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Case report

A case of Hallermann-Streiff syndrome with aphakia

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= Abstract =

Hallermann-Streiff syndrome is a rare disease. Approximately 150 cases have been reported, including 6 cases in Korea. The authors experienced a case of Hallermann-Streiff syndrome in a 6-year-old female with aphakia. The syndrome is characterized by a bird-like face, dental abnormalities, hypotrichosis, atrophy of the skin, bilateral microphthalmia, and proportionate dwarfism. A brief review of the literature was conducted. (Korean J Pediatr 2008;51:646-649)

Key Words : Hallermann-Streiff syndrome, Aphakia, Bird-like face

Introduction

Hallermann-Streiff Syndrome is a rare genetic disorder that is characterized by bird-like face, dental abnormalities, hypotrichosis, atrophy of skin, congenital cataracts, bilateral microphthalmia, and proportionate nanism. It was first published by Aubry in 1893[1], but the case was incomplete. This syndrome was first described completely in 1948 by Hallermann[2] and then in 1950 by Streiff[3]. In 1958, Francois reviewed literatures, analyzed the manifestations of 22 published cases and described diagnostic criteria for this syndrome[4]. Approximately 150 cases have been reported in the literatures. In Korea, 6 cases have been reported. Bilateral microphthalmia and congenital cataracts are the most common ophthalmologic signs of this syndrome. We report a rare case of aphakia in Hallermann-Streiff Syndrome.

Case report

Sex : Female

Age : 5 years and 7 months

Chief Complaint : Decreased visual acuity

Physical Findings : This child was admitted to the Department of Ophthalmology in Maryknoll Medical Center for the diagnosis and surgery for congenital cataracts. Her birth history showed a normal vaginal delivery at the fetal age of 40 weeks and her mother's obstetric history revealed no record of systemic disease or drug administration. Her parents and only brother showed no specific finding. On admission, she was 5 years and 7 months old and her height was 83.2 cm (less than 3rd percentile) while her body weight was 13 kg (less than 3rd percentile), showing a growth disorder. She showed a developmental disorder showing unassisted self-ambulation at the age of 4 years. She had a pointed nose and frontal bossing as well as bilateral microphthalmia (Fig. 1). Her hair and eyebrows were thin, her skin showed atrophy while the vessels were prominent. Her mandible was small and her teeth were irregularly developed showing a malocclusion (Fig. 2). Her respiratory sounds were regular but stridor was heard while she was sleeping. Her heart sounds were regular and

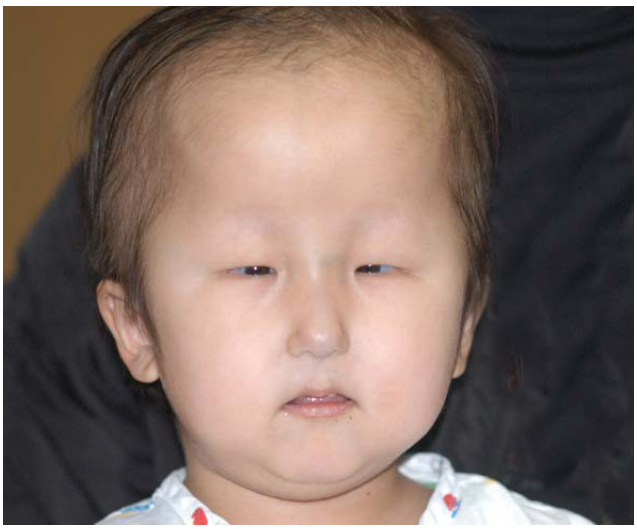

Fig. 1. The nose is pointed, the frontal head protruds, the hair and eyebrows are thin and scanty, the skin is atrophied, and the vessels are prominent.

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no cardiac murmur was heard. There was no abdominal distention.

Test Findings : On admission, her blood test and chemistry lab report showed as follows: WBC 7,400/mm^3 (neurophil, 51.5%; lymphocytes, 43.5%; monocytes, 3.7%); Hb, 11.0 g/dL; hematocrit, 33.1%; platelet, 477,000/mm^3 AST, 38 IU/L; ALT 17 IU/L, BUN, 11.0 mg/dL; creatinine, 0.4 mg/dL; protein 6.7 g/dL; albumin, 4.2 g/dL; and bilirubin, 0.2 mg/dL. Her skull x-ray revealed developmental failure in the mandible and face showing a two-fold increase in cranio-facial ratio (Fig. 3). She showed developmental retardation corresponding to development quotient (DQ) <75 on Denver developmental screening test (DDST).

