CASE NOTES

TUBEROUS SCLEROSIS

(A case report)

By

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Cases of tuberous sclerosis (Syn: Bournville's Disease) are not frequently reported in our country. It is comparatively a rare disease which is of autosomal dominant inheritence.

Tuberous sclerosis is a term applied to nodules of potato like appearance and consistency, that are found distributed over the cortex of the cerebral hemispheres. It is the key stone of an incompletely dominant trait characterized by widespread involvement of the skin and internal organs. The disease commonly occurs in children. It affects both sexes equally. Vogts described a clinical syndrome of a triad i. e., adenoma sebaceum, epilepsy and mental deficiency which is found in tuberous sclerosis. This triad is known as epiloia. Most commonly the mental defects and epilepsy are noted in infancy or early childhood, whereas the adenoma sebaceum develops between the fourth and ninth years. The convulsive states may appear in any of the familier epileptic forms such as petitmal, grandmal, jacksonian or psychic equivalent.

Frequently there are other systemic disorders in the form of cysts and tumours found in the kidneys and / or heart. Most patients who are affected with tuberous! sclerosis die before the age of 20 years from epilepsy, intercurrent infection and cardiac or renal tumors.

CASE REPORT

N. D. a 12 years old Hindu female attended the dermatologic out patient department of S. M. S. Hospital, Jaipur on 5-6-64 with the complaint of painless, papular eruptions over her face. Duration of this papular growth was about 1½ years.

Past History :—Patient had been much aggressive and quarrelsome since her early childhood. She never showed interest in reading or writing. About 5 years back she fell down from a height of 15 feet and became unconscious. On the same day patient was admitted in S. M. S. Hospital in comatose state and regained consciousness next day. She remained in the hospital for 7 days. At the time of discharge from the hospital she was perfectly well. After 3 months of this illness,

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she developed high fever about 106°F, which remained for about 6 days. On the 3rd day of fever she developed rash of small-pox. During the febrile period she was delirious and had convulsions. The treatment given for this illness was chloroform inhalation, icecap, some injections and tablets, the details of which is not known to the patient's relation. During the convalescence period of small-pox she suddenly developed madness. She started abusing and beating her relation without any reason, tearing her clothes and running from bed. For this illness she was admitted in mental hospital of Jaipur as a case of Behaviour disorder and was given electro-convulsive therapy along with largactil tablets. During her stay in mental hospital her blood K. T. and V. D. R. L. tests were found positive. For this she was given a course of PAM injectious. K. T. of parents was negative. There was moderate improvement in her behaviour after her stay of 1 year in mental hospital.

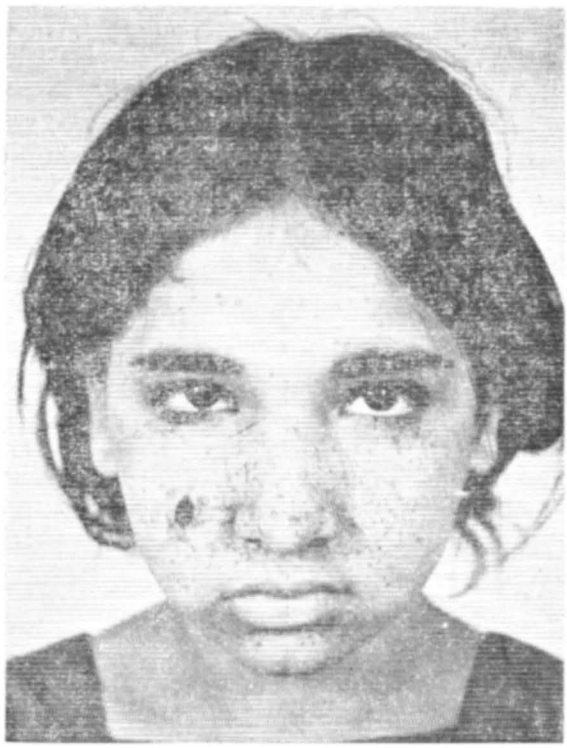

Fig. 1. Classical distribution of the papules of adenoma sebaceum.

History of present illness:—A papular growth appeared over face 1½ years back. Papules have been increasing in number since then. The earliest lesions were seen in nasolabial folds. There have been no complaint of itching, irritation or pain of any sort in these papules. There is no history of epilepsy. Her parents revealed that the mental illness which started about 5 years back is still containued.

Family History:—Not contributory.

Personal History:—Patient was unable to continue her studies because she could not qualify even for primary standards.

Physical Findings:—

General Examination:—Patient of sthenic built and well nourished. She laughed without any reason. She was rather of shy nature. Her intelligence was subnormal for her age. She was very slow in responding to even simple orders.

Skin Lesions:—Papular lesions of brownish red colour were present over the cheeks, forehead, chin and lowerlip. There was great confluence of lesions in nasolabial folds. In other places they were discrete. No capillary telangiectasia were seen. The arrangement of lesions was bilateral and symmetrical. The temporal regions and forehead showed a few widely scattered papules. Lower eyelids were also affected and revealed some papulas near the outer canthus. The upper lip was singularly free of lesions. The individual papulas were firm in consistency. The size of the papules varied from millet to that of a lentil seed. A few lesions on the left side of face near naso-labial folds showed definite tendency towards pedunculation.

