BRIEF COMMUNICATIONS

AN UNUSUAL VARIANT OF HALLERMANN - STREIFF SYNDROME

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Hallermann-Streiff syndrome or mandibulo-oculo-dyscephaly is a condition in which patients have stunted growth, characteristic facial appearance with beaked nose, small mouth, irregular dentition and microphthalmia (Kirman and Bicknell, 1975). About a hundred cases are reported and the genetics of the condition is uncertain.

The cranial sutures have delayed ossification and fontanelles remain open for a long time and as a result brachycephaly occurs with frontal and parietal bossing and thin calvarium, facial bones are hypoplastic, small beaked nose with receding, hypoplastic mouth and chin. Low-set ears, thin and light hair, hypotrichosis of scalp, eyebrows and eye lashes, microphthalmia with congenital cataract (total or incomplete in 90% of cases), microstomia, high arched palate, hypoplasia and/or malimplantation of teeth and partial anodontia, micrognathia with hypoplasia of the rami and displacement of the temporo-mandibular joint can occur. Nystagmus and squint are common with stiff joints, thin skin and many visible blood vessels, stunted growth is common.

Rarely the patient may exhibit scaphocephaly, microcephaly, platybasia, shallow sella turcica, absence of mandibular condyles, downward slant of palpebral fissures, coloboma of optic disc, glaucoma and various chorioretinal pigment alteration, syndactyly, winged scapulae, lordosis, scoliosis, spina bifida, funnel chest, mental retardation (1/3 or less), hypogenitalism, cryptorchidism in the male.

These patients may have feeding and respiratory problems, which often causes death. The peculiar physiognomy and shortness of stature may impair their psychological adjustment, but ocular defects leading to blindness is major handicap.

From India only one case has so far been reported by Narayanan (1978).

Case Report

A 19 years old female brought with complaints of sleeplessness, wandering, talking irrelevently and continuously, refusing feeds since 15 days. She claimed great power and flexed her muscles to show her 'great strength' and said she could do any work.

Patient is the last of 4 sibs, the others

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being healthy. No history of consanguinity, mental illness, epilepsy or any chronic illness among other members of the family.

Born of a full term normal delivery, patient was very small in size for age. She attained neck control by 8 months, spoke few words by 2 years, started walking by 4 years and has had 15 secondary teeth. She is illiterate and earns a substantial amount by singing film songs in 6 different languages. Patient has to be helped in toilet activities. She attained menarche 1 year back with periods lasting 6-11 days every 1-2 months.

No significant past history was observed.

Premorbidly she was affectionate with poor intellectual interests. On mental status examination, patient was overtalkative with increased psychomotor activity, restlessness, singing and dancing, picking cues from the environment. She talked continuously and irrelevantly with pressure of speech and grandiose delusions, elated mood, poor abstraction, moderate mental retardation, poor judgement and absent insight.

**Clinical Examination**

Head circumference 45 cms, filtrum 1/2 cm, height 112 cms, Weight 15 kgs. The skull is brachycephalic with bossing of frontal bones, palpable anterior and posterior fontanelles, with prominent suture lines and a prominent occipital protuberance. The patient had microcephaly, the nose was thin and small, parrot shaped with a flattened bridge, micrognathia and microstomia with microphthalmia and no cataract. Except two central incisors in lower jaw and one left premolar in upper jaw, the patient had anodontia. Patient has a long neck with a receding jaw and low-set ears, thin and sparse hair, poorly built, no breast development, no axillary hair, poor growth of pubic hair, no syndactyly.

CVS, RS, PA, CNS were normal. Fundus was normal.

**Investigations**

Haematological and biochemical tests were normal. X-rays of the skull, A.P, and lateral views revealed microcephaly, multiple vermian bones in the occipital area, the sella is shallow and J shaped with almost complete absence of both maxillae and hardly any maxillary sinuses, frontal sinuses being rudimentary. The mandible is dysplastic and the angle
is more obtuse with two lower central incisors and one upper premolar.

Chromosomal Analysis

The cytogenetic studies revealed that the patient had an interstitial deletion of long arm of chromosome 10, with a karyotype of 46, XX del (10) (p ter → q 21.3 : : q 22.2 → q ter).

Dermatoglyphic Studies

Revealed a normal pattern, however, all dermal ridges were ulnar loops. The total ridge count was 137. The atd angle was 45° in both palms.

Psychodiagnostic Evaluation

Could not be done, as the patient was unco-operative. Intelligence quotient (I.Q.) was 30 on Binet-Kamat test.

Discussion

This case had psychoses, chromosomal anomaly, microcephaly, brachycephaly, fontanellae and sutures being palpable, micrognathia, microstomia, microphthalmia, poor development of secondary sexual characteristics, X-ray showing the absence of sinuses and a receding jaw.

The differential diagnoses include:

1. Cockayne's syndrome - where photophobia and light sensitive skin are prominent features with cataracts, coarse skin and mental retardation.

2. Seckel's syndrome - here extreme microcephaly (less than 7 standard deviation) with short stature and beak like nose is common.

3. Treacher's - Collins syndrome - External ear deformities are extreme and there is anti-mongoloid slant of eyes with absence of zygoma in the X-ray.

This case differs from the classical "Hallermann - Streiff" in having no opthalmological problems (a common finding in Hallermann - Streiff syndrome) and in having chromosomal anomaly, but many other features like mandibulo-oculo-dyscephaly were present. This case we believe could be an unusual variant of "Hallermann - Streiff" syndrome. Only further chromosomal studies especially with high resolution chromosome banding technique would reveal whether chromosomal anomaly is a feature of "Hallermann - Streiff" or these cases with such anomaly can justify for a
Separate syndrome. The presence of psychoses is for the first time being reported in this type of syndrome. The psychoses is of manic type, which is not a common occurrence in such cases.

Only a further perusal by other workers in identifying such cases and doing careful chromosomal study will reveal, whether this is a variant of "Hallermann – Streiff" and can qualify for a new syndrome.

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