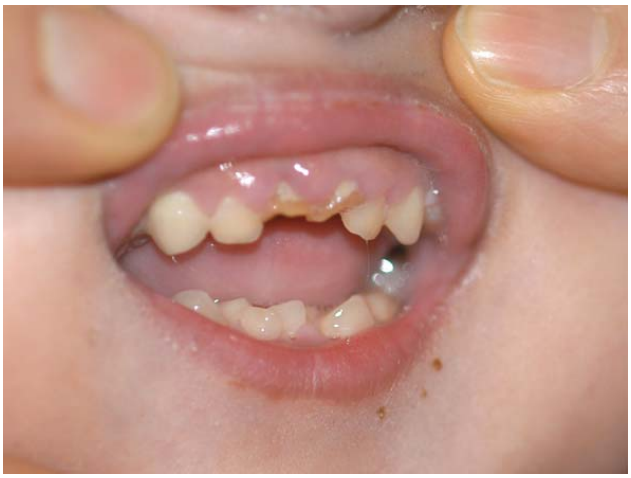

Fig. 2 The mandible is small and the teeth are irregularly developed, showing malocclusion.

In the ophthalmologic test, epiblepharon of the upper palpebra and voluntary horizontal nystagmus of both eyes, as well as esotropia of 30 prism diopter (PD) were observed. Miosis due to posterior synechiae was observed. Thus, a funduscopic examination was not possible.

An ultrasound test revealed no retinal detachment and A-scan showed the distance of the optic axis to be quite short, 17.4 mm for the right eye and 16.1 mm for the left eye, exhibiting microphthalmia. A slit lamp examination confirmed the finding of microcornea.

Treatment and Progress : She underwent ophthalmologic surgery of both eyes in turn throughout the period of two weeks. There was no difficulty in securing endotracheal intubation. As a result of surgery, aphakia covered with double-folded membrane was verified. Intraocular lenses that would fit the child's eyes could not be obtained due to microphthalmia. Intraocular lens implantation was not performed due to a concern of a possible glaucoma as a postoperative complication. She was put under observation after she was prescribed for eye glasses.

Discussion

Hallermann-Streiff syndrome, a rare congenital anomaly, was first reported incompletely by Aubry[1] in 1893. In 1948, Hallermann[2] made a complete report for the first time, which was followed by Streiff[3] Report in 1950. In 1958,

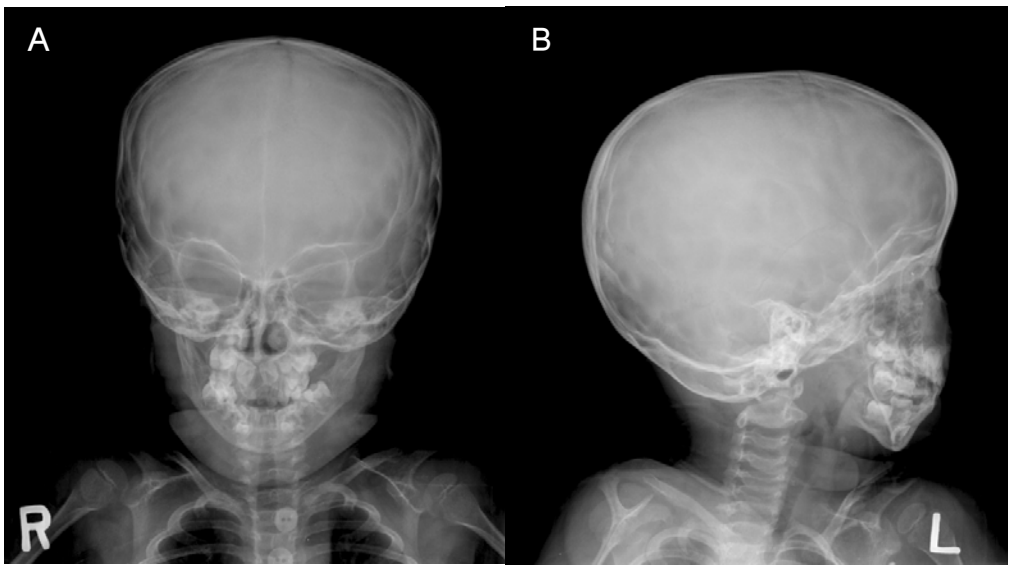

Fig. 3. The mandible and the facial bone are hypoplasic. The skull is large and poorly ossified with frontal bossing.

Table 1. Diagnostic Criteria for Hallermann-Streiff Syndrome[4]

1	Dyscephalia and bird face
2	Dental anomalies
3	Proportionate nanism
4	Hypotrichosis
5	Atrophy of the skin
6	Bilateral Microphthalmia
7	Congenital Cataract

Francois[4] analyzed clinical manifestations of 22 cases, which had been described in various designations, and sorted out 7 characteristic features of this syndrome (Table 1) and reported under the name of "A New Syndrome." Falls and Schull[5] named it 'Hallermann-Streiff syndrome' for the first time as they reported 6 cases in 1960. In 1965, Hoefnagel and Benirschne included a finding of developmental retardation of motor and intelligence[6]. In 1970, Steel and Bass[7] analyzed 51 cases including 1 case of their own, and included 4 findings - frontal and occipital prominence, dehiscence and of the cranial sutures and opening of the fontanelles, high-arched palate and nystagmus. In 1975, Golomb and Poster[8] analyzed 80 cases including 3 of their own, classified them into major and minor features, and indicated the rate of positivity for each finding.

