Hallermann–Streiff Syndrome: Two Typical Cases with Dental Treatment and Primary Aetiological Investigation

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Abstract: *Presented are two case reports of the rare congenital anomaly Hallermann-Streiff Syndrome. One patient received dental treatment and cytogenetic examination for primary aetiological investigation.*
Key words: *Hallermann–Streiff syndrome*

Hallermann–Streiff syndrome (HSS) is a rare congenital anomaly characterised by a distinctive malformation of the craniofacial region (bird-like face), mandibular and maxillary hypoplasia, dyscephaly, congenital cataracts, microphthalmia, hypotrichosis, skin atrophy, and proportional short stature (OMIM® 234100)[1].

This syndrome chiefly affects the head and face. Dental abnormalities requiring treatment are common. The diagnosis is usually difficult owing to lack of knowledge of the syndrome, which can be incorrectly diagnosed as ectodemal dysplasia or even only hypodontia. Two patients with most of the typical characteristics of HSS are presented. One patient received dental treatment and cytogenetic examination for primary aetiological investigation.

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Case reports

The two patients were both outpatients of the School and Hospital of Stomatology, Peking University. Routine oral and x-ray examination was performed. The diagnosis of HSS was finally established. Family histories were recorded. Peripheral blood taken from the patient in case 1 was sent to the Department of Cytogenetics, Health Science Center, Peking University, for cytogenetic examination to explore whether or not there was chromosomal abnormality. Both patients and the family members gave informed consent and the study was conducted under permission of the Ethics Committee of Peking University Health Science Center.

Case 1

General information

Case 1 was a 20-year-old female with obvious face and dental abnormalities (Fig 1). She was a slim and short girl, with a peaked nose, bilateral microphthalmia and crossed eyes, prominent mandibular angles and angular chin, showing a typical 'bird-like' face (Fig 1a). Suffering from congenital hypotrichosis, she wore a hairpiece and her eyebrows were enhanced with brow pencil. Although she suffered from xerotic skin and sparse hair, she could perform common sports, but heavy exercise

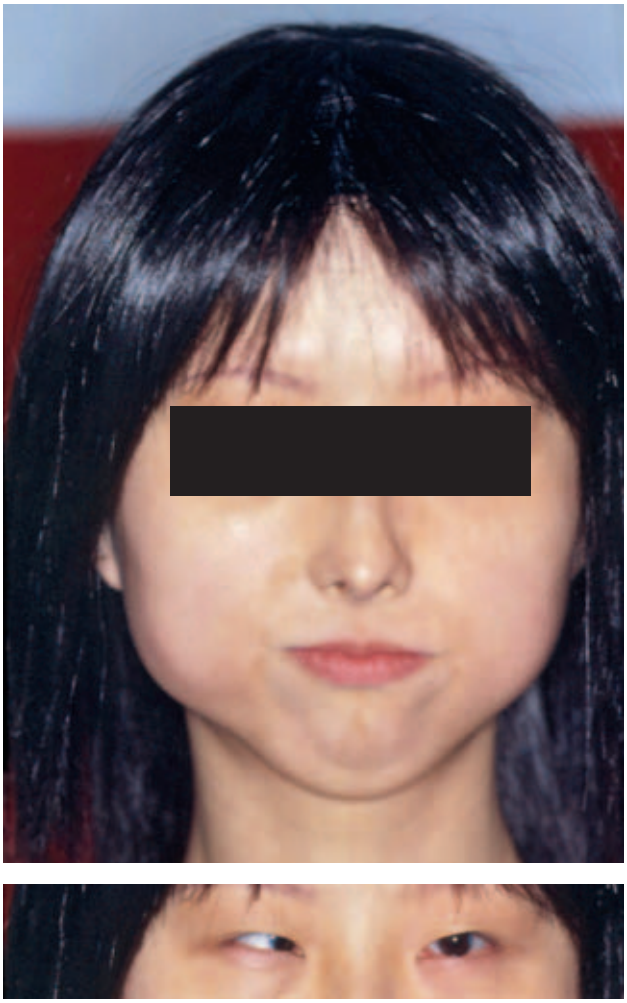


Fig 1a 'Bird-like' face, with all the typical characteristics of Hallermann–Streiff syndrome (note hairpiece and enhanced eyebrows).

could not be endured because of hypotrichosis and hypohidrosis. No mental retardation was found and she had passed the national college entrance exam and could communicate with others normally.

