DIDACTIC IMAGES

RETINAL FINDINGS IN TUBEROUS SCLEROSIS SYNDROME (TSS)

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Tuberous sclerosis syndrome (TSS) is an autosomal dominant multisystem disease characterized by hamartomatous growths in the brain, skin, kidneys, eyes, and heart. Mutations in the genes TSC1 and TSC2 account for the majority of cases.

Clinical features are classified into major (facial angiofibroma or forehead plaque, ungual fibroma, hypomelanotic macules, shagreen patch, multiple retinal nodular hamartomas, cortical tuber, subependymal nodule or giant cell astrocytoma, cardiac rhabdomyosarcoma,

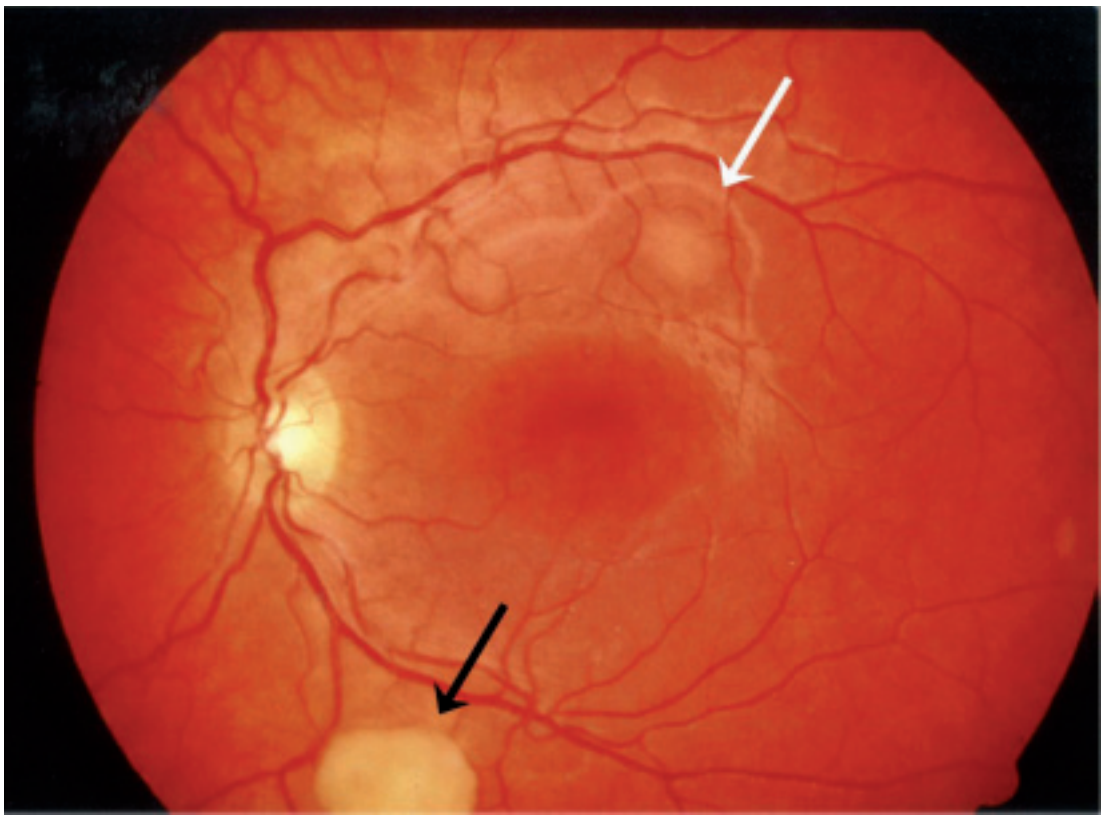


Figure: Two morphological types of retinal hamartomas in TSS:

1. A flat smooth, noncalcified, translucent lesion (white arrow)
2. An elevated, multinodular, calcified, opaque, lesion resembling mulberries (black arrow)

lymphangiomyomatosis and renal angiomyolipoma) and minor features (dental enamel pits, hamartomatous rectal polyps, bone cysts, cerebral white matter migration lines, gingival fibromas, nonrenal hamartomas, retinal achromatic patch, white skin macules and multiple renal cysts). Two major features or one major plus two minor features constitute a definite diagnosis of TSS (1,2). Ophthalmic features associated with TSS can be divided into retinal and non-retinal. Retinal hamartomas are one of the major diagnostic criteria for TSS. There are three basic morphological types: A flat smooth, noncalcified ,translucent lesion, an elevated, multinodular, calcified, opaque, lesion resembling mulberries and a lesion with the features of both (3).

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