Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/mjafi

Case Report

Hallermann Streiff syndrome: 'Bird faced' but not 'bird brained'

Maj Abhishek Mallick^{a,*}, Lt Col R.K. Singh^b, Col R.K. Thapar^c^a Resident (Pediatrics), Command Hospital (Central Command), Lucknow, India^b Classified Specialist (Pediatrics), Command Hospital (Central Command), Lucknow, India^c Senior Adviser (Pediatrics & Neonatology), Command Hospital (Central Command), Lucknow, India

ARTICLE INFO

Article history:

Received 5 July 2016

Accepted 7 February 2017

Available online 22 March 2017

Keywords:

Bird face

Hallermann Streiff syndrome

Hemihyperplasia

Hypotrichosis

Microphthalmia

Patent Ductus Arteriosus

Introduction

Hallermann Streiff syndrome or Francois dyscephaly syndrome is a rare condition with approximately 200 reported cases in the world. It is of unknown aetiology but it has been suggested that developmental disturbance of both ectoderm and mesoderm may be involved. Most cases are sporadic, with some mutations in gap junction alpha-1 protein (GJA1) gene reported.¹ Here we report a case which presented to us with stunted growth.

Case report

2-Year-old female patient, only issue of a non-consanguineous marriage was born by institutional term vaginal delivery, with

no antenatal risk factors, with a birth weight of 2.5 kg and normal perinatal transition. She had history of suck rest suck cycles and recurrent chest infections needing multiple hospitalizations since 2 months of age. 2D Echo at 6 months had revealed a 2.6 mm PDA. A device closure procedure was done at 1 year of age. She was then lost to follow up and admitted for evaluation at 2 years of age. Her weight was 8.3 kg (weight for age = -3SD), length - 71 cm (length for age < -3SD), weight/length = median, US/LS ratio was 1.4, head circumference - 45 cm (between -1 SD and -2SD). Head to toe examination revealed brachycephaly, hypotrichosis of scalp and eyebrows, bird like face with a beak shaped nose, micrognathia, bilateral microphthalmia and microcornea with iris coloboma (Fig. 1a and b), submucous cleft palate (Fig. 2b), maloccluded dentition. There were no digital anomalies but there was hyperplasia of the left leg from thigh below (Fig. 3a and b). Her ears, nails, skin and genitalia were normal. Her respiratory, cardiovascular, abdominal and neurological examination was normal. Developmental examination showed that child had just started to walk without support but not run. She was able to scribble when given a pencil. She started saying 'baba', 'mama' by the age of 14 months and now could string two words to indicate her needs, but her speech was unclear. She was able to point to body parts when named. Thus, gross motor development was around 15 months and the rest of the domains were around 18 months of age (DQ = 75%). The child was subjected to assessment by a psychologist who applied VSMS and the Social Quotient (SQ) was scored at 90. Radiograph skull showed no intracranial calcifications or evidence of synostosis (Fig. 2a). Ophthalmologic evaluation revealed bilateral iris and fundal colobomas, inferior corneal opacities in both eyes with no cataract or

* Corresponding author.

E-mail address: abhishekm046@yahoo.com (A. Mallick).<http://dx.doi.org/10.1016/j.mjafi.2017.02.005>

0377-1237/© 2017 Published by Elsevier B.V. on behalf of Director General, Armed Forces Medical Services.

Fig. 1 – (a) Full face frontal view showing 'bird face', microphthalmia and microcornea. (b) Profile view showing brachycephaly, beak shaped nose, micrognathia and hypotrichosis.

glaucoma. Bilateral Oto-acoustic emissions, TORCH screening, T3, T4 and TSH, rickets work up (radiograph wrist AP view, serum alkaline phosphatase, calcium, phosphorus) were normal. Karyotype was normal female XX. Ultrasonography of abdomen and Doppler study of the venous system of the left leg was normal. The parents were unwilling to undergo clinical exome sequencing for their child at this point of time.

