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HYPOGLOSSIA-HYPODACTYLIA (HANHART'S) SYNDROME WITH SENSORINEUROL HEARING LOSS*

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SUMMARY: Tüysüz B, Erginel A, Unutmaz T, Cenani A. (Department of Pediatrics, Istanbul University Cerrahpaşa Faculty of Medicine, Istanbul, Turkey). Hypoglossia-hypodactylia (Hanhart's) syndrome with sensorineural hearing loss. Turk J Pediatr 1994; 347-352.

Hanhart's syndrome, an uncommon disorder characterized by severe micrognathia, hypoglossia and absence of the extremities, belongs to the oromandibular-limb hypogenesis group of diseases.

Our patient was admitted with the complaints of abnormality of the mouth and absence of the upper extremities. Physical examination revealed severe micrognathia, a small tongue adhered to the base of the mouth, absence of bone, muscle and skin tissue at the lower edge of the right humerus and at the middle zone of the left ulna and radius. According to these findings, the diagnosis of hypoglossia-hypodactylia (Hanhart's) syndrome was made. At the age of 1 1/2 years, tympanometric examination revealed conductive hearing loss due to dysfunction of the eustachian tube. Auditory brainstem responses also revealed bilateral severe sensorineural hearing loss.

A literature search revealed two cases of Hanhart's syndrome with conductive hearing loss, but no report of sensorineural hearing loss as part of this entity. Therefore, we present this case of Hanhart's syndrome with severe sensorineural hearing loss. *Key words: Hanhart's syndrome, hypoglossia-hypodactylia, aglossia-adactylia, sensorineural deafness.*

Cases presenting with absence of the extremities and oral cavity malformations have, since the beginning of this century, been reported under various names, including aglossia-adactylia syndrome, hypoglossia-hypodactylia syndrome, glosso-palatine ankylosis, ankyloglossia superior syndrome, and Hanhart's syndrome[1,2]. The syndrome, which is one of the diseases grouped under the name of oromandibular limb hypogenesis, was described by Hanhart[2] in 1947 and is characterized by severe hand and/or foot defects together with oral abnormalities.

Oral malformations consist of extreme micrognathia, hypoglossia-to-aglossia, adhesion of the tongue to the law and fibrous bands between the upper and lower jaws[2,4,5]. There may also be paralysis of cranial nerves III, V, VI, VII, IX

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and XII[4,6]. Hearing loss is rare in Hanhart's syndrome[5], but two cases with conductive hearing loss have been reported[7,8]. In this report, a case of Hanhart's syndrome with bilateral sensorineural hearing loss is presented.

Case Report

A three-day-old boy was admitted because of absence of upper extremities and abnormality of the mouth. He was born at term. His birth weight was 2310 gr (< 3rd percentile), length 49 cm (25th-50th percentile) and the head circumference 36 cm (90th percentile). He was the first child of unrelated healthy parents (mother 20, father 26 years old). The mother had previously had a spontaneous abortus at two-and-one-half months of gestation. There was no similar case in the family. During pregnancy the mother had received no drugs, was not x-rayed and had no illness with fever and rash.

Severe microretrognathia, a defect in the symphysis of the jaw and hypoplasia of the tongue were detected (Fig. 1). The tongue was adhered to the base of the mouth and the palate was high-arched. Two hemangiomas (3×5 cm) were

