

Hallermann-Streiff syndrome with bilateral microphthalmia, pupillary membranes and cataract absorption

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Dear Editor,

We write to present a case report of Hallermann-Streiff syndrome (HSS; oculo-mandibulo-dyscephaly with hypotrichosis) with persistent pupillary membranes and cataract absorption.

CASE REPORT

All data and photos were taken with oral and written consent from the guardians. The patient is a five-year old girl. She was a full-term baby with a normal vaginal delivery with a birth weight of 2735 g and birth height of 50 cm. She was the only baby in the family with no notable family history. Her mother's obstetric history revealed no record of systemic disease or drug administrations. Her mother noticed that her daughter had a poor vision five years ago. They visited local ophthalmic clinics two years ago and she was diagnosed as having congenital cataract. No medical intervention was given. Recently, they resorted to us showing typical dyscephalia of the HSS (bird face) (Figure 1A, 1B).

The patient was once diagnosed as proportionate short stature and growth retardation at three-year old. She has received growth hormone injection therapy for a year already. On admission to our facility, she was five years and four months old, and her height was 94 cm while the body weight was 12 kg. The head circumference was 48 cm and normal. Four

limbs were thin with normal muscle forces. She had a pointed nose and frontal bossing (Figure 1A, 1B). Her hairs were thin and brows absent. Her skin was so thin that the vessels were prominent. All nails were normal. She had a small mandible and irregular cone-shape teeth. Her ears were small and close to the mastoid process without listening problems. The patient had no cardiac or respiratory disorders, or mental retardation.

Entropion and trichiasis were observed on the upper lids. Epiblepharon of the upper palpebra were observed. Palpebral fissure was 24 mm in width and 8 mm in height. Bilateral inner canthus distance was 28 mm. Both eyes showed microphthalmia and microcornea [cornea diameter of right eye vertical/horizontal (V/H)=8.5/8.3 mm, left eye (V/H)=8.5/8.3 mm]. Involuntary horizontal nystagmus of both eyes as well as esotropia of near 30 prism diopter (PD) (Figure 1C) was noted. Vision acuity of both eyes was finger counting at 1 m. The intraocular pressures were normal.

Corneas and anterior chambers were clear. Pupillary membrane and miosis due to posterior synechiae were observed, making fundus examination impossible (Figure 2A). The pupil diameter was 1.8 mm of the left eye and 1.6 mm of the right eye. Depths of anterior chamber of both eyes were 24 mm. The opaque lenses were thin. Ultrasound test revealed mild vitreous opacity in the left eye and no retinal detachment in both eyes. No clear echoic reflection of posterior lens capsule was detected indicating lens absorption. A-scan showed short optic axis lengths, 16.45 mm for the right eye and 15.54 mm for the left eye. However, due to binocular nystagmus, errors do exist. The electroretinogram showed decreased amplitudes of a-wave and b-wave after both dark and light adaptation. And flash-vision evoked potential of both eyes was normal.

SURGICAL TREATMENT

Resection of the pupillary membranes and lensectomy were performed in both eyes under general anesthesia without problem in securing endotracheal intubation. During the surgery, lens absorption and thin lens covered with double-folded membrane was verified (Figure 2B, 2C). These thick membranes presumably were remnants of the anterior and posterior capsules. Intraocular lens implantation was not performed due to microphthalmia.

The first day after the surgery, the pupils were round, regular and equal (Figure 2D). The optic axis was clear and fundus was

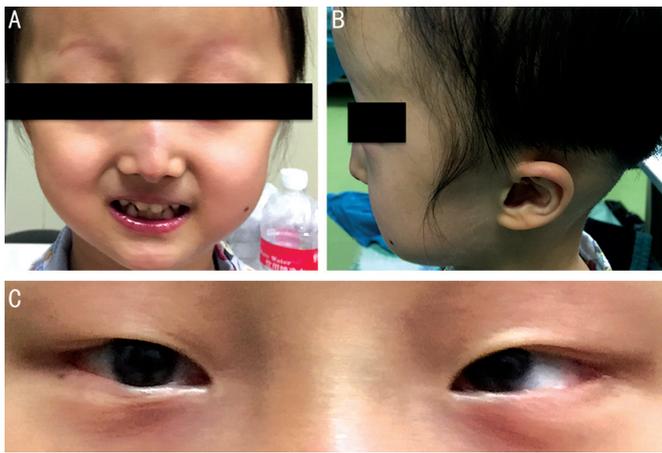


Figure 1 The typical “bird-face” with mandibular hypoplasia A: Frontal view; B: Profile view; C: Esotropia.