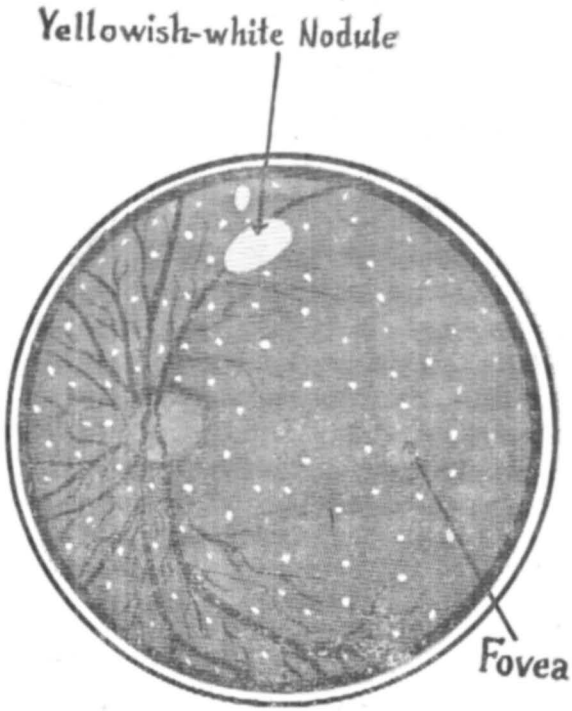

Fig. 2. Diagrammatic representation of the fundus changes in Left eye.

Some vitiligeneous areas were present over both the arms and the lumbosacral region. A big patch of vitiligo was present in left inguinal region extending right up to labia majora. A few hair present in this patch were also white. There were neither any nevoid lesions nor any fibrous hyperplasias.

Systemic Examination :—No tumour or cyst could be detected in internal organs like kidenys or heart. Nervous system examination revealed no abnormality.

Fig. 3. "Cotton ball" Calcification in the Skiagram of skull.

Fundoscopy :—A nodule of size about 6 mm. by 3 mm, was seen in supero-temporal quadrant of left fundus. It was about two disc diameters away from the disc, and was homogeneous and avascular in appearance. It was yellowish white in colour with distinct margins. The underlying retinal vessels were obscured. Another small nodule of size 3 mm x 2 mm was seen just above and nasal to the previous one. Fine, discrete colloid bodies were seen throughout the fundus, more so in between the superior and inferior temporal vessels and around the macula. A few of these small bodies were also seen on the nasal side of the disc. Macula and foveal reflex were normal.

Investigations:—

1.	Urine and stool.	N. A. D.	
2.	Haemogram.	Hb	11.5 gm%
		T. R. B. C.	4.1 million/c. m. m.
		T. L. C.	5.900/c. mm.
		D. L. C.	Polymorphs 64%
			Lymphocytes 34%
			Eosinophils 12%
			E. S. R. 10 mm / 1 Hour.

3. Blood V. D. R. L. Negative.
4. Screening chest—N. A. D. in heart and lungs.
5. Flat plate of abdomen—No abnormality detected in the region of kidneys, ureters and bladder.
6. Skiagram skull—" Cotton ball " calcification present above the Sella and sorrounding it.

Histopathological Examination of the biopsy material taken from papular lesion:—There was an increase in the sub-epithelial fibrous connective tissue with thinning of the overlying epithelium at places. There was prominence of hair follicles and hyperplasia of their lining epithelium. No hyperplasia of the sebaceous glands could be revealed. Other appendanges of skin were normal.

DISCUSSION

Of three cardinal features of tuberous sclerosis i. e. adenoma sebaceum, epilepsy and mental deficiency, our case had only two viz. adenoma sebaceum and mental deficiency. The symptoms of mental deficiency were present from her early childhood, but because the lesions of adenoma sebaceum did not appear until 1½ years back her diagnosis of Tuberous sclerosis was missed by a number of clinicians. Diagnosis made by Physicians about 4 years back was behaviour disorder. Her skiagram skull at that time was normal. Clinicians referred her to the child guidance bureau of Delhi. There also she could not improve much. But when she approachedus for consultation the lesions of adenoma sebaceum were quite apparent.

The typical distribution of the lesions on face with history of mental deficiency left little doubt in diagnosing the case as that of tuberous sclerosis. The discovery of " Cotton ball " calcification in roentgenogram of skull and the flat retinal tumours (described by van der Hoeve) in fundus examination further helped us in clinching the diagnosis, which was later confirmed by histopathological examination.

From the above discussion we come to the conclusion that while in most cases the above mentioned triad of " Epiloia " is well developed, abortive forms of tuberous sclerosis are not uncommon as in our case. Any phase of the triad may be poorly developed or absent, for instance our case had no epileptic fits.

Among the other disturbances found in cases of Tuberous sclerosis our patient had widely distributed vitiligenous areas. The lesions present over the legs were of the nature of pale freckles, while the other patches present in upper part of left thigh and lumbo-sacral region were milky white in colour.

Presence of moderate eosinophlia however, could not be explained.

SUMMARY

A typical case of tuberous sclerosis is reported. Out of three symptoms of "Epiloia" i. e. adenoma sebaceum, mental deficiency and epilepsy, our case had only the first two. Associated with these lesions, patient had pigmentary disturbance in the form of vitiligenous patches and areas of freckling.

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