HALLGREN'S SYNDROME

(A Case Report)

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Introduction

Hallgren's syndrome, a rare complex syndrome transmitted by a single autosomal recessive gene in both sexes with complete penetrance and pleiotropic effects, gives rise invariably to retinal pigmental dystrophy and congenital deafness, and in the majority of cases to vestibular and mental abnormalities [1,2]. A case of Hallgren's syndrome is reported here because of its rarity.

Case Report

A 32-year-old married male hailing from rural Tamil Nadu presented with night visual deficit and headache of two years duration. His cousin (maternal uncle's daughter) had a similar illness and also had mental subnormality. There was no past history of psychiatric illness. He could not pass any services examination and had been superseded. Unit report on AFMSF-10 was good.

General physical and systemic examination were within normal limits. He did not have nystagmus, ataxia or skeletal abnormalities. Ocular examination revealed a visual acuity of 6/6 in both eyes, orthophoria and no abnormality in the anterior segments. Fundus examination revealed clear media with normal optic discs and bright fovea. Equatorial retina showed early changes of pigment degeneration. There was no vascular attenuation. Intra-ocular tension was 14.6 mm Hg (Schiotz) both eyes. Lister's perimetry showed concentric constriction of about 25-30° to 3/330 white. Bjerrum's scotometry was within normal limits. The complaint of night visual difficulty with equatorial pigment degeneration and visual field constriction in an otherwise normal eye led to the diagnosis of an early pigmentary retinal dystrophy (retinitis pigmentosa). ENT examination with pure tone audiometry revealed congenital bilateral sensoryneural hearing loss (40-50 dB).

In the ward, the patient was noted to remain aloof and depressed which led to psychiatric referral. Patient complained of headache, forgetfulness, difficulty in falling asleep and early morning awakening. Mental status examination showed well kempt appearance, psychomotor retardation, extreme preoccupation with somatic symptoms, anxious and depressed mood with fleeting suicidal ideation, feeling of helplessness and hopelessness and ideas of reference. Appetite was reduced. He had initial insomnia with early morning awakening.

Investigation including hemogram, urinalysis, blood sugar (fasting and post-prandial), serum cholesterol, blood urea, serum creatinine, VDRL, skull radiography and CT scan (head) were within normal limits. Radiogram of the paranasal sinuses showed mucosal thickening of maxillary antra. Intelligence quotient by Bhatia's battery of performance test of intelligence was 91. With a diagnosis of major depression the patient was treated with antidepressants, anxiolytics, ECT (7) and psychotherapy with amelioration of his psychiatric symptoms.

Discussion

Pigmentary degenerations of the retina have been associated with congenital deafness ever since von Graefe reported their simultaneous occurrence in 1858, an observation confirmed by Liebreich (1861) who found this retinal degeneration in 5.8% of congenitally deaf persons in Berlin [3]. Several references to this combination are to be found in the literature which were summarised by Hallgren (1959) who investigated 177 patients belonging to 102 families and in a genetical-statistical study established a syndrome (Hallgren's syndrome) [4]. The incidence of this syndrome in Sweden was approximately 3 : 100,000 people. The clinical appearance of the primary retinal pigmentary dystrophy

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is characteristic of that seen in patients with retinitis pigmentosa and is associated with night blindness. All patients suffer from a congenital sensorineural deafness. More than 90% of the clinically deaf have a gait disturbance of vestibulo-cerebellar type, possibly due to labyrinthine disorder, since other signs of cerebellar deficiency are not generally found. There is a positive correlation between the degree of hearing loss and vestibulo-cerebellar ataxia. About 25% of patients show mental retardation and 25% show a psychosis. Nystagmus occurs in 10% of patients. Cataract usually develops before the age of 40 [1,3,5]. Skeletal anomalies (short stature, genu valgum, kyphosis, club foot) are occasional findings. Mortality among the patients is not increased whereas fertility is greatly reduced [6].

An interesting feature of this syndrome is that while the ocular and aural anomalies are inherited traits, the mental retardation and psychotic behaviour may be a nonspecific

response in some patients to a double sensory handicap [5]. While there is no treatment for the ocular and aural defects, the psychological disorders are amenable to therapy.

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