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Hallermann-Streiff Syndrome*

(Dyscephalia Mandibulo-Oculo-Facialis)

Sevim Balcı, M.D. ** / Burhan Say, M.D. ***

Although this syndrome was first reported by Aubry[1] in 1893, a more detailed description was made by Hallermann[2] in 1948 and Streiff[3] in 1950. Since then over 60 cases of the Hallermann-Streiff Syndrome have been reported from different parts of the world including Turkey.[4-16]

The classical picture presents bilateral congenital cataract, dyscephalia with mandibulo facial malformation, microphthalmia, mental retardation, cutaneous atrophy, localized alopecia, beaked nose and nanism. Ponte in 1962 reviewed the findings in reported cases and found only the first three as constant features but many reported cases even do not show all of these manifestations.[4] More recently detailed descriptions of the radiological changes of this condition have been reported.[9]

The purpose of this communication is to report a typical case with characteristic radiological findings and cytologic and dermatoglyphic studies.

Case Report

N. S. was a 4-year-old girl with chief complaints of mental and motor retardation. The history revealed that she was the product of a full-term pregnancy, complicated with bleeding during the fourth month. She had neonatal jaundice associated with fever and convultions on the fifth day after birth. When she was one month old she had further

* From the Department of Pediatrics, Hacettepe University, Faculty of Medicine, Ankara-Turkey.
** Specialist in Pediatrics and Fellow in the Department of Genetics
*** Professor of Pediatrics

convulsions but at that time fever was not observed. These attacks continued until one and a half years of age. After that the attacks decreased but her body became stiff.

Family History : This is the seventh child of the family. The other 6 children are in good health. There was no consanguinity between the parents.

Physical Examination : The patient's weight was 10 kgr., height 85 cm. and head circumference 43 cm. (all below the third percentile). The head was flattened posteriorly. Her face had a bird-like appearance with a beakshaped nose and hypertelorism. Both eyes were microphthalmic and showed cataracts (Figures 1 a, b). The mouth was small and the palate had a high arch. Teeth showed irreguler dentition. The neck and sternum were short. There was sparse hair on the anterior part of the head. The tongue was very small and it could not be moved easily. She had flexion contractures of the extremities as well as generalized muscular spasticity (Figure 2). There was severe micrognathia with marked limitation of movements in both temporomandibular joints. Dermatoglyphic studies revealed six whorls and 4 ulnar loops

