Neurofibromatosis type 2.

Evans DG.

1 University Department of Genomic Medicine, University of Manchester, St Mary's Hospital, Manchester, UK. Electronic address: gareth.evans@cmft.nhs.uk.

Type 2 neurofibromatosis (NF2) is an autosomal dominant disorder caused by mutations in the NF2 tumor suppressor gene NF2 on chromosome 22. Around 1 in 33000 people are born with an NF2 mutation although more than one-third of the 60% of de novo cases are not conceived with the mutation but this develops later in embryogenesis (mosaics). NF2 has a substantial effect on life expectancy and individuals with a constitutional truncating mutation have the worst prognosis. The vast majority of people with NF2 will develop bilateral vestibular schwannomas with many developing schwannomas on other cranial, spinal and peripheral nerves. Cranial and spinal meningiomas and intraspinal low grade indolent ependymomas are the other major tumor features. Cutaneous features can be subtle with only 70% having evidence of intracutaneous plaque-like schwannomas or subcutaneous lesions on peripheral nerves. Café-au-lait patches are more frequent than in the general population but in only around 1% will meet NIH criteria for NF1.

KEYWORDS: NF2; café-au-lait; cataract; ependymoma; hamartoma; meningioma; schwannoma

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