Nothing was known about the cause. It seemed that this syndrome did not have typical autosomal dominant or autosomal recessive traits, despite its sporadic manifestation[9]. In the beginning, there was an assertion that it could be a mutation that occurred in the period between the 5th week through the 7th week of the fetal age[4]. The evidence implicating contributing factors in Hallermann-Streiff syndrome is questionable. There were familial case reports, but all were atypical cases, pseudoprogeria-Hallermann-Streiff syndrome, or poorly documented[10].

The facial aspect showed a characteristic bird-like appearance, a pointed, small and thin nose with a small receded mandible as if it were a parrot. To put it concretely, the cranium was generally large, the frontal and parietal bones were prominent, ossification was inadequate and cranial suture closure was delayed. This patient showed a bird-like appearance, large cranium size in comparison with the face and small sized ramus and body of mandible with an obtuse gonial angle.

With respect to teeth, this patient seemed to keep primary teeth or have absence of permanent teeth, which might often show malocclusion. In this case, permanent teeth remained, and the medial and lateral incisors as well as the first molar of the mandible were already erupted. The medial incisor of the upper jaw was presently undergoing an eruption, was crowded and showed a state of malocclusion.

Hypotrichosis might often be common and would be limited to the cephalic region. Alopecia in the periphery of the suture line would be characteristic. In this case, hypotrichosis was observed in the head. Hairs in the eyelash and eyebrow were quite scanty. It was reported that complete absence or a roundabout flaw of hair epidermis was observed under an electron microscope and that fragile hairs were seen sporadically.

Cutaneous atrophy is common[11] and the skin becomes thin and shiny, which is often accompanied by telangiectasia[12]. In addition café-au-lait spot neurofibromatosis could also be manifested. In this case, skin atrophy and telangiectasia were witnessed, but other dermatologic findings could not be found.

The body would show proportionate dwarfism. Developmental failure of the skeleton1[3], delayed bone age[5], failure of the rib and clavicle, spina bifida and scoliosis were often seen[12]. In view of growth hormone increase following arginine infusion, it seemed that developmental failure could not be due to hormonal deficiency[14]. In this case, only proportionate dwarfism was observed.

With respect to ocular findings, bilateral microphthalmia and congenital cataract were most often manifested. This case also showed bilateral microphthalmia and aphakia due to spontaneous synchysis of congenital cataracts. Pinhole pupil and strabismus due to posterior synechiae were also observed. Other than that, there were reports that nystagmus, bluish sclera, defect of the ciliary body, glaucoma and IOP rise were observed[5, 8].

Francois[4] insisted that there was no mental disorder. However, 12 out of 80 cases reported by Golomb and Poster[8] showed a mental disorder, and some of these cases were thought to be associated with ocular abnormalities. Except for patients in the 15% category, intellectual ability was reported to be normal[12]. In this case, there was no consideration in the intellectual ability, but this child showed developmental retardation corresponding to DQ<75 in all items on DDST despite the current patient's age of 5 years and 6 months.

A patient with Hallermann-Streiff Syndrome would show complications due to anatomical structural abnormalities of the upper airway. For instance, feeding problems, respiratory embarrassment, recurrent respiratory infection, ob-

structive sleep apnea and cor pulmonale could be manifested. Endotracheal intubation could be difficult during anesthesia. Fortunately, endotracheal intubation during general anesthesia was not largely difficult in this case, but severe stridor was heard in her sleep and the stridor-induced respiratory difficulty were often experienced. Thus, we learned that there was an anatomical structural abnormality in the upper airway.

Differential diagnosis for progeria (Hutchison-Gilford type), cleidocranial dystosis, Franceschetti's mandibulofacial dystosis, and oculodentodigital dysplasia should be made.

Securing and maintaining a proper airway would be important in the treatment. Repeated respiratory infection would be quite common. Continuous positive airway pressure (CPAP) was reported to be effective for patients with obstructive sleep apnea[15]. In the meanwhile, protection and maintenance of visual acuity could be important, and surgery at an early age would be indicated for congenital cataracts[16]. Rhinoplasty, facial contouring plastic surgery and mandibular contouring surgery could be performed for the face[10].

Nothing was known about the prognosis. In the management of a patient with this syndrome, one should pay special attention to ophthalmologic, dental and upper respiratory problems.

한 글 요 약

무수정체안을 동반한 Hallermann-Streiff 증후군 1예

메리놀병원 소아청소년과

이명철 · 최임정 · 정진화

Hallermann-Streiff 증후군은 새 모양의 두개 기형, 치아 이상, 털 감소증, 피부위축, 선천성 백내장, 양측 소안구증, 비례적 왜소증 등의 7가지 특징을 동반한다. 소하악증이나 후두 연화증으로 인한 상기도 폐쇄가 문제가 되며, 이로 인한 수면 무호흡증이나, 호흡기 감염, 폐성심, 섭식 곤란 등이 자주 나타난다. 전 세계적으로도 150례 정도가 보고되어 있는 드문 질환으로써, 이에 저자들이 경험한 무수정체를 동반한 Hallermann-Strieff 증후군 1례를 보고하는 바이다.

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