Discussion

Falls et al coined the name of this syndrome after German ophthalmologist, Wilhelm Hallerman and Swiss ophthalmologist, Enrico Bernardo Streiff who identified this condition and distinguished it as a separate entity from mandibulo-facial dysostosis.² Francois (1958) gave the following seven essential features constituting this syndrome: dyscephalia and bird-face, dental anomalies, proportionate nanism (dwarfism), hypotrichosis, atrophy of the skin, bilateral microphthalmia and congenital cataract. The findings which distinguish this condition from other related mandibulofacial dysostoses, ectodermal aplasia and dysplasias are absence of anomalies of ears and palpebral

aperture, nails and extremities, musculo-arthritis anomalies and neuropsychological deficit.³

Our patient had significant dysmorphology as mentioned above with proportionate stunting. She had achieved her language milestones but speech was unclear (owing to the submucous cleft palate). Her gross motor milestones were delayed (most probably due to the recurrent chest infections) but she had good social skills. In our case, five of the seven essential features listed by Francois were present with the exception of cataract and atrophy of skin. The cataracts in the condition have been reported to undergo spontaneous rupture and absorption making it quite unique.⁴ None of the negative signs reported by Francois were present in our case. In addition, the child had features like congenital heart disease – PDA (reported in 5% of cases), submucous cleft palate (reported once)⁵ and hemihyperplasia (reported once).⁶

Since a genetic diagnosis of this syndrome is not available we would like to list the following differentials and discuss the basis of diagnosis in our case.

1. The closest differential in this case would be Oculo-Facio-Cardio-Dental syndrome where there are facial, ophthalmic, cardiac and dental abnormalities. This is a very rare,

Fig. 2 – (a) Radiograph skull showing lack of synostosis with presence of maloccluded dentition. (b) Submucous cleft palate.

Fig. 3 – (a) Digits show no anomalies. (b) Hemi hyperplasia of left leg with no lower digital anomalies.

X linked dominant condition and has been mapped to the BCOR gene of X chromosome. The typical facial gestalt of long narrow face, high nasal bridge, pointed nose with cartilages separated at the tip, was missing in our patient and she lacked a septal/valve defect which is an essential diagnostic feature. Also these patients are not usually growth stunted.

2. The next close differential would be Oculo-Dento-Digital-Dysplasia Syndrome. The characteristic features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares, with a narrow nasal bridge. The teeth may be small and carious. Typical eye findings include microphthalmia and microcornea. Digital anomalies like syndactyly or camptodactyly are characteristic. There may be neurological manifestations like ataxia, neurogenic bladder, seizures and spastic paraparesis with basal ganglia calcifications. They have characteristic GJA1 mutations. Our patient lacked the typical nasal features, had no digital anomalies or neurological manifestations.
3. The 'bird face syndrome' like, Seckel syndrome is characterized by the typical gestalt of bird face and severe microcephaly along with severe intellectual disability. Our patient lacked the latter two features.
4. Hutchinson Gilford Progeria Syndrome is characterized by a senile appearance, lipodystrophy and other ageing features. Our patient had no ageing features.
5. Cockayne syndrome is characterized by cutaneous photosensitivity, a progeroid appearance, progressive pigmentary retinopathy, sensorineural hearing loss. None of them were present in our patient.

Our patient had features of a rare syndrome and some unique findings rarely mentioned in literature. Also we have

strived to compare syndromes with similar features and tried to point out the essential differences in them. We would like to conclude by saying that although she has a bird like face, she has a good social quotient (not "bird brained") but lags behind in her milestones. Nutritional and occupational rehabilitation will go a long way in helping her achieve her milestones.

Conflicts of interest

The authors have none to declare.

REFERENCES

1. Abrams CK, Scherer SS. Gap junctions in inherited human disorders of the central nervous system. *Biochim Biophys Acta*. 2012;1818(8):2030–2047.
2. Falls HF, Schull WJ. Hallermann-Streiff syndrome, a dyscephaly with congenital cataracts and hypotrichosis. *Arch Ophthalmol (Chicago Ill: 1960)*. 1960;63:409–420.
3. 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(5):842–862.
4. Wolter JR, Jones DH. Spontaneous cataract absorption in Hallermann-Streiff syndrome. *Ophthalmologica*. 1965;150(6):401–408.
5. Spaepen A, Schrandt-Stumpel C, Fryns JP, et al. Hallermann-Streiff syndrome: clinical and psychological findings in children. Nosologic overlap with oculodentodigital dysplasia? *Am J Med Genet*. 1991;41(4):517–520.
6. Dogan DG, Karabiber H, Erhan MD, et al. Hallermann-Streiff syndrome with hemihypertrophy. *Genet Counsel (Geneva Switzerland)*. 2010;21(3):329–333.