Her mother reported that the patient had been born with two natal teeth and soon these teeth were lost. At one year old, she received an ophthalmologic operation of phacocystectomy because of her congenital cataracts on both eyes.

The patient's parents were normal, non-consanguineous and had no relatives with analogous appearance. The parents had another daughter 3 years younger who was completely normal and healthy. According to her mother's recollection, nothings seemed unusual in the patient's embryonic period and childhood except that the mother had worked in a ^{3}H radioisotope laboratory before and during the pregnancy.

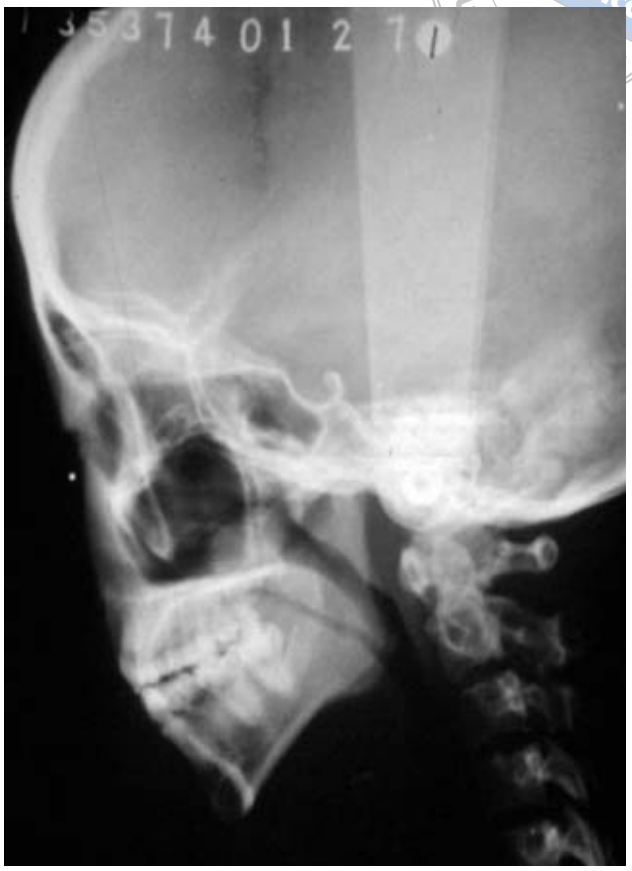


Fig 1b The cephalogram showed a large notch in the inferior border of the mandible and a high Frankfort-mandibular plan angle.

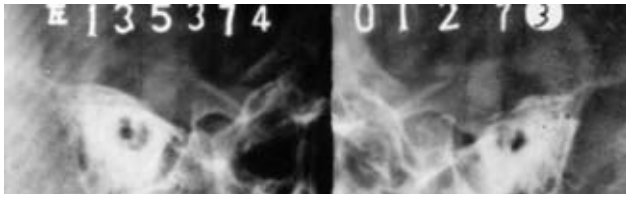


Fig 1c Bilateral glenoid fossa were shallow and no cortical bone coating on condyle was seen on the Schüller's radiographs.