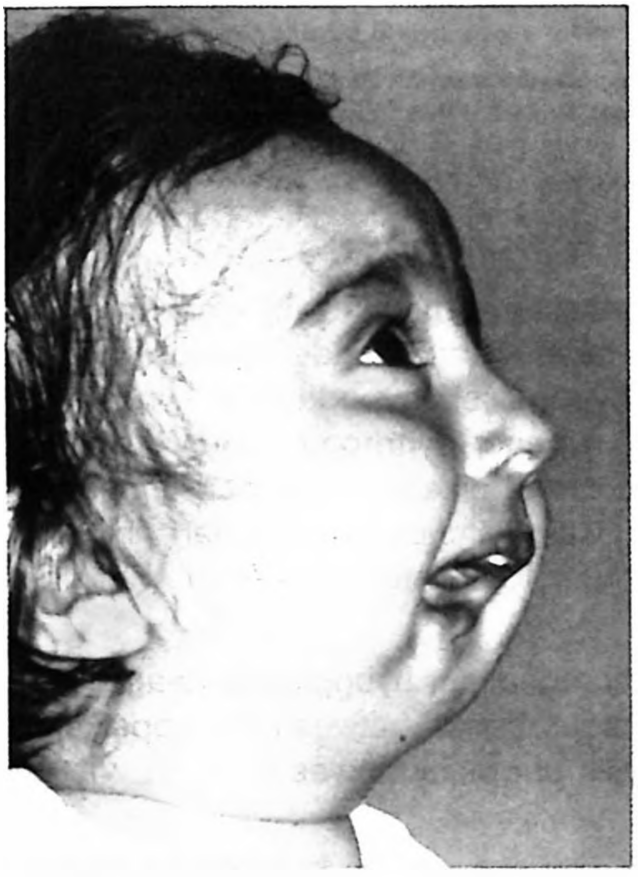

Fig. 1: Lateral view of the patient displaying severe microretrognathia, defective jaw and hypoplastic tongue.

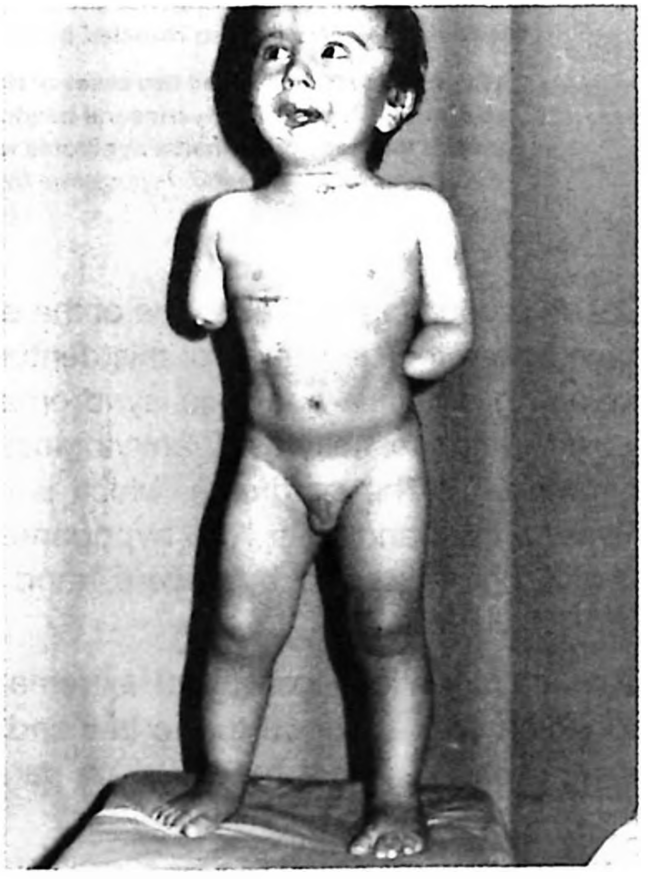

Fig. 2: Photograph displaying the absence of the distal end of the right humerus and part of left forearm.

present between the right eyelid and eyebrow and on the forehead. The eyes and ears were not malformed. Bone, muscle and skin tissues were absent at the distal end of the right humerus and in the middle of the left radius and ulna. There was a rudimentary finger at the end of the right arm consisting of skin and subcutaneous tissue (Fig. 2). No malformation of the lower extremities was noted. Neurologic examination revealed no paralysis of the cranial nerves. The eyes and the other systemic findings were normal.

Complete blood count and urinalysis were normal. Biochemical findings of the blood were normal. Humoral and cellular immunological studies were normal. Chest x-ray, electrocardiography, abdominal ultrasonography and intravenous pyelography revealed no congenital malformations. Cranial computed tomography was normal except slight dilatation of the ventricles. The karyotype was 46XY by G-banding technique.

The patient was considered to be a case of Hanhart's syndrome. When he was admitted again at the age of one and a-half years, his weight was 11 kg (25th-50th percentile) and his height was 78 cm (25th percentile). He had six hypoplastic and irregular teeth. His neuromotor development was reported to be normal. His speech was limited to a few words. Denver test revealed that fine and gross motor skills were compatible with his age. Speech development was compatible with 14 months.

Tympanometric evaluation revealed a conductive hearing loss which was due to dysfunction of the eustachian tube. Auditory brainstem responses were evaluated. Wave responses to acoustic stimulations of 80 decibel intensity in both ears were examined; configuration of the waves was irregular and the fifth wave was not observed. In conclusion, severe bilateral sensorineural type hearing loss was detected.

