosis and basophilic stippling. Total proteins were 7 gm/100 ml, albumin 4.5 gm/100 ml, globulin 2.5 gm/100 ml, blood sugar 73 mg/100 ml, serum calcium 10.5 mg/100 ml, serum phosphorus 4.5 mg/100 ml, serum alkaline phosphatase 5.2 Bodansky units and cholesterol 206 mg/100 ml. The serologic test for syphilis (VDRL) was negative, serum iron was 108 gamma/100 ml, serum iron binding capacity was 417 gamma/100 ml and PBI was 2.56 gamma/100 ml. The sex chromatin was positive in a buccal smear and chromosome analysis was normal (46/XX). Total amino acids in the urine were within normal limits (48 Taurin units). The cerebrospinal fluid analysis was normal.

E.E.G. showed slight disseminated dysrythmia. The patient's bone age was revealed to be two years by wrist x-rays. Although radiography of the long bones showed generalised osteoporosis, the mandibulae and temporo-mandibular joints were normal. Pneumoencephalography showed the presence of cortical atrophy and in the first chest x-ray there were bilateral bronchopneumonic infiltrations.

Her infection was treated with antibiotics. Bilateral cataract extraction was performed and the patient was discharged from the hospital.

Comments

The symptoms in the present case are typical of dyscephalia oculo-mandibulo-facialis with abnormalities of the skull and face, bilateral congenital cataract, dwarfism, and mental and motor retardation.

Although localised hypotrichosis, skin atrophy and dental abnormality have been described frequently in the literature, our patient had none of these. She had no history of rubella syndrome. Tests for galactosemia and congenital syphilis were negative.

Previous authors found the chromosome analysis normal as it was in our case.[2, 3, 4, 5] Our patient's PBI was low and the bone age was retarded. This may be due to growth retardation. Pneumoencephalography was not performed in the reported cases of the literature; however, because we found cortical atrophy, we suggest that in such patients pneumoencephalography be done and the presence of cortical atrophy investigated.

Falls and Schull reported a patient with neurofibromatosis, duplication anomaly of the urogenital system and deafness along with other anomalies.[6] François and Hoefnagel described degenerative retinal disease (chorioretinal atrophy).[5]

At first glance, dyscephalia mandibulo-oculo-facialis is commonly confused with progeria[7] (Hutchinson-Gilford syndrome), but in the latter the presence of a generalised atrophy of the skin and muscles, arthritis, early arteriosclerotic changes and the absence of cataracts are some of the points of difference.

Hallermann-Streiff syndrome and oculodentodigital syndrome appear very similar. If cataracts are not present, roentgen features are most helpful in the differentiation of the these two syndromes. The oculodentodigital syndrome shows a broad mandible, broad long bones and hypoplasia of one or more middle phalanges of the toes; the Hallermann-Streiff syndrome shows hypoplasia of the rami of the mandible, natal teeth, thin bones of the calvarium and very slender long bones.[1, 2]

The face of patients with Hallermann-Streiff syndrome resembles the one that is seen in Cockayne's syndrome, but in the latter deafness, ataxia and light sensitivity of the skin are the differentiating findings.

Above all, Hallerman-Streiff has such characteristic features that if it is seen once, the diagnosis will not be a problem.

Summary

A case of dyscephalia mandibulo-oculo-facialis is presented. The patient had the characteristic bird face, microcephaly, mental and motor retardation and congenital bilateral cataract. Chromosome analysis revealed no anomalies. She had cortical atrophy proven by pneumoencephalography.

REFERENCES

1. Gellis, S. S.: Dyscephaly (Hallermann-Streiff-François syndrome), in Year Book of Pediatrics, Chicago, Year Book Medical Publishers, 1966-1967, p. 493.
2. Kurlander, G. J., Lavy, N. W. and Campbell, J. A.: Roentgen differentiation of the oculodentodigital syndrome and Hallermann-Streiff syndrome in infancy, Radiology 86: 77, 1966.
3. Lamy, M., Jammet, M. L., Maroteaux, P. and Ajjan, N.: [Dyscephaly, Hallermann-Streiff-François syndrome], Arch. Franç. Pédiat. 22: 928, 1965.
4. Forsius, H. and De la Chapelle, A.: Dyscephalia oculomandibulo-facialis; two cases in which the chromosomes were studied, Ann. Paediat. Fenn. 10: 280, 1964.
5. Hoefnagel, D. and Benirschke, K.: Dyscephalia mandibulo-oculo-facialis (Hallermann-Streiff syndrome), Arch. Dis. Child. 40: 57, 1965.
6. Falls, H. F. and Schull, W. J.: Hallermann-Streiff syndrome, a dyscephaly with congenital cataracts and hypotrichosis, A.M.A. Arch. Ophthal. 63: 409, 1960.
7. Moehlig, R. C.: Progeria with nanism and congenital cataracts in a five year old child, J.A.M.A. 132: 640, 1946.