THE MULBERRY TUMOUR

(A Case Report)

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ABSTRACT

A case of tuberous sclerosis with the classical triad of adenoma sebaceum, mental retardation and epilepsy is reported in a 10 year old boy. Fundus examination revealed a typical "mulberry" tumour and CT scan showed multiple "brain - stones". Fundus examination as aid to early diagnosis is emphasized.

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KEY WORDS : Tuberous sclerosis; Phacomata; Mulberry tumour

Introduction

Tuberous sclerosis was first described by Bourneville in 1880 as a syndrome characterised by mental retardation, epilepsy and a vesiculo-papular eruption (adenoma sebaceum) on the nose, cheeks and forehead [1]. In England, in a general survey the incidence was reported as 1 in 300,000. In Mayo Clinic it was seen in 1 in 20,000 registered patients [2]. Retinal phacomata are seen in approximately half the cases but the classical "mulberry" tumour is unusually rare. We report one such case of a retinal lesion in a young boy.

CASE REPORT

A 10 years old male child was a product of a non-consanguineous marriage. The child started having tonic-clonic

seizures from 6 months of age which increased to 20-35 per day at 1 year of age, at present controlled at 1-2 seizures per day with anti-epileptics. A rash developed over the face at 6 months of age. There were no ocular complaints. There was no positive family history.

On examination, the child was aggressive. There was adenoma sebaceum on the face and ash leaf macules on trunk and buttocks. There was Grade II/VI ejection systolic murmur best heard over pulmonary area. IQ was calculated as 50 on Binet Kamat.

Ocular examination revealed visual acuity of 6/9 both eyes. Fundus examination of eyes revealed clear media, normal optic disc, vessels and macula. Fundus of right eye showed one well circumscribed oval lesion 1 DD in size and 1 DD infra temporal to the disc. Left eye revealed similar lesion infra temporal to the disc which was 'gelatinous' while another lesion supra-temporal to the disc was white, well circumscribed, "mulberry" calcified type, with small vessels on the surface.

Routine haematological investigations, renal function tests, USG abdomen and X-ray skull did not reveal any

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Fig. 1. Left eye showing nodular lesion above and flat lesion below the disc.

abnormalities. EEG was suggestive of a structural lesion of the fronto-parietal area. CT scan of brain showed widespread calcific areas - "brainstones".

Discussion

Seizures are the most common presenting feature of tuberous sclerosis starting as infantile "salaam" spasms later becoming grand mal in type occurring in 93% of affected children. Mental deficiency occurs in 60% and adenoma sebaceum in 80-90% of cases. The benign tumour nodules may be detected on skull X-ray or CT scan as circumscribed calcifications, "brain stones", in 60% cases by 10 years of age [2]. Other lesions described are shagreen patches, sub ungual fibromata, sclerotic areas in the skeletal system, rhabdomyomata of the heart, renal cysts, pulmonary fibrosis etc [7].

Inheritance is autosomal dominant, but upto 80% of cases may occur as new mutations. Approximately 75% of patients die before the third decade of life [3].

Retinal lesions occur in half the cases [4] and are bilateral in 15% [5]. Three types have been described. First is the flat, smooth, translucent lesions, second is the classical mulberry or salmon-egg appearance and a third

intermediate form. The lesions are retinal astrocytic hamartomas. The flat lesions occasionally evolve into the nodular form, but both are extremely slow growing and clinically silent. Visual prognosis is good.

The retinal tumours are reported as early as 6 months of age and may be the first objective signs of tuberous sclerosis in children with retardation and epilepsy. The mulberry tumours are elevated, usually 1/4 - 4 DD across, and multinodular, often occurring near or along the disc margin. They contain large blood vessels and calcareous deposits. Pathologically they consist of elongated astrocytes, with long processes and small oval nuclei arising from the ganglion cell layer of the retina.

Paediatricians are often confronted with the problem of detecting tuberous sclerosis in the infants presenting with infantile spasms. This is required for genetic counselling. Williams *et al* [2] found an average delay of approximately 3 years in confirmation of diagnosis and by then the parents would have had other children. They also found that in 25% cases an ophthalmic referral led to early diagnosis of tuberous sclerosis.

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