Hanhart syndrome: A rare case report and review of literature

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ABSTRACT

Hanhart syndrome is a rare congenital and genetic disease, in which the most common signs are short, incompletely developed tongue (hypoglossia), absent or partially missing fingers and/or toes (hypodactylia), malformed arms and/or legs (peromelia), and small jaw (micronathia). Here, we report a case of Hanhart syndrome in an 18-year-old boy. The boy presented with few extraoral and intraoral abnormalities such as short toes and phalanges along with partial syndactyly in the left hand only were the most relevant. Other features such as micro and retrognathic face, incompetent lips, and wide nasal bridge were also significant. The boy was suffering for difficulties in speech and swallowing due to small tongue size, high arched palate, crowding, and few missing teeth. To provide adequate treatment to a patient with Hanhart syndrome, this study aimed to review and to analyze this literature and treatment protocols.

Key words: Brachydactyly, Hanhart syndrome, Microglossia, Partial syndactyly, Retrognathia

Hanhart syndrome is a rare developmental disorder that affects males and females equally. Fewer than 1 in 20,000 children are affected with this syndrome [1]. In 1950, Hanhart described this anomaly as bird-like face profile caused by micrognathia, protruding nose, opisthodontia, peromelia, and small growth [1]. Grislain et al. also observed aglossia and hypoglossia in few cases of Hanhart syndrome [2].

The etiology of Hanhart syndrome is not fully understood. However, researchers suspect that genetic and/or environmental factors may be the cause for this condition. No specific genes have been identified. Possible causes such as exposure of the pregnant mother to radiation, teratogenic medications, hypothermia, trauma, or disrupted blood flow to the baby in the womb have been suggested [3,4]. Consanguineous marriage increases the risk to develop this condition. It is important to note that having a risk factor does not mean that one will get the condition and not having a risk factor does not mean that an individual will not get the condition [5]. We report the case of Hanhart syndrome diagnosed in an 18-year-old boy that had retrognathia, microglossia, hypodontia, severe malocclusion, brachydactyly, and unilateral partial syndactyly.

CASE REPORT

An 18-year-old boy was referred to the Department of Oral Pathology at our hospital at Kolkata with complaints of speech problem for 13 years and bird-like appearance (Fig. 1). There was no family history of similar anomalies. A history of consanguineous marriage was present in his family.

Upon the first visit to the Oral Pathology OPD, a thorough extraoral clinical examination revealed short toes and phalanges along with partial syndactyly in the left hand only (Fig. 2a and b). He could cope with his limb anomalies from everyday life. Intraoral examination revealed microglossia, retrognathia, deep palatal arch, small size tongue, and the mobility of tongue was slightly restricted due to tongue tie and almost no chin contour (Fig. 3a). The lips were incompetent with the protruding upper incisors. Mouth opening was 40 mm that is within normal range.

After thorough clinical examination, the patient was advised for complete blood count, blood sugar estimation both fasting and postprandial, bleeding time, clotting time, erythrocyte sedimentation rate, and echocardiogram, and all the investigations were within normal limits. Viral markers for hepatitis B, C, and HIV were negative. Radiological investigation (orthopantomogram [OPG], chest X-ray) was done. The OPG revealed few missing teeth and severe malocclusion (Fig. 3b). Chest X-ray was also normal.

The patient was provisionally diagnosed as Charlie M. syndrome or Hanhart syndrome. Since there were ocular hypertelorism, cleft palate, conical teeth, cleft tongue, an absence of cranial nerve palsy and the patient was not associated with any neurological problem, a final diagnosis of Hanhart syndrome was made. The patient was sent to the Department of Oral Surgery for orthognathic surgery followed by orthodontic treatment to correct the malocclusion. After the operation, the patient was very satisfied with his facial appearance. He felt better psychologically and enabled him to socialize.
Hanhart syndrome is a rare anomaly with many characteristics that are not present in all cases. It is also known as aglossia adactyla, hypoglossia-hypodactylia syndrome, and peromelia with micrognathia. Individuals of all racial groups may be affected [6]. Some clinicians said that the factors responsible for Hanhart syndrome may occur in the developmental stage when there is a clot formed within a blood vessel (thrombus) or has traveled through the bloodstream and become lodged in a vessel (embolus) resulting in the deficient of blood supply to the parts of the embryo that develops into the arms, legs, hands, feet, tongue, mouth, jaw, and/or some parts of the brain. Such a clot may result from exposure of the embryo to certain drugs taken during pregnancy or could result from the death of another embryo in the uterus that was formed from the same fertilized egg (discordant monozygotic twins) [5]. In the present case, after taking proper family history, it was agreed that consanguineous marriage was present in his family. In this view, it can be pointed out that how a consanguineous marriage increases the chances of getting this type of disorder compared to an individual without the risk factors.

