Floating harbour syndrome with medial entropion: a rare association and brief review

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DESCRIPTION

A 6-year-old girl presented to us complaining of watering with associated frequent blinking in her both eyes since 2 years. There was history of delayed speech and delayed dentition. She was born out of consanguineous marriage; with no similar illness in any of her family members.

The child was alert, conscious with stable vitals. There was unusual dentition, large nose with an overhanging large columella, smooth philtrum and thin upper lips (*figure 1A*). On ophthalmological examination, best corrected visual acuity in both eyes was 6/6 and Intra ocular pressure (IOP) of 14 mm Hg. On further examination, bilateral epicanthal folds were seen along with medial entropion of both upper and lower eyelids with lashes touching the globes. The ocular surface was healthy; there was no epithelial defect or surface keratinisation (*figure 1B*). Fundus examination was within normal limits.

Routine blood investigations along with hormonal levels were within normal limits. Dental pantomogram showed absence of several teeth in lower jaw, indicating delayed dentition (*figure 2*). Diagnosis of floating harbour syndrome with bilateral medial entropion was made. She underwent bilateral Z-plasty for correction of medial entropion. Postoperative follow-up was unremarkable. A common differential diagnosis could be a medially localised epiblepharon of both lids. In case of an epiblepharon, on pulling the skin downwards, lashes should turn out with the lid still remains in apposition to the globe; however, this was not seen in our case.

The syndrome gets its name from the hospitals, Boston Floating Hospital and Harbour General Hospital where the first two cases were reported respectively by Pelletier and Feingold¹ and Leisti *et al.*² The condition is exceeding infrequent and until only around 65 cases of Floating Harbour Syndrome (FHS) have been reported.³ Most of the documented cases are sporadic in origin. Cases with family history of consanguinity have also been reported.⁴ A loss of the C-terminal portion of SRCAP protein, including the AT hook motifs have been implicated in the pathogenesis.³

Patients of FHS usually presents with delayed bone mineralisation and growth deficiency, often associated with mental retardation, skeletal and craniofacial abnormalities.³ Significant delay in bone age (−2SD) is a consistent feature in all patients. However, epiphyseal closure takes place at puberty with normal endocrine studies of growth hormone, somatomedin C and thyroid function.⁴ Speech delay

Figure 1 (A) Clinical picture of the patient showing triangular facies, prominent large nose with overhanging columella, smooth philtrum and thin upper lips. (B) Clinical picture showing bilateral upper and lower eyelid medial entropion with lashes touching the globe.

is a consistent feature of this syndrome. Striking facial features include broad and bulbous nose with a prominent nasal bridge and a wide columella with thin upper and lower lips; most recognisable during mid-childhood. The syndrome is also characterised by long lashes and dental malocclusion. There may be other associated features like clinodactyly of the fifth finger occasionally with a hypoplastic nail on that digit, accessory thumb, pulmonary stenosis and abnormal Electro Encephalogram (EEG).⁴

Nikkel *et al* gave the clinical features for coming to a diagnosis, which included mandatory features like: characteristic face and language delay, frequent features like: short stature, delayed bone age, skeletal anomalies and

Figure 2 Dental pantomogram showing the absence of multiple teeth.

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intellectual disability, recurrent problems like: eye anomalies, ear anomalies and dental disorders, infrequent problems like: genitourinary malformations, cardiac malformations, gastrointestinal features, seizures and hypothyroidism. The presence of both mandatory criteria along with the presence of any one frequent feature confirms the clinical diagnosis.⁵ Ocular disorders reported with this condition include hyperopia, strabismus and nystagmus; however, entropion of eyelids has never been reported.^{6 7} The pathophysiology of the medial entropion could be a localised overaction of medial fibres of orbicularis muscle. It may also possibly be a by chance association. We, here, document the first case of entropion in FHS.

Patient's perspective

Our child had eyelid problems with watering. We were relieved after the surgery as the doctor assured of no eye symptoms later and also a full body check-up assured us of no systemic complications.

Learning points

- Floating harbour syndrome is a rare entity. It is a triad of delayed bone age, abnormal speech and facial features.
- Although ocular features like strabismus, nystagmus and hyperopia have been previously documented with floating harbour syndrome; entropion can also be associated and is to be kept in mind when managing such a case.

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Disclaimer Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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REFERENCES

- 1 Pelletier G, Feingold M. Case report 1. In: Bergsma D, ed. *Syndrome identification*. White Plains, NY: National Foundation-March of Dimes, 1973: 8–9.
- 2 Leisti J, Hollister DW, Rimoin DL. Case report 2. In: Bergsma D, ed. *Syndrome identification*. White Plains, NY: National Foundation-March of Dimes, 1974: 305.
- 3 Messina G, Atterrato MT, Dimitri P. When chromatin organisation floats astray: the SRCAP gene and Floating-Harbor syndrome. *J Med Genet* 2016;53:793–7.
- 4 Patton MA, Hurst J, Donnai D, et al. Floating-Harbor syndrome. *J Med Genet* 1991;28:201–4.
- 5 Nikkel SM, Dauber A, de Munnik S, et al. The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. *Orphanet J Rare Dis* 2013;8:63.
- 6 Hood RL, Lines MA, Nikkel SM, et al. Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *Am J Hum Genet* 2012;90:308–13.
- 7 Le Goff C, Mahaut C, Bottani A, et al. Not all floating-harbor syndrome cases are due to mutations in exon 34 of SRCAP. *Hum Mutat* 2013;34:88–92.

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