International Journal of Science and Research (IJSR)
ISSN (Online): 2319-7064
Index Copernicus Value (2013): 6.14 | Impact Factor (2014): 5.611

Neurofibroma

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Abstract: ***Neurofibromatosis is a genetic disorder that causes tumors to form on nerve tissue. These tumors can develop anywhere in your nervous system, including your brain, spinal cord and nerves.***

Keywords: Nerve Axons Heringbone Pattern

1. Introduction

Neurofibromatosis is usually diagnosed in childhood or early adulthood. The tumors are usually benign, but sometimes can become malignant. Symptoms are often mild. However, complications of neurofibromatosis can include hearing loss, learning impairment, heart and blood vessel (cardiovascular) problems, loss of vision, and severe pain.

It is also called as von Recklinghausens disease of skin. It is autosomal dominant trait which arises from connective tissue sheath of Schwann cells and axons. NF1is characterised by the development of multiple café-au-lait spots, inguinal/axillary freckling and multiple neurofibromas

Symptoms usually appear during childhood and may become more pronounced during puberty, pregnancy, or when hormonal changes take place. Range and severity of symptoms can vary greatly among affected individuals even between family members.

Neurofibromas are diagnosed when two of the following clinical features are present:

- Six or more café-au-lait spots, 0.5cm diameter before puberty, or1.5cm in adults
- Two or more neurofibromas of any type or one plexiform neurofibroma Freckling under the arms or in the groin
- Benign tumour of the optic nerve (glioma)
- Two or more Lisch nodules (iris hamartomas)

A distinctive osseous lesion such as a sphenoid dysplasia or thinning of the long bone cortex with or without psuedoarthrosis

Additional but not diagnostic features:

- Precocious puberty or delayed sexual development may occur
- About 50% have specific learning disabilities in reading, spelling mathematics
- Growth may be reduced
- Macrocephaly
- Scoliosis
- Hypertension
- Epilepsy

Genetics

NF1 is caused by mutations in the NF1 gene that encodes a protein called neurofibromin, which functions as a tumour suppressor. Many different mutations in the NF1 gene have been identified in individuals with the condition.

Prevalence •

NF1 affects about 1 in 3000 people. There is a wide range of severity of symptoms. Many people with the condition will only be affected mildly. For most people, NF1 does not significantly affect their health but for a few, NF1 can cause major health problems at certain stages of their lives.

Investigations •

Genetic testing is not needed to diagnose the condition after birth because most people with NF1 will have enough signs of the condition by age 5 years for a specialist to diagnose them with confidence. Genetic testing for NF1 is not widely available and is currently expensive, but it can be helpfull in some situations, such as where prenatal diagnosis is requested. Prenatal genetic testing can be done where one of the parents is affected and wants to know if the fetus is affected, provided the specific NF1 mutation in the affected parent has been identified.

Management •

An annual review by GP for complications of the condition and for management advice (eg referral to plastic surgeon) should be undertaken. •Be aware that:

Neurofibromas can cause cosmetic problems and wrap around or penetrate the nerves causing pain. There is about a 5% increase in risk for various cancers, including brain tumour. Sometimes plexiform neurofibromas and, very rarely, simple neurofibromas can become malignant.

Hypertension is more common in NF1 patients than in the general population. At least annua l blood pressure should be undertaken on all individuals with this condition. If hypertension is identified, then investigations for a secondary cause such as renal artery stenosis and phaeochromocytoma should be undertaken.

There is also an increased rate of scoliosis in NF1. This should be looked for and there should be a low threshold for referral to an orthopaedic surgeon for investigation and

management. As with scoliosis in other conditions, it most commonly presents and progresses around the time of puberty. • Areas of surveillance should include:

Ophthalmology for optic gliomas; growth of these is rare over 10 years of age. Education, as specific learning disabilities in reading, spelling or mathematics may be present. Children also may have short attention span, low muscle tone, reduced co-ordination and emotional immaturity.

2. Conclusion

The concept of over viewing neurofibroma is to create awareness among the general public about this particular tumor of nerve cells and it is still an interesting topic to be discussed and delivered to upcoming medic0 pathological students.

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