The signs and symptoms of Hanhart syndrome includes small mouth (microstomia), incompletely developed tongue (hypoglossia), small or absence of tongue (microglossia or aglossia), absence/partially missing or shortened fingers and/or toes (adactyly/hypodactyly or brachydactyly), jaw abnormalities such as micrognathia, retrognathia, or partially missing lower jaw, high-arched, narrow, or cleft palate, absent or unusually formed arms and/or legs (limb phocomelia), missing teeth (hypodontia), absence of major salivary glands, abnormality of oral frenula, hypoplasia of the zygomatic bone, clubbing or fusing of fingers (finger syndactyly), increased space between the eyes (telecanthus), and wide nasal bridge [5].

Additional abnormalities such as spleen and gonads (i.e., testes in males and ovaries in females) may have fused together during fetal development (splenogonadal fusion), absence of kidney (unilateral renal agenesis) and cyst in the brain (porencephalic cyst) may occur in association with Hanhart syndrome. The complications of Hanhart Syndrome may include severe physical deformities, intellectual disability, neurological speech impairment, feeding difficulties, and death in infancy.

The differential diagnoses for these disorders are hypoglossia-hypodactylia syndrome, glossopalatine ankylosis, Moebius syndrome, limb deficiency splenogonadal fusion syndrome, Charles M. syndrome, ankyloglossia superior syndrome, Poland syndrome, and phocomelia syndrome [5]. When micrognathia is associated with microglossia, it is difficult to make a distinction between Hanhart syndrome and hypoglossia-hypodactylia syndrome. Under the latter diagnosis, Purohit et al. [7] and Arshad and Gosh [8] reported that hypoglossia and micrognathia are associated with anterior maxillo-mandibular fusion.

The treatment of Hanhart syndrome requires the coordinated efforts of a team of specialists (pediatricians, plastic and orthopedic surgeons, dental specialists, speech pathologists, and physical therapists). In infants with Hanhart syndrome, any feeding difficulties resulting from tongue, mouth, and/or jaw malformations must be treated immediately through surgical correction, use of artificial devices (prostheses), and/or physical therapy to ensure proper nutrition and growth. Difficulties with speech can be treated with speech therapy. Depending on the severity of any limb abnormalities, children may have difficulty performing skills that require coordination of motion (motor skills), such as walking and writing. Another treatment is symptomatic and supportive, based on the individual’s case [5]. As far as the face is concerned, Wexler and Novark described a procedure that consisted of reduction rhinoplasty, sagittal split of the mandible with advancement, iliac crest onlay bone graft to the lower border of the mandible, and deepening of the vestibular sulcus with a split-skin graft [9]. The long-term prognosis for people with Hanhart syndrome varies and largely depends on the signs and symptoms.
Signs of Hanhart syndrome may be identified before birth by ultrasonography. Therefore, in the case of consanguineous marriage, ultrasonography is compulsory in prenatal life to minimize these complications. Early detection will help to take necessary action against the abnormalities. Limb and/or craniofacial abnormalities may be treated with surgery and/or prosthesis. Affected children may also need speech therapy, physical therapy, and/or occupational therapy [10].

CONCLUSION

Hanhart syndrome is mainly diagnosed based on the presence of characteristic signs and symptoms. The diagnosis may be suspected in prenatal life if concerning features are seen on ultrasound. Few craniofacial abnormalities can be life threatening that may impair breathing, eating, and/or swallowing. However, early diagnosis and treatment can improve survival rate and the quality of life. Regular medical screening at periodic intervals with physical examinations is recommended. The affected individuals may also benefit from social support, special education, vocational, and occupational services.

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