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Hallermann-Streiff Syndrome – A Case Report

KEYWORDS: Hallermann Streiff Syndrome, genetic condition, chronic renal failure.

Dr. Prabhakar Shivshankar Jirwankar
Associate Professor, Department of Medicine, Government Medical College, Aurangabad

Dr. Shailaja Rao
Assistant Professor, Department of Medicine, Government Medical College, Aurangabad

ABSTRACT *Hallermann-Streiff syndrome is a rare genetic condition in which patients have characteristic facial features, visual abnormalities, tooth problems, short stature, and occasionally mental impairment. The distinctive facial features of Hallermann-Streiff syndrome include a very small head that is unusually wide with a prominent forehead, a small underdeveloped jaw, an unusually small mouth, and/or a characteristic beak shaped nose, small eyes and premature cataracts. Most patients of this syndrome present to the ophthalmologist for visual problems. We present a 22 year girl of Hallermann-Streiff syndrome with chronic renal failure.*

INTRODUCTION :

Hallermann-Streiff syndrome is a rare genetic condition in which patients have characteristic facial features, visual abnormalities, tooth problems, short stature, and occasionally mental impairment. The distinctive facial features of Hallermann-Streiff syndrome include a very small head that is unusually wide with a prominent forehead, a small underdeveloped jaw, an unusually small mouth, and/or a characteristic beak shaped nose, small eyes and premature cataracts[1]. Most patients of this syndrome present to the ophthalmologist for visual problems. We present a 22 year girl of Hallermann-Streiff syndrome with chronic renal failure.

CASE HISTORY :

A 22 years proportionate dwarf girl presented for breathlessness, decreased urine output and vomiting since one month. Patient had history of gradual loss of vision, due to bilateral premature cataracts, for which she underwent cataract surgery with IOL implantation two years back. She attained menarche at 14 years of age with irregular cycles. She was amenorrhic since one year. She is forth child, first three being male children of 30, 28, 24 years old, having normal growth and development. Both parents were normal. There was no history of infection to mother during pregnancy and had full term normal delivery.

Detail physical examination revealed short stature with 93 cm height and 11 kg weight. Upper to lower segment ratio was normal. Vital parameters were normal. Her head was small with broad forehead and parrot beak nose, small mouth, under developed maxilla and mandible. Her primary dentition was normal however secondary dentition revealed partial anodontia in the form of missing upper and lower anteriors and hypo plastic teeth. Her eye brows were sparse, skin was dry, rough and hyper pigmented.

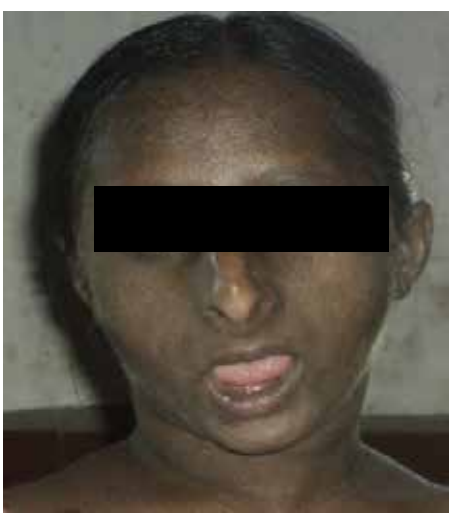

Figure – 1: Showing hypotrichosis, beaked nose and partial anodontia.

Patient's mental development appeared to be normal with average intelligence. She had poorly developed secondary sex characters. Her voice was low pitch with nasal twang. Systemic examination did not reveal any cardiac septal defects, respiratory infections, organomegaly or neurological deficit.

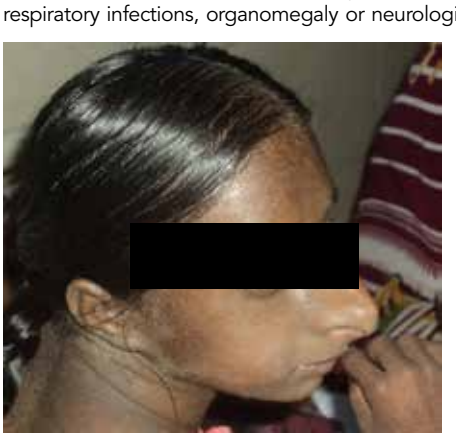

Figure – 2: Showing small head, sparse hairs , pinched nose and small mandible.

