



The Molecular Biology of Neurofibromatosis Type 1 (Paperback)

By Meena Upadhyaya

Morgan Claypool Publishers, United States, 2014. Paperback. Condition: New. Language: English . Brand New Book ***** Print on Demand *****.Neurofibromatosis type 1 (NF1) is a common autosomal dominantly inherited, tumour predisposition syndrome affecting 1/3,000-4,000 individuals worldwide. This inherited disorder results from the mutational inactivation of the NF1 gene on human chromosome 17. The NF1 gene contains 61 exons that give rise to 12kb mRNA encoding neurofibromin. The 327kDa (2,818 amino acid) neurofibromin protein is expressed in most tissues and has a number of alternative isoforms. Neurofibromin is a tumour suppressor protein and down-regulates cellular Ras. Increased active Ras-GTP levels also stimulate the important PI3K/AKT/mTOR signalling pathway that protects cells from apoptosis. The major clinical features of NF1 include multiple cafe-au-lait macules, skinfold freckles, iris Lisch nodules, and neurofibromas. The diagnostic criteria for clinical diagnosis have been well established. However, there are a small number of cases in which the diagnosis is not certain. The germline mutation rate for the NF1 gene is 10-fold higher than that observed for most other inherited diseases. Using a combination of different techniques, almost 95 of germline mutations can be detected. To date, only two firm genotype phenotype correlations have been reported. NF1 phenotype exhibits...



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