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Cancer Prone Disease Section

Short Communication

Schwannomatosis

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Identity

Note: A third major form of neurofibromatosis.
Inheritance: Up to 90% of schwannomatosis are isolated cases. An annual incidence of newly identified cases was estimated to be approximately 1 in 1,700,000. Inheritance in familial cases is autosomal dominant with incomplete penetrance.

Clinics

Clinical spectrum of schwannomatosis has similarity to neurofibromatosis type 2 (NF2) and overlap to some extent with that of NF2. Both disorders share the predisposition to multiple schwannomas.

Phenotype and clinics

Critetia for definite diagnosis: two or more pathologically sampled schwannomas and lack of evidence of vestibular nerve tumor on magnetic resonance imaging performed after age 18 years.
Criteria for presumptive diagnosis: two or more pathologically ascertained schwannomas without symptoms of eighth nerve dysfunction at age above 30 years or two or more pathologically sampled schwannomas in an anatomically limited distribution without symptoms of eighth nerve dysfunction at any age

Neoplastic risk

Benign schwannomas of peripheral nerve.

Treatment

Surgical resection upon indication for pain and neurological symptoms. Electromyographical or electrohpysiological monitoring can be used to minimize the risk of iatrogenic injery to the nerve or spinal cord during surgery.

Prognosis

Schwannomatosis-associated tumors are basically of benign nature. Surgical outcome depends on anatomical localization and size of the tumor. No reduction in life-span expectation.

Cytogenetics

Inborn condition

No special feature.

Genes involved and Proteins

Note: The genetic cause for schwannomatosis has not yet be identified. Linkage analysis has exclude the NF2 gene region and located the responsible locus to a 5 mega-basepair interval proximal to the NF2 gene on chromosome 22.

Mutations

Though alterations of the NF2 gene have been found in schwannomatosis-associated tumors, none of them has ever been found in any non-tumor tissues such as peripheral leukocytes of the patients.
Somatic: Somatic mutations and allele loss of the NF2 gene have been found in schwannomatosis-associated schwannomas.

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