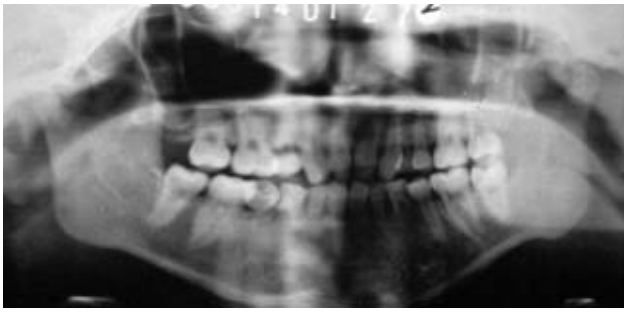
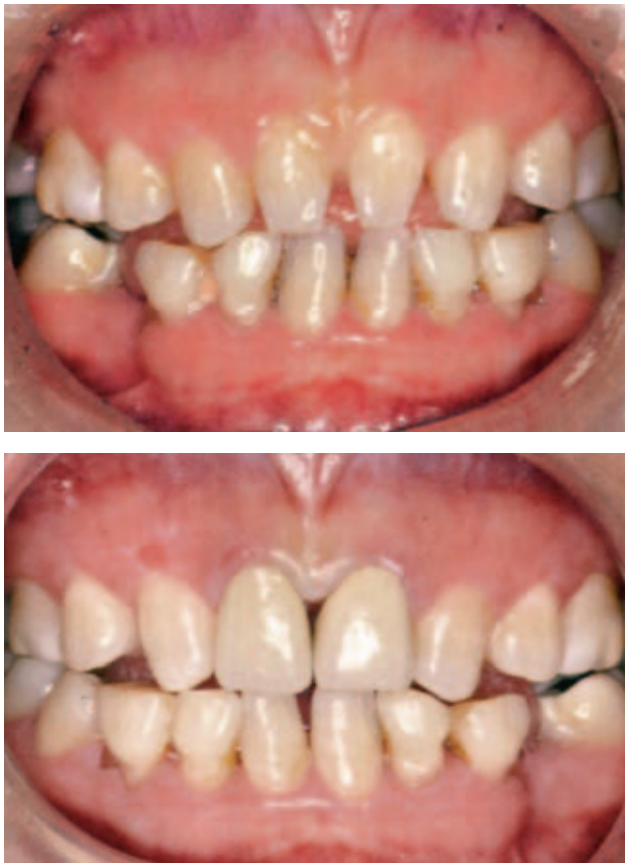


Fig 1d The panoramic radiograph showed congenitally missing teeth 15, 12, 22, 25, 34, 33, 32, 42, 43, 44, and 45, and some mandible primary teeth remained.



Figs 1e and 1f Pre- and post-restoration of the abnormal maxillary incisors.

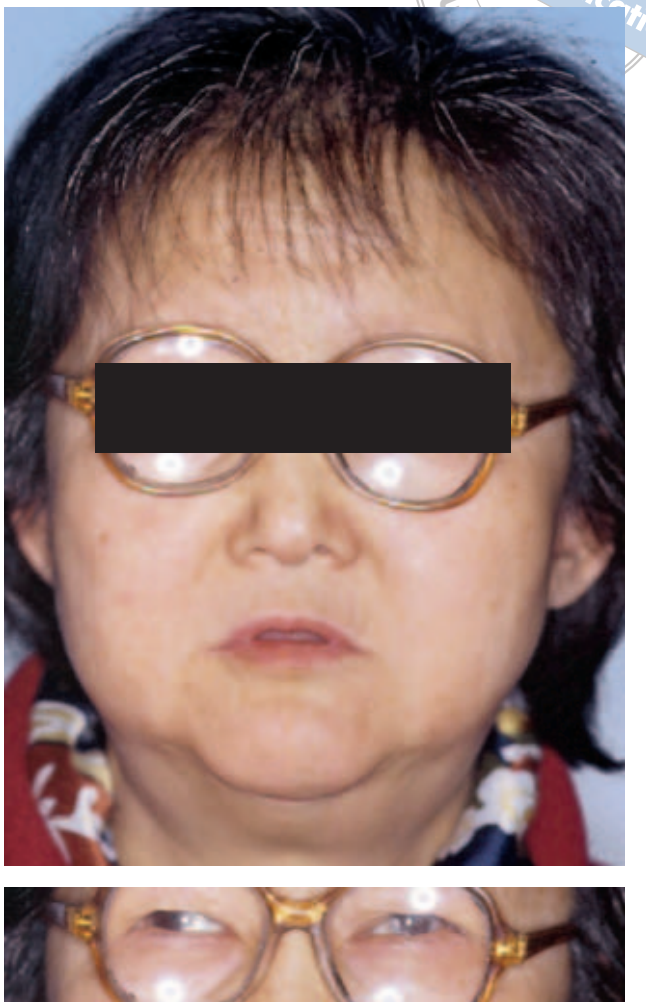


Fig 2 'Bird-like' face of the second patient. Note that the patient has no eyebrows.

Dental and maxillofacial abnormalities

With obvious developmental dyscephaly, the patient had the distinctive appearance of HSS. A cephalogram showed large notches in the inferior border of the mandible and a high Frankfort-mandibular plane angle (Fig 1b).