The patient was referred to the plastic surgery department. The defect in the mandibula was repaired and the tongue was surgically released.

Discussion

In 1976, Hermann et al[6] evaluated seven personal and 62 previously reported cases of oromandibular-extremity hypoplasia. Patients with oro-acral malformations accompanied by nuclear agenesis of the cranial nerves and pectoral defects were classified as having Poland-Mobius syndrome. Patients with ocular hypertelorism, cleft-palate and variable degrees of hypodactyly of hands and feet were classified as having Charlie-M syndrome. Cases of distal agenesis of the extremities and severe micrognathia-microglossia and sometimes cranial nerve palsy were classified as having Hanhart's syndrome. There is considerable overlap between these syndromes. In the same study, 35 patients

with Hanhart's syndrome were coded according to the severity of the clinical findings: Grade-0: no pathology in the extremities; Grade-I: partial or complete cutaneous syndactyly, with or without short digits; Grade-II: presence of oligodactylia; Grade-III: complete or partial agenesis of the hands and/or feet; Grade-IV: complete or partial transverse agenesis of a forearm or leg; Grade-V: partial or complete hemimelia. According to this classification, our patient was compatible with Grade-V. In this study, severe micrognathia was observed in 26 cases while five cases had slight micrognathia. Four patients lacked this pathology. The anterior two-thirds of the tongue was absent in 21 cases. Even though some authors name Hanhart's syndrome as "aglossia-adactylia", the tongue is expected to exist, and rudimentary or not, it adheres to the base of the mouth. In our case the tongue was hypoplastic, adhered to the base of the mouth and there was a defect in the symphysis of the jaw. In Hanhart's syndrome, the palate is narrow and high-arched, maybe cleft, and the teeth are hypoplastic and irregular[1-5]. In our patient, the palate was high-arched, and the teeth were irregular and hypoplastic.

Chandra-Sekhar et al.[7] reported two patients: one of them had atresia of the external auditory canal and temporal bone, and the other had bilateral conductive hearing loss. Our patient had no malformation in the external ear but he had conductive hearing lose due to dysfunction of the eustachian tube and severe bilateral sensorineural hearing loss determined by auditory brainstem responses. It is known that paralysis of cranial nerves. III, V, VI, VII, IX, XII might be present in Hanhart's syndrome[4,6]. The tests to determine whether our patient's sensorineural hearing loss was due to paralysis of the eighth cranial nerve or whether there was an accompanying cochlear pathology cannot be done in this age group.

Our patient was born at term and was small for gestational age. In the literature [1-8], most of the cases were also born small for gestational age.

Congenital heart and renal disease, cryptorchidism and anal atresia are other possible features of the syndrome[1-6], but none of them was present in our patient. Cases with hydrocephaly and porencephaly have been reported[4,8]. In our patient physical examination revealed no increase in intracranial pressure, and cranial tomography revealed only minimal ventricular dilatation. A case of Hanhart's syndrome with colobomatous microphthalmia has been reported[9]; however, examination of the eyes was normal in our patient.

Most of the reported patients had normal intelligence[1-6]. Even with hypoglossia, speech is surprisingly good. In our patient the Denver test results revealed that fine and gross motor skills were in accordance with his age at 18 months, while speech development was compatible with a child of 14 months.

The prevalence of the disease has been reported by two different authors to be one in every 500,000 births and less than one in every 20,000 births [4,10].

The etiology of Hanhart's syndrome is not clearly understood. Thirty-five cases which were analyzed by Herman et al.[6] were all sporadic, chromosome analyses were normal and the parents were not relatives. Tunçbilek et al.[11] reported three cases whose parents were relatives and suggested that the syndrome might be autosomal recessive. However, in Turkey the incidence of consanguineous marriages is high and this condition might be a coincidence. Temtamy[12], McKusick[13] and later Scott[14] determined dental malformations in first-degree relatives of patients and suggested that an autosomal dominant gene with low penetrance might play an etiological role. In our case the karyotype was normal. Similar cases and consanguineous marriage were not reported in the family history.

Many investigators[11,15-18] have suggested that between the 5th and 7th weeks of gestation, thrombosis or vasoconstruction of the vessels caused by drugs or other teratogenic factors resulted in these clinical malformations of the Meckel cartilage, the tongue and the extremities developing from the same embriological region. In our patient the mother had not been exposed to any teratogenic agent.

Prognosis is usually good if aspiration due to malformations of the mouth does not occur[1,5,6,19].

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