

CLINICAL QUIZ

Left Facial Tumour. What Is It?

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Figure 1: Left facial tumour protruding from the orbital cavity displacing the eyeball downwards (eyeball not seen in picture)

QUESTION

A 30-year Malay gentleman (South-East Asian) ethnicity has been suffering from a left facial tumour, progressively increasing in size since birth. The tumour exits from the left orbital cavity and has displaced his left eye globe inferior posteriorly (Figure 1). Left eye vision has been progressively deteriorating in his left eye with the ability to only perceive bright light. He is of a short stature with multiple facial nodules. Spot the diagnosis and what other symptoms the patient might exhibit?

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ANSWER

The diagnosis is left Plexiform Neurofibromata, easily mistaken with an optic glioma. The tumour has protruded through his left orbital socket over his left cheek in a progressive manner, with multiple neurofibromatosis nodules throughout his torso and four limbs. The overlying skin is non-tender with normal tactile sensation. Plexiform Neurofibromata is a subset of neurofibromatosis type 1. Other symptoms include café-au-lait spots, axillary or inguinal freckles, Lisch nodules (pigmented hamartomatous nodules in the iris), sphenoid dysplasia with a strong genetic preponderance. There is no cure, a routine follow-up to monitor complications is recommended. Cosmetic tumour resection can be considered but they have high rates of recurrence.