Bilateral glenoid fossae were abnormally shallow, especially in right side, and no cortical bone coating on the condylar processes was seen on the Schüller radiographs (Fig 1c), indicating temperomandibular joint (TMJ) dysplasia on both sides at the corresponding age. The patient had normal primary teeth according to the mother's description but her permanent teeth showed hypodontia and malocclusion. As well as all the third molars, teeth 15, 12, 22, 25, 34, 33, 32, 42, 43, 44 and 45 were missing congenitally. Some mandibular primary teeth remained, however (Fig 1d).

Some of the existing teeth were malformed, especially the incisors, which were 'screwdriver-like' (Fig 1e). According to the patient's requirement, two metal-ceramic crowns were made on the maxillary central incisors to improve aesthetics (Fig 1f). Abnormal tension of skin and mucosa around the mouth could be felt when taking the dental impression.

Cytogenetic examination

Cytogenetic examination showed that the patient had no chromosome aberrations.

Case 2

The second patient was a 50-year-old female who also had all the typical features of HSS, showing crossed eyes

and a typical 'bird-like' face (Fig 2). She had poor vision because of congenital cataracts.

With congenitally missing teeth and periodontitis, she had lost most of her teeth when she presented in the hospital. Her mouth could not open sufficiently wide for oral examination owing to the fibrosis of the lips and cheek skin and mucosa. She had xerotic skin and sparse hair, but these were less severe than in case 1, and she said that hypohidrosis was much relieved with advancing age. According to the patient's memory, nothing particular was related to development of the malformations before or after her birth. No family history was found and she had a healthy son of 20 years old.

Discussion

Diagnosis

Hallerman (in 1948) and Streiff (in 1950) described a syndrome characterised by a 'bird-like' face, congenital cataracts, mandibular hypoplasia and dental abnormalities, and later defined this as HSS[2]. HSS is an extremely rare developmental abnormality, and no information on incidence exists, with only 150 cases reported[3]. The diagnosis of this syndrome is mostly based on clinical features and clinical course, and characteristic changes on radiographs of the TMJ[4].

Although many developmental abnormalities affecting ectodermal or oculomandibular organs may have similar symptoms to HSS, the criteria established by Francois[5] and the differential diagnosis demonstrated in detail by Limeres et al[3] make it easy to differentiate the syndrome. On the basis of typical clinical characteristics and thorough review of the literature, the present two patients were diagnosed as having HSS without any doubt. Both of the cases reported here had all the typical features of HSS: dyscephalia with 'bird-like' face, dental anomalies, proportionate nanism, hypotrichosis, bilateral microphthalmia and congenital cataracts on both eyes.

Aetiology

The aetiology of the HSS remains obscure. Most reported cases were sporadic with no sex predilection. Familial cases either result from a consanguineous marriage or inheritance pattern[1]. Cohen[2] comprehensively reviewed this syndrome. He pointed out that all cases were sporadic, and the disorder had been both concordant and discordant in monozygotic twins[2]. Normal karyotypes and chromosomal examination were found in many cases[6,7], and HSS patients with normal children are not rare[7,8].

David et al[7] suggested that this syndrome is a sporadic mutation and the inheritance pattern is unknown. In their report of 15 patients with HSS, no patients had similarly affected siblings, five patients had produced normal cytogenetic studies and showed normal chromosomes, and four patients had normal unaffected children.

Pizzuti et al[9] reported a homozygous *GJA1* gene mutation in a patient of oculodentodigital dysplasia, which shared several clinical characteristics similar to HSS. However, in their research, a case of full-blown HSS phenotype showed no mutations on *GJA1*.

Hou[10] reported a case of 2-month-old baby with HSS. Besides typical HSS features, choanal atresia and small cerebellum, he also reported a very low insulin-like growth factor I level, hypothyroidism and generalised organic aciduria in the baby. The associated anomalies in this patient may broaden the clinical spectrum of HSS.

At present, the aetiology of HSS remains unknown. The two cases reported also appeared sporadic. Case 1 had no inherited trait and no chromosomal aberration, and case 2 had a normal son of 20 years old. However, the mother of case 1 had worked in a ^{3}H radioisotope laboratory before and during her pregnancy. Whether or not this was a potential factor for the mutation is unknown and has not previously been reported in the literature.

