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Case Report

Multisystemic Masses in a Child with Von Recklinghausen's Disease

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Abstract

Neurofibromatosis type-1 (NF) is an common autosomal dominant disorder that can involve tissues of neuroectodermal and mesenchymal origin. Specifically, type 1 neurofibromatosis involves multiple systems of the body. We present a child affected by von Recklinghausen's disease with skin, intracranial, thoracic, abdominal and skeletal tumors.

Presentation of Case: A 11 year old male child was admitted to our Emergency Department with right lateral thoracic injury. His physical examination revealed multiple neurofibromas all over the skin, scoliosis and multiple cafe au lait spots. Abdominal ultrasound showed a tumor between liver right kidney and pancreas. Brain, abdominal and thoracic MRI showed a huge abdominal mass, minor thoracic lesions and small lesions in the basal ganglia.

Discussion: NF is a common genetic disorder with no treatment. Patients without symptoms must be followed and operated only in case of a growing lesion. In patients with NF1, any symptoms with other systems should be managed carefully for underlying malignity

Conclusion: We would like to underline the need of close observation of children with von Recklinghausen disease with regard to manage multi system complications.

Keywords: *Neurofibromatosis Type-1 (NF); Von Recklinghausen's Disease*

Introduction

Neurofibromatosis (NF) is a multifocal neurocutaneous disease. It is most commonly prevalent autosomal dominant disease. The disorder affects all neural crest cells (Schwann cells, melanocytes and endoneurial fibroblasts). Cellular elements from these cell types proliferate excessively throughout the body forming tumors.

Case Report

An eleven year old male patient was admitted to our Emergency Department after right lateral thoracic and abdominal injury. His physical examination revealed that, arterial blood pressure was 100/65 mmHg, heart rate was 102 1/min, multiple neurofibromas all over the skin, scoliosis, and multiple cafe au lait spots. His neurological examination was normal. His hemoglobin level was 13.6 mg/dl and hematocrit level was 36%.Biochemical analysis results were in normal range. He was hemodynamically stable on follow up period and no blood level decrease detected. Abdominal ultrasound showed a tumor between liver right kidney and pancreas.

Contrast-enhanced CT and MRI examination was performed. The examination revealed a large, well-defined, polypoidal soft tissue thoracic and abdominal masses with mild to moderate post-contrast enhancement. The masses were located on paravertebral regions,

along esophagus, around the aorta and between liver, porta hepatis, peripancreatic region and retroperitoneum. In addition, a similar lesion was noted in the right lateral and posterior thoracic wall. The lesions were hypodense on non-contrast images with minimal to mild heterogeneous enhancement on post-contrast images. Also, was noted scoliosis. Radiological probability of neurofibromas was suggested. After all these, we performed brain MRI scan that showed small lesions in the basal ganglia.

Discussion

NF1 is a multi-systemic genetic disorder that may affect any organ in the body. NF-1 is a tumor disorder that is caused by the mutation of a gene on chromosome 17 that is responsible for control of cell division. NF-1 is inherited in an autosomal dominant fashion, although it can also arise due to spontaneous mutation. Prenatal testing may be used to identify the existence of NF-1 in the fetus.

Clinical manifestations are variable and include cutaneous (multiple café au lait spots, axillary and inguinal freckling, neurofibromas, neurologic (neuropathies secondary to neurofibromas), orthopedic (skeletal dysplasia, scoliosis, osteoporosis), and ophthalmologic (Lisch nodules on the iris, optic gliomas). Malignant tumors of neuroectodermal origin as well as pheochromocytoma are possible.

In NF1, gastrointestinal tumors occur most frequently followed by thoracic tumors, arterial involvement and endocrine tumors in the decreasing order of their frequency. Intra-thoracic tumor association with other tumors like pheochromocytoma, gliomas of the brain stem and of the optical nerve, is also reported.

Clinical presentation depends on the organ system involved. Diagnosis is usually clinically based using National Institute of Health (NIH) criteria. Genetic testing is available but has limitations.

Two of these seven NIH criteria are required for positive diagnosis.

- Six or more café-au-lait spots over 5 mm in greatest diameter in pre-pubertal individuals and over 15 mm in greatest diameter in post-pubertal individuals.
- Two or more neurofibromas of any type or 1 plexiform neurofibroma
- Freckling in the axillary or inguinal regions
- Optic glioma
- Two or more Lisch nodules (pigmented iris hamartomas)
- A distinctive osseous lesion such as kyphoscoliosis, sphenoid dysplasia, or thinning of the long bone cortex with or without pseudarthrosis.
- A first degree relative (parent, sibling, or offspring) with NF-1 by the above criteria.
- Discovered mutations of the NF1 gene, which is located at chromosome 17q11.2

NF is a common genetic disorder with no treatment.

Patients without symptoms must be followed and operated only in case of a growing. In patients with NF1, any symptoms with other systems should be managed carefully for underlying malignity

Manifestations of the disorder occur over time and require referral to multiple specialists: ophthalmology, neurology, orthopedics, cardiology, and surgery. Learning disabilities range from mild to severe in > 50% of those affected.

Children with NF require a multidisciplinary team approach with regular developmental assessment and monitoring of manifestations. Regular BP monitoring is necessary because of increased prevalence of essential hypertension.

Café au lait patches and skin-fold freckling do not usually cause complications. There is no evidence to support the routine use of laser treatment for cafe´ au lait patches. Subcutaneous neurofibromas are evident on palpation of the skin, may be tender to touch and cause tingling in the distribution of the affected nerve. Malignant change rarely occurs and if removal is contemplated, expert advice should be sought from NF1 specialists or soft tissue tumour/peripheral nerve surgeons as removal occasionally results in neurological deficit.

NF1 children need yearly assessment of the spine, and individuals with clinical evidence of scoliosis should be referred for expert orthopaedic advice and imaging.

Neurological examination should be undertaken during annual assessment. Any unexplained neurological signs and symptoms merit referral to a neurologist.

Abdominal bloating, pain, dyspepsia, haemorrhage and constipation may denote a gastrointestinal neurofibroma. Carcinoid tumours have a predilection for the duodenum where they give rise to facial flushing, diarrhoea, right sided cardiac lesions, facial telangiectasia and bronchoconstriction. Increased urinary levels of the serotonin metabolite 5-hydroxyindoleaceticacid confirm the diagnosis. Gastrointestinal stromal tumours, the commonest mesenchymal tumours of the gastrointestinal tract, have been observed recently in association with NF1. Patients present with anaemia and gastrointestinal bleeding but the majority of NF1 related tumours have a good prognosis.

Close collaboration between NF1 clinicians will facilitate a uniform approach to the diagnosis and management of NF1and its complications. Reliable clinical and radiological assessment will be helpful in determining the value of potential therapeutic agents [1-4].

Conclusion

We would like to underline the need of close observation of children with von Recklinghausen disease with regard to manage multi system complications.

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