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General Medicine

DIPLOPIA AS A PRESENTING MANIFESTATION OF TUBEROUS SCLEROSIS

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ABSTRACT Tuberous sclerosis complex is a rare autosomal dominant genetic disorder caused by mutations in TSC1 and TSC2 characterized by benign tumours in various organs like brain, heart, skin, lung, liver usually presenting with dermatological features and seizures. Atypical presentations of ocular, renal, pulmonary features are not so uncommon .we report such a rare case with no other family members affected and presenting late with ocular symptoms like diplopia .we report this case because of the rarity of disease with atypical manifestations.

KEYWORDS : Autosomal dominant, Tuberous sclerosis, Tumours

INTRODUCTION

Tuberous sclerosis is a rare inherited neurocutaneous disorder with multi-organ involvement with varied expression in different people[1]. It usually presents in infancy or adolescence with seizures and dermatological features. Late presentation with ocular symptoms like diplopia as in our case is rare. Here we report such a rare case to highlight the unusual presentations.

CASE STUDY

18yrs old male who is studying intermediate 1st year came to hospital with chief complaint of doubling of vision since 20 days , which is sudden in onset on waking up from sleep more on looking towards right side associated with headache in frontal region, with earache in both ears with occasional tinnitus without any hearing loss ,vomiting , colour vision abnormalities. In the past, pt had a history of seizures at 7 months of age for which he took antiepileptics for 3yrs and consulted a dermatologist 2yrs back for lesions over forehead which subsided after some sort of intervention. The patient was born out of 2nd degree consanguineous marriage with no similar complaints in the family and developmental history revealing late to join the school at age of 5 and passed 10th class with an 8.8/10 GPA. On examination, the patient has multiple bilaterally symmetrical fibromatous lesions over face more on the malar region and roughened leathery shagreen patch over back and hypomelanotic lesion over face and his vital signs are normal. Nervous system examination revealed right 6th cranial nerve palsy with normal intelligence, Fundus reveals normal study. CECT brain and MRI brain showed evidence of b/l multiple subependymal small calcification s/o sub ependymal nodules and a well defined lobulated intra ventricular isodense mass near left foramen of Munro with dilatation of ventricle s/o sub ependymal giant cell astrocytoma. ECG, ECHO, USG abdomen, chest Xray, chest CT were normal with out any evidence of rhabdomyoma, angiomyolipoma, lymphangioleiomyo matosis; routine blood investigations like CBP, LFT, RFT, LIPID profile were in normal range. the patient was conservatively treated with mannitol and eye patching but with out much improvement and hence referred to a neurosurgeon for intraventricular shunt procedure after 1 week.

FIGURES AND IMAGES

IMAGE 1: clinical image showing adenoma sebaceum and shagreen patches.

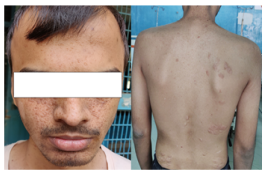
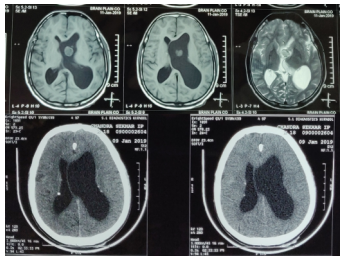


IMAGE 2: MRI brain showing subependymal giant cell astrocytomas, CT Brain showing subependymal nodules and left side hydrocephalous.



DISCUSSION

Tuberous sclerosis complex(TSC) is a very rare autosomal dominant genetic disorder caused by mutations in TSC1 and TSC2 genes[2]characterized by benign tumours in various organs like brain, heart, skin, lung, liver. Almost all patients with TSC have one or more skin lesions characteristic of disorder like angiofibromas, shagreen patches, ash leaf macules[3]. Brain lesions are the second most common characteristic features which include cortical tubers, Subependymal giant cell nodules and tumours presenting as epilepsy in childhood followed by cognitive and learning disabilities[4]. Other features which are characteristic of tuberous sclerosis include rhabdomyoma of heart, angiomyolipoma of kidneys, lymphangioleiomyomatosis of the lung.

Diagnosis of TSC is based up on genetic testing and clinical criteria. Genetic testing for TSC1 or TSC2 is sufficient to make a diagnosis but is not required for those who fulfil the clinical criteria for definite TSC[5]

Genetic diagnostic criteria	
Identification of either TSC1 or TSC2	
Clinical diagnostic criteria	
MAJOR	MINOR
Hypomelanotic macule	Confetti skin lesions
Angiofibroma	Intraoral fibroma
Ungual fibrosis	Dental enamel pits
Shagreen patch	Retinal achromatic patch
Retinal hamartoma	Multiple renal cysts
Cortical dysplasias	Non renal hamartomas
Subependymal nodules	

Subependymal gaint cell astrocytomas	
Cardiac rhabdomyoma	
Lymphangioleiomyomatosis	
Angiomyolipoma	
Definite TSC: 2 major or 1major + 2minor clinical features	
Possible TSC: Either 1 major or 2 or more minor clinical features.	

TSC is a progressive disorder and the expression of TSC varies significantly among patients and within families hence the individuals have different natural histories. Usually, patients present at infancy with seizures, later at childhood with dermatological features. Few were diagnosed at adulthood with renal and lung malignancies. In our patient the scenario was different where both the parents and no other sibling were not affected and the individual present late in the course of disease with ocular manifestations which is rare., which could be result of new mutations or mosaicism in one of the parent – hence both the parents were adviced to undergo complete body examination with genetic testing in rest of the family but was deferred by the parents as of now.

CONCLUSIONS

A similar case has been reported in a British medical journal by Tiago Maio et al[6], where a young woman presented with horizontal binocular diplopia and decreased visual acuity later diagnosed to have tuberous sclerosis. Hence a comprehensive knowledge of rare diseases like TSC with both typical and atypical features is much needed for a physician to diagnose and regular monitoring of the patient is adviced as it is a progressive disease. Genetic counselling for the rest of the family members must be made available to warn the 'at risk' relatives.

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