Dental treatment and psychological care

Characteristics of craniofacial and dental abnormalities or dental treatment considerations in patients with HSS have been well decribed[6,11-17]. Potential complications such as early pulmonary infection, respiratory embarrassment, obstructive sleep apnoea and anaesthetic risk related to a narrow upper airway caused by the craniofacial configuration have been considered[2], but most of these patients could undergo conventional dental procedures and local anaesthesia successfully. Previous studies suggested that young patients with HSS and other similar syndromes should be involved in personalised oral health prevention programmes as early as possible, and more attention should be paid to developmental changes and psychological health[3,18]. If necessary, a treatment plan may consist of orthodontic, endodontic and periodontic procedures and proper restorations to help the patients improve function and aesthetics.

In the present report, the patient of case 1 had two maxillary incisors restored with porcelain-fused-to-metal crowns to improve the aesthetics (Figs 1b and 1c). Local infiltration anaesthesia was performed during preparation of the vital teeth. The patient was satisfied

with the restoration. Further restorations may be applied to fulfil the functional and aesthetic requirements when the retained primary teeth are lost.

References

1. MIM 234100, OMIM (Online Mendelian Inheritance in Man), available at: http://www.ncbi.nlm.nih.gov.
2. Cohen MM Jr. Hallermann–Streiff syndrome: a review. Am J Med Genet 1991;41:488-499.
3. Limeres J, Abeleira M, Tomas I, Feijoo JF, Vilaboa C, Diz P. An atypical Hallermann–Streiff syndrome. Focus on dental care and differential diagnosis. Quintessence Int 2004;35:49-55.
4. Bissonnette B, Dalens B. Syndromes: Rapid Recognition and Perioperative Implications. McGraw-Hill Professional, 2006:357-358.
5. Francois J. A new syndrome: dyscephalia with bird face and dental anomalies, nanism, hypotrichosis, cutaneous atrophy, microphthalmia, and congenital cataract. AMA Arch Ophthalmol 1958;60:842-862.
6. Sharp EC, Kopel HM. Craniofacial and oral manifestations in oculomandibulodyscephaly (Hallermann–Streiff syndrome). Oral Surg Oral Med Oral Pathol 1970;30:488-496.
7. David LR, Finlon M, Genecov D, Argenta LC. Hallermann–Streiff syndrome: experience with 15 patients and review of the literature. J Craniofac Surg 1999;10:160-168.
8. Ponte F. Further contributions to the study of the syndrome of Hallermann and Streiff. (Congenital cataract with 'bird's face'). Ophthalmologica 1962;143:399-408.
9. Pizzuti A, Flex E, Mingarelli R, Salpietro C, Zelante L, Dallapiccola B. A homozygous GJA1 gene mutation causes a Hallermann-Streiff/ODDD spectrum phenotype. Hum Mutat 2004;23:286.
10. Hou JW. Hallermann–Streiff syndrome associated with small cerebellum, endocrinopathy and increased chromosomal breakage. Acta Paediatr 2003;92:869-871.
11. Defraia E, Marinelli A, Alarashi M. Case report: orofacial characteristics of Hallermann–Streiff syndrome. Eur J Paediatr Dent 2003;4:155-158.
12. Gopakumar M, Hedge AM. Atypical Hallerman–Streiff syndrome: a case report. J Clin Pediatr Dent 2005;30:73-76.
13. Honda E, Inoue T, Domon M, Sasaki T, Uchida T. Dental radiographic signs characteristic to Hallermann–Streiff syndrome. Oral Surg Oral Med Oral Pathol 1990;70:121-125.
14. Ohishi M, Murakami E, Haita T, Naruse T, Sugino M, Inomata H. Hallermann–Streiff syndrome and its oral implications. ASDC J Dent Child 1986;53:32-37.
15. Malerman AJ, Album MM. Hallermann–Streiff syndrome: report of case. ASDC J Dent Child 1986;53:287-292.
16. Hutchinson D. Oral manifestations of oculomandibulodyscephaly with hypotrichosis (Hallermann–Streiff syndrome). Oral Surg Oral Med Oral Pathol 1971;31:234-244.
17. Steele RW, Bass JW. Hallermann–Streiff syndrome. Clinical and prognostic considerations. Am J Dis Child 1970;120:462-465.
18. Nowak AJ. Dental treatment for patients with ectodermal dysplasias. Birth Defects 1988;24:243-252.