Original Research Paper

Volume - 12 | Issue - 11 | November - 2022 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Dermatology

A CASE REPORT OF TUBEROUS SCLEROSIS COMPLEX IN MOTHER AND 3 CHILDREN

Dr. Chandni Nair* Final year postgraduate, Sri Venkateswara Medical College, DR.NTR University of Health Sciences *Corresponding Author

Dr. T.Ajay Pradeep Final year postgraduate, Sri Venkateswara Medical College, DR.NTR University of Health Sciences

ABSTRACT Tuberous Sclerosis Complex (TSC) is a rare genetic disorder – with incidence approximately 1 in 5000 to 10,000 live births. It is a debilitating condition for both the patient and his family. It is characterised by the triad of epilepsy, low intelligence and adenoma sebaceum. Here we present one such case with classic clinical features.

KEYWORDS : Tuberous sclerosis complex, hamartoma

INTRODUCTION

Tuberous sclerosis complex (TSC) is a genetic disorder of hamartoma formation in many organs particularly the skin, brain, eye, kidney and heart.(2,4).Characteristic skin lesions include angiofibromas, shagreen patch, periungual fibromas and ash leaf macules seen in association with epilepsy and intellectual impairment. (1)
Inherited as an AD disorder caused by mutations in hamartin or tuberin genes Chromosome 9q34. Tsc 1 and 16p13 Tsc 2.(2)

CASE REPORT

Here we describe three cases of patients belonging to the same family, a mother aged 34 years and her son aged 9 years and youngest daughter aged 4 months. History of similar facial lesions and intellectual disability with multiple episodes of seizures in eldest daughter who didn't accompany them to the hospital.

The mother presented with facial angiofibromas which were firm, discrete, red brown telangiectatic papules 1-3 mm in diameter, symmetrically extending from the nasolabial folds to chin with relative sparing of upper lip and lateral face, which she developed 12 years back. She also has a shagreen patch in the lumbosacral region measuring 5cm x 6cm since birth and multiple hypopigmented macules over upper limbs and trunk. She has history of single episode of seizure at 4 years of age. MRI brain showed subependymal nodules along bilateral lateral ventricles and cortical tubers at bilateral frontal cortex and subcortical white matter. USG abdomen showed prominent renal pelvis.

Her son has similar facial lesions since 1 year and multiple hypopigmented macules over the back since birth . He had 2 episodes of seizures 3 years back.

Her 4 month old daughter has multiple hypopigmented macules over both hands and trunk. She has multiple intracardiac rhabdomyomas detected on paediatric echocardiography.

Fig 1-facial angiofibroma in mother

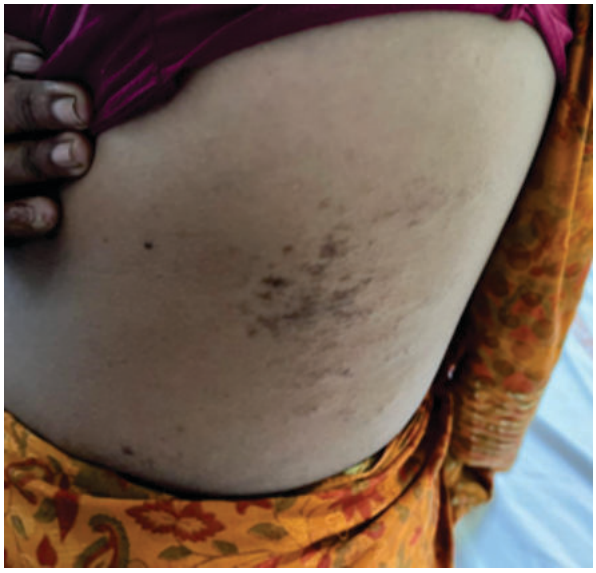

Fig 2-shagreen patch

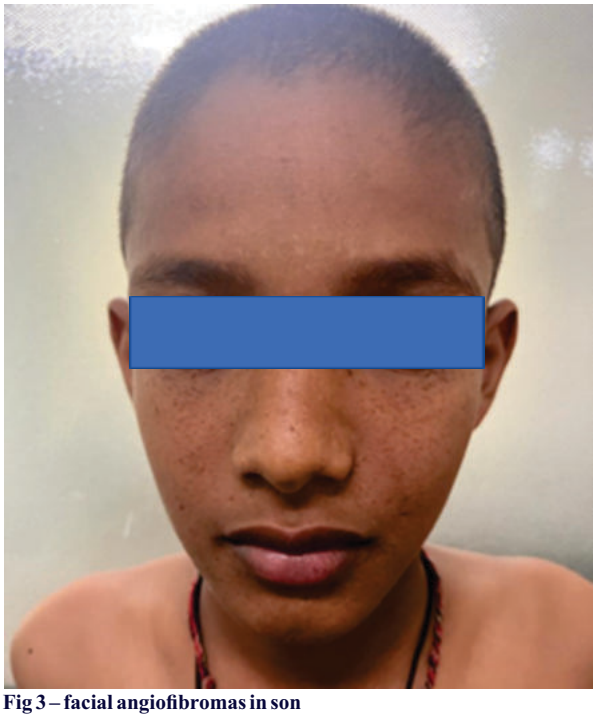

Fig 3 – facial angiofibromas in son

DISCUSSION

Major features:
- Facial Angiofibroma/ Forehead plaque
- Non Traumatic Ungual/periungual fibroma
- Hypomelanotic macules (>3)
- Shagreen patch (Connective tissue naevus)
- Multiple retinal nodular hamartoma
- Cortical tuber
- Subependymal nodules
- Subependymal Giant Cell Astrocytoma
- Cardiac Rhabdomyoma – single/multiple
- Lymphangiomyomatosis
- Renal Angiomyolipoma

Minor features
- Multiple randomly distributed pits in dental enamel
- Hamartomatous rectal polyps
- Bone cysts
- Cerebral white matter migration lines
- Gingival fibromas
- Non renal hamartomas
- Retinal achromic patch
- Confetti skin lesions
- Multiple renal cysts

Diagnostic criteria
Definite TSC: 2 Major/ 1 Major + 2 Minor
Probable TSC: 1 Major + 1 Minor
Possible TSC: 1 Major / ≥ 2 Minor

Investigations
Histopathology :
Angiofibroma: Hyperplastic blood vessels and sebaceous glands of immature hair follicles.

Periungual fibroma: Distal part of fibroma has loose collagen and many blood vessels and the large proximal part has dense collagen with fewer capillaries.

Radiological features:
MRI Brain: Cortical tubers, radial migration bands, Subependymal nodules/ Giant Cell Astrocytoma.

Chest: Lymphangiomyomatosis, Chylothorax, Rhabdomyomas(5)

Kidneys: Angiomyolipomas

Bones: Cysts.

Management:
Cosmetic treatment for angiofibroma: Pulsed dye vascular laser – reduces redness; systemic Rapamycin for visceral tumor and neurological complications including epilepsy.

Epilepsy is treated symptomatically ; neurosurgery is considered for non responsive cases.

Family screening and genetic counselling are most important modalities for prevention.(3)

CONCLUSION
Clinical suspicion is the key for diagnosis.Genetic screening and radiological follow up forms an integral part in its management.
This case is presented because of its rarity and presence of numerous clinical and radiological classical findings in our patient.

CONFLICT OF INTEREST: None

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