Hallerman Streiff Francois Syndrome

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Abstract

18 Year old male child presented with abnormal facial features and operated for congenital cataract surgery with successful visual rehabilitation.

Introduction

Hallerman Streiff syndrome is a rare syndrome, which involves multiple congenital abnormalities affecting chiefly the head and face.

Case Report

18 year old male child presented with diminished vision and abnormal facial features (Fig. 1). Patient was operated for both eyes congenital cataract surgery at 5 months of age after which visual rehabilitation was done by aphakic glasses. At present patient is 6/60 both eyes with a 16 D sph lens in both eyes on refraction.

Examination of both eyes revealed that the child had sparse eyebrows and eyelashes (Fig. 2). There was a mild antimongoloid slant. Sclera appeared bluish. There was bilateral aphakia with a peripheral buttonhole iridectomy in the right eye (Fig. 3). Intraocular pressure was normal in both eyes. Fundus was within normal limits. There was no evidence of squint. Ultrasonography of both eyes revealed a normal posterior segment.

The boy had typical retrognathia and beak shaped nose. Skin of the face appeared atrophic. The mouth and tongue appeared small and palate was high arched. Examination of the oral cavity demonstrated 4 yellowish discoloured natal teeth in upper jaw and absence of teeth in lower jaw (Figs. 4,5). The child was short in stature (Fig. 6).

At present the child is being visually rehabilitated with aphakic contact lenses and on follow routine IOP measurements is being done. The patient has been screened for respiratory illnesses and at present he has no such illness.

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Fig. 1 : Abnormal facies

Fig. 2 : Sparse eyebrows and lashes
Hallerman Streiff Francois syndrome (oculomandibulofacial dyscephaly)

The first record of this disorder was made by Aubry in 1893. More than 150 cases have been reported till date. Virtually all cases are sporadic and thus there is no obvious pattern of inheritance.

The most likely hypothesis is that of a single mutant gene (dominant) with most
cases representing fresh mutations. Recently a defect of elastin and abnormal glycoprotein metabolism has been reported. Several pairs of affected siblings have also been reported, suggesting autosomal recessive inheritance.\(^1\) Jules Francois analysed 21 cases and reported 7 essential signs for what he regarded as a ‘new syndrome’ to be linked into the long chain of heredity of ectodermal dysplasias.

These signs are: 1. Dyscephalia and bird face; 2. dental abnormalities; 3. proportionate short stature; 4. hypotrichosis; 5. Atrophy of skin especially on nose; 6. bilateral microphthalmos and; 7. congenital cataract.\(^2\)

The characteristics feature in this patient include dyscephaly with bird like facies, hypoplastic mandible and beaked nose, proportionate short stature, hypotrichosis, congenital cataract (operated), dental anomalies and cutaneous atrophy. In contrast to the craniostenoses, delayed closure of the fontanelles and dehiscence of sutures associated with an odd shaped brachycephalic skull are often observed in patients with Hallermann Streiff syndrome. Other systemic abnormalities of potential importance include tracheomalacia and upper airway obstruction, which can lead to chronic respiratory embarrassment. Intellect is normal in most cases.\(^3\) The cause of Hallermann Streiff syndrome remains unclear. The lenses of patients with Hallermann Streiff syndrome are described as white and liquefied and often resorb spontaneously. Other ophthalmic findings that have been reported include blue sclera, nystagmus, strabismus, down slanting palpebral fissures, glaucoma, aniridia and sclerocornea. Posterior pole abnormalities including retinal folds, pigmentary degeneration and Coats disease have also been reported.

References

PULMONARY REHABILITATION FOR MANAGEMENT OF CHRONIC OBSTRUCTIVE PULMONARY DISEASE

A 61-year-old black woman with chronic obstructive pulmonary disease is referred for enrollment in a pulmonary rehabilitation programme. Pulmonary rehabilitation includes high-intensity exercise to improve muscle function and reduce dynamic hyperinflation of the lungs, as well as education to promote collaborative self-management strategies. Patients with unstable angina or recent myocardial infarction may not be good candidates for pulmonary rehabilitation.