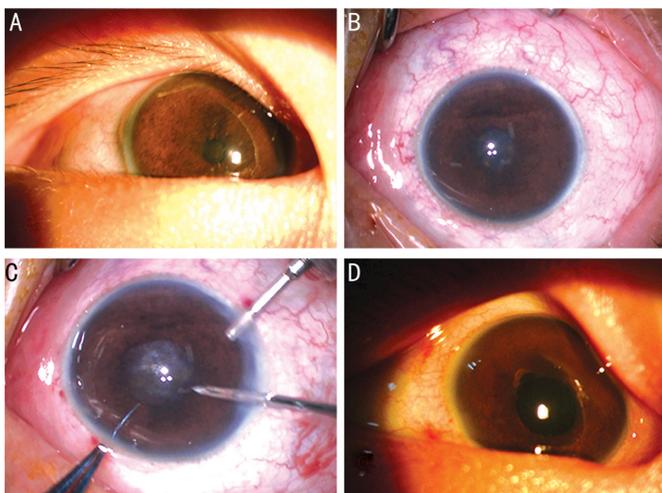


Figure 2 Images of the right eye A: Before the surgery, pupillary membrane and miosis were observed; B: After pupil dilation, miosis remained due to posterior synechiae; C: The lens was absorbed and replaced by opaque membranes; D: The first day after the surgery, the pupil was round and no uveitic reaction was noted.

normal. However, examinations of the periphery or funduscopy were not possible due to nystagmus and poor cooperation. Eyeglasses and amblyopia training were prescribed. The best corrected vision acuity of both eyes one week after the surgery were 0.02 (OD: +16.00DS/-2.50DC \times 170 $^\circ$; OS: +17.50DS/-2.75DC \times 20 $^\circ$) by Chinese Standard Logarithmic Visual Acuity Chart. The intraocular pressure was normal.

DISCUSSION

HSS is a rare disease. It is characterized by bird-like face, dental abnormalities, hypotrichosis, atrophy of skin, proportionate nanism, congenital cataracts and bilateral microphthalmia^[1]. HSS was initially noted in 1983, however the description was not complete. This syndrome was first described by Hallermann in 1948 and by Streiff in 1950. In 1953, it was first identified as an independent syndrome. Well-accepted diagnostic criteria was established by Francois^[2] in 1958. Virtually all cases are sporadic. There is no sex predilection^[1].

Over 150 cases of HSS have been reported worldwide since it was first described as HSS by Francois^[2-3] in 1958. The diagnostic criteria of this entity include dyscephalia and bird face (98%-99%), dental anomalies (80%-85%), proportionate nanism (45%-68%), hypotrichosis (80%-82%), atrophy of the skin (68%-70%), bilateral microphthalmia (78%-83%) and congenital cataract (81%-90%)^[1-2]. This patient had all seven features.

Ocular anomalies are a major problem, with the most common changes being microphthalmia and cataracts, presenting in 81%-90% of HSS patients^[1,3]. The lenses may have been absorbed spontaneously after birth, which sometimes occurs in the setting of HSS^[3-6]. Other ocular finding of HSS include microphthalmia (78%-83%), nystagmus (32%-45%), strabismus (33%-37%), blue sclera (22%-31%), sparse eyelashes and eyebrows (29%), fundus anomalies (18%-22%), conjunctival defects (11%), cornea abnormalities (9%-14%), down-slanting palpebral fissures (12%-13%) and so on^[1].

In this case, entropion and trichiasis, epiblepharon, nystagmus, esotropia, absence of brows, microphthalmia, microcornea, pupillary membrane, miosis, posterior synechiae and bilateral lens absorption were found. The diagnosis is easily made. Bilateral microphthalmia and congenital cataract are often manifested^[7]. Cataract absorption is the result of untreated congenital cataract. The pupillary membrane could be the result fibrinous reaction after cataract rupture and absorption.

For this case, poor vision was found in the first year of life. However, the treatment was delayed until the patient was five-year old, making vision rehabilitation more difficult. This makes it urgent for early ocular screening and treatment for HSS patients.

Besides, one of the most severe complications in HSS is respiratory embarrassment^[1]. Fortunately, this patient had no problem in tracheal intubation during the anesthesia and recovered from anesthesia without any respiratory complications. Close follow-up is needed.

CONCLUSION

In conclusion, we present a case of successful surgical repair for HSS in a five-year old patient who had ophthalmic features of pupillary membranes, posterior synechiae and lens absorption. In the setting of HSS, ocular examinations should be performed immediately when the patient was diagnosed. Cataract should be removed as early as possible to restore the vision functions.

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