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Figure – 3: Showing short stature with normal upper and lower body segment ratio.

Her investigations revealed presence of Chronic Renal Failure with anemia.

Table No.1 : Showing investigations

Hb	6.3 gm/dl	Bl. Urea	137 mg/dl	Urine -R	Normal
TLC	10100/cmm	Sr.cret	7.4 mg/dl	T3,T4,TSH	Normal
P	56 %	Sr.Na	151 meq/l	2 D ECHO	Normal
L	34%	Sr.K	4.7 meq/l	Bone age	> 20 yrs
E	3%	Sr.Bili	0.7 mg/dl	NCCT brain	normal
M	7%	FBS	100 mg/dl	RBC	Normocytic, Normochromic
Platelets	54 lakh/cmm	PLBS	115 mg/dl		

USG abdomen revealed bilateral small size kidneys for her height (Rt. Kidney - 4.1 cm x 1.6 cm, Lt.Kidney 4.1 cm x 2.1 cm) with slightly raised cortical echogenicity . USG pelvic revealed uterus measuring 23mm x 16mm x 16mm. with midline echo, both ovaries appeared normal containing multiple follicles. Serum levels of GH, LH & FSH could not be done.

Considering typical facial features, short stature, pinched beak shaped nose, underdeveloped maxilla and mandible, hypotrichosis, partial anodontia and skin changes, we considered the diagnosis of Hallerman Streiff Syndrome with Chronic Renal Failure. Further studies may be required to correlate occurrence of CRF in individuals with this syndrome.

DISCUSSION:

Hallermann-Streiff syndrome, also known as Francois dyscephaly syndrome, Hallermann-Streiff-Francois syndrome, oculomandibulodyscephaly with hypotrichosis, and oculomandibulofacial syndrome[1].

Genetic Profile: Hallermann-Streiff syndrome is a rare genetic disorder. The specific gene responsible for Hallermann-Streiff syndrome has not yet been identified. Most cases of Hallermann-Streiff syndrome has sporadic occurrence. Some reports in the literature indicate that Hallermann-Streiff syndrome is inherited as a recessive condition.

Signs and Symptoms: Hallermann-Streiff syndrome affects the face, skull, hair, skin, eyes, teeth, and overall growth and development.

Face and Skull: The facial features of individuals with Hallermann-Streiff syndrome are distinctive. The face is small with a thin, tapering, pinched nose, and small chin. The head is small and unusually wide with a prominent forehead, a small underdeveloped jaw, and a small mouth[2]. Hairs are usually sparse, particularly that of the scalp, eyebrows, and lashes. Often there are no hairs around the front and sides of the head. The skin of the scalp is thin and taut, and scalp veins are prominent.

Eyes: Congenital cataracts are the most common eye disorder. Other eye problems may include nystagmus, squint, decreased visual clarity, and in some cases, blindness[3].

Teeth: Dental problems are very common. They may include underdevelopment of tooth enamel and cavities. There may be absence, malformation, and/or improper alignment of certain teeth.

Growth and development: Short stature is seen in about half of the individuals with Hallermann-Streiff syndrome. The average final height for females is about 150 cms and for males it is about 155 cms. Most individuals are of normal intelligence; however 15-30% of individuals with Hallermann-Streiff syndrome show some degree of mental impairment or slow development[4].

Other: A small number of individuals with Hallermann-Streiff syndrome have atrial or ventricular septal defects. Chronic Renal Failure has not been described in literature in patients of Hallerman Streiff Syndrome.

Prognosis: Individuals diagnosed with Hallermann-Streiff syndrome typically have normal intelligence and life-spans when complications of this disorder are properly managed. A major difficulty for individuals has visual problems which can often lead to blindness, despite surgery. Lung infections can be life-threatening and must be treated immediately. Breathing problems are another serious complication resulting from the abnormal skull formation that narrows the upper airway. Individuals with significant mental impairment may require life-long supervision.

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