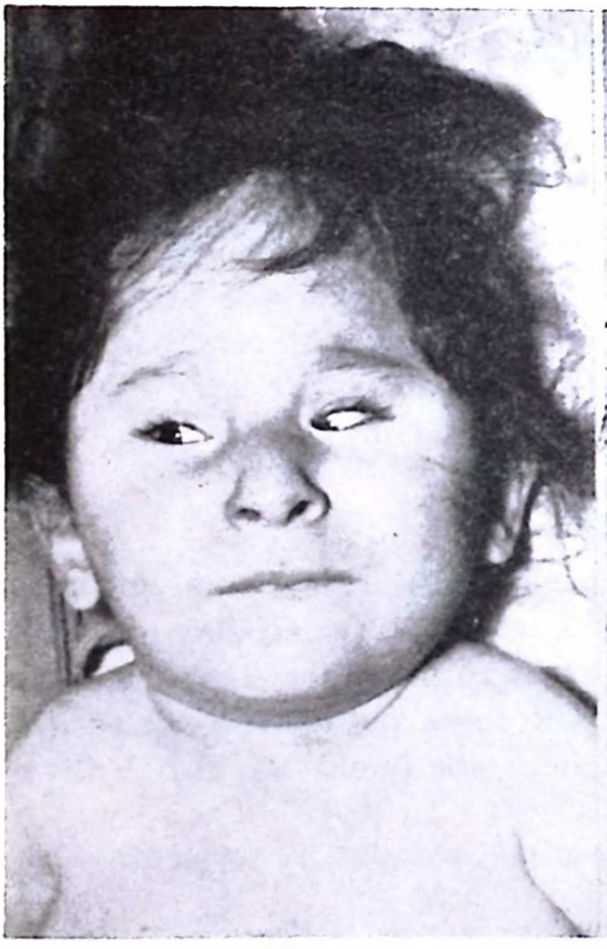

Figure 1 a

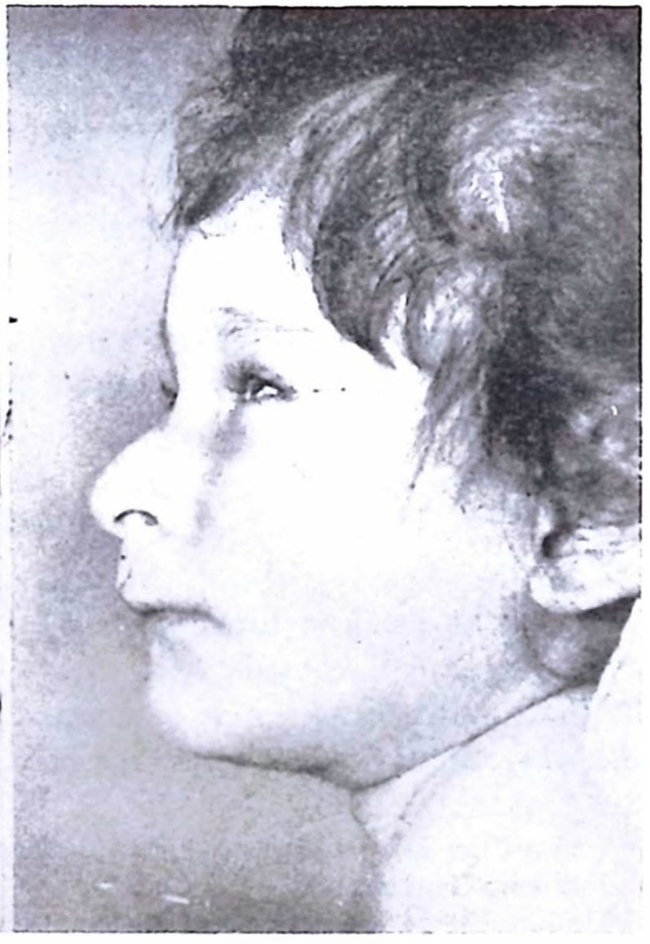

Figure 1 b

Figure 2

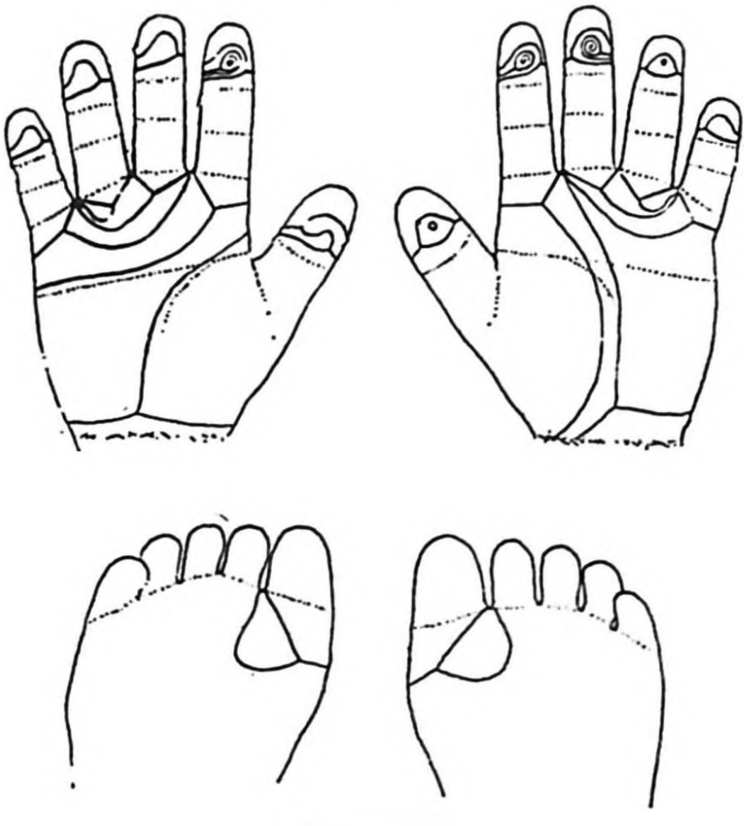

Figure 3

on the finger tips. The main line was placed proximally in the thenar region. Other findings included bilateral simian line and wide distal loops on the hallucal areas (Figure 3).

Laboratory Studies : Routine blood counts and urine analyses were normal. Buccal smear and chromosomal analyses showed normal female patterns.

X-Ray Studies : The rami of the mandibular bone were very hypoplastic (Figure 4). The anterior and posterior fontanelles were open. There was a generalized osteoporosis of the skeleton. X-rays of the wrist showed a bone age of 4 years. Terminal phalanges were smaller than normal (Figure 5). The patient died at the age of 5 years with an acute severe respiratory illness.

Figure 4

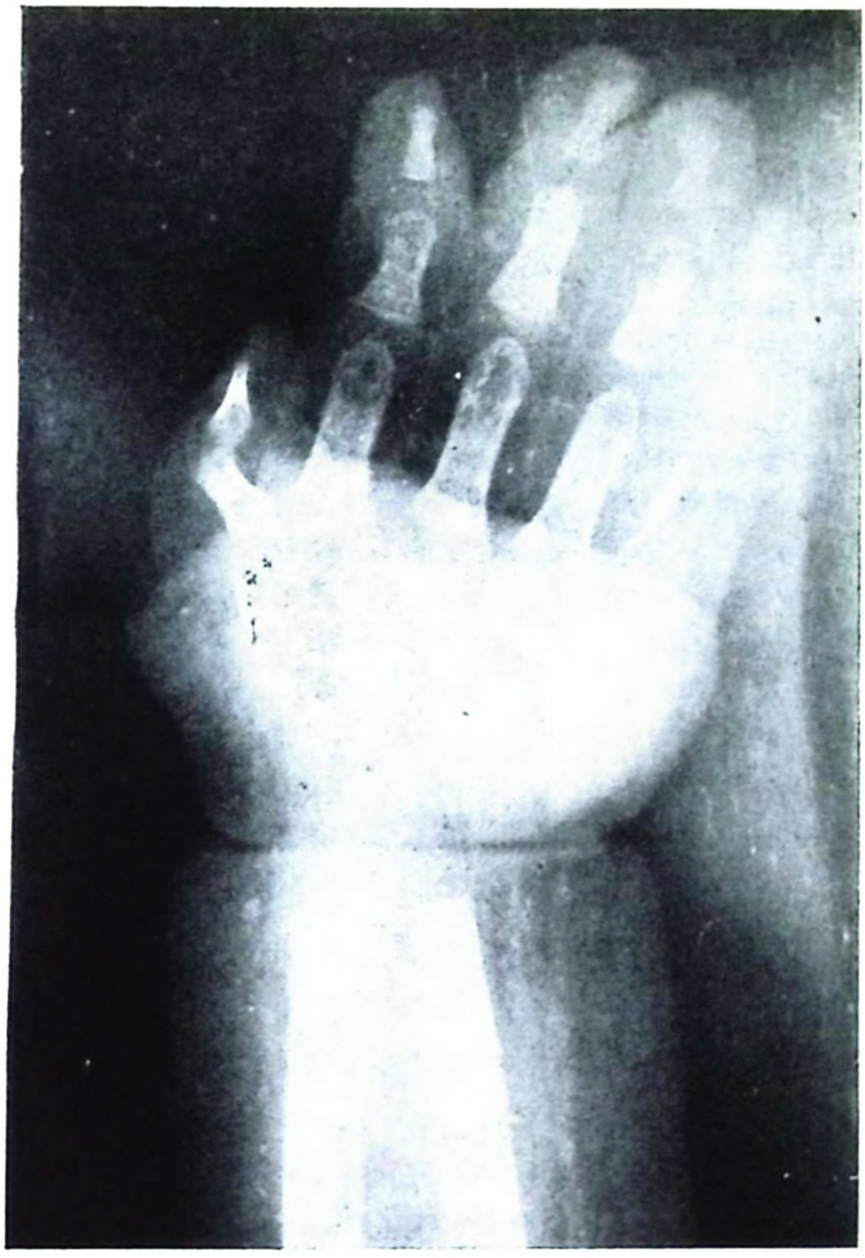

Figure 5

Discussion

An extensive review of this condition covering 25 cases from the literature was published by François[17] in 1958. The syndrome has been presented under the following synonyms : Hallermann-Streiff syndrome, François syndrome,[17] Ullrich-Fremerey-Dohna syndrome[18] and also as a dyscephaly with congenital cataracts and hypotrichosis.[10]

The patients show many abnormalites involving various systems. The most common eye findings are microphthalmia, bilateral congenital cataracts, blue sclera, corneal vaulting, iris atrophy posterior synechia, glaucoma, nystagmus, strabismus and ptosis. The cataracts are unique in this syndrome : It has been reported that they might undergo spontaneous rupture and resorption.[10 19]

Dental anomalies comprise aplasia of five or six permanent teeth, malposition and enamel hypoplasia. Furthermore deciduous and permanent teeth erupt irregularly.[6] Sometimes one or more of the upper incisors are absent altogether, but very rarely erupted teeth are present at birth. For this reason the possibility of dental anomalies must be kept in mind. Therefore every case, if possible, should be examined by an orthodontist.

Growth is severely retarded but dwarfism is proportionate. The hair and eyebrows are very scanty, fine and sometimes whitish. Eyelashes are short or sometimes lacking. The skin of the scalp and face, especially over the nose, is thin and atrophic. The anterior fontanelle is large and remains so for a long time. The skull is usually brachycephalic. The zygomatic process is hypoplastic. The most impressive change concerns the mandible and temporomandibular joints.[9] There is hypoplasia of the mandibular rami with anterior displacement of temporomandibular joints and opening of the mouth is very difficult because of this deformity. Platybasia, and shallow sella tursica,[19] calcification of the falx cerebelli may be noticed on the skull-rays.

Public and axillary hair are very sparse and external genitalia are underdeveloped. Undescended testes are very common.

Although mental and motor retardation and hyperextensibility of joints are common, sometimes psychomotor retardation is not present.

The condition is generally accepted as an autosomal recessive trait.[20] It is seen equally in both sexes.[5 7 9] Familial occurrence has been reported[11] and Van Balen described this syndrome in two male twins, allegedly monozygotic.[21] It is also possible that it represents a new dominant gene mutation.[22]

Vaginal bleeding in the first trimester of pregnancy, as in our case, has been reported. There is no chromosomal abnormality.[5 13 23 24] Environmental factors such as teratogens or infectious agents may be responsible for this condition.

It is interesting to note that our patient had many dermatoglyphic features similar to those seen in the patient described by Walbaum et al, with the exception that their case did not have bilateral simian lines.

Summary

Clinical, radiological, cytogenic and dermatoglyphic findings are described in a case of dyscephalia mandibulo-facialis (Hallermann-Streiff Syndrome).

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