Arachnoid cyst in a case of Neurofibromatosis type 1

Ashish Varma*, Abhilasha Singh, Revat Meshram, Rupali Salve, Anjali Kher, Jayant Vagha

Department of Pediatrics, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences (Deemed to Be University), Sawangi (M), Wardha. 442001, India.

*Corresponding author
Associate Professor, Department of Pediatrics, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences (Deemed to Be University), Sawangi (M), Wardha. 442001, India
Email: avarma2055@gmail.com

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ABSTRACT

Neurofibromatosis (NF) is categorized into two major subtypes: neurofibromatosis 1 (NF-1) and neurofibromatosis 2 (NF-2). The commonest subtype is NF-1. NF-1 is also called peripheral neurofibromatosis or Von Recklinghausen’s disease and NF-2 is also called central neurofibromatosis. NF 1 is a rare form of the genetic disorder, in which multiple noncancerous (benign) tumours develop in the nerve and skin (neurofibromas). There also occurs the development of areas of abnormally decreased or increased colouration of the skin. NF1 is a multisystem disorder, which requires management by multiple disciplines. In this report, we present
a case of a 7-years-old male child diagnosed with neurofibromatosis type 1. The symptoms of NF1 began to appear in childhood in the form of multiple hyperpigmented skin macules. He developed lower back pain from the last two months. The diagnosis NF-1 for the present case was made according to the presence of at least two among the seven clinical features.

Keywords: Nevus, Neurofibroma, Hyperpigmented, Recklinghausen’s disease, cafe-au-lait

1. INTRODUCTION

Although the first historical evidence of neurofibromatosis (NF) emerged in the 13th Century, it was identified as a distinct disorder after Friedrich Daniel von Recklinghausen, in 1882, wrote about NF in his publication (Boyd et al., 2009; Crowe et al., n.d.). Neurofibromatosis type 1 (NF-1) or Von Recklinghausen’s disease is a rare form of autosomal dominant neurogenetic disorder with variable expression and total penetrance, although it is sporadic in 50% of cases. It tends to mutate and evolve spontaneously with time (Sforza et al., 2016). It evolves in bursts after birth, particularly during growth, puberty, and pregnancy. Among all genetic diseases in humans, it has the maximal rate of spontaneous mutation (Ghalayani et al., 2012). It has an incidence of 1 to 2 in 2000 to 3000 of the total population (Ghalayani et al., 2012). The NF-1 tumour suppressor gene present on chromosome 17 (17q11.2) undergoes mutations giving rise to NF-1. This gene encodes neurofibromin, which is a protein responsible for inducing cell proliferation by downregulating the Ras-Raf/MAPK signalling pathway (Harrisingh and Lloyd, 2004). The mutations of this gene result in unrestrained cell proliferation and greater risk of developing cancer as a consequence of the impairment or loss of function of the negative regulator of cellular differentiation and growth of neurofibromin (Ferner et al., 2006; Huson, 2008).

In accordance with the National Institute of Health Consensus Development Conference, of the following, a minimum two criteria must be present for diagnosing NF-1 ("National Institutes of Health Consensus Development Conference," 1981):

- In the prepubertal patients, the occurrence of 5 or more cafe-au-lait spots having a diameter more than 5 mm, and in the postpubertal patients, the occurrence of 6 or more cafe-au-lait spots having a diameter more than 15 mm; The occurrence of any kind of neurofibromas 2 or more in numbers, or presence of at least one plexiform neurofibroma; The occurrence of fleckling in inguinal or axillary regions, The occurrence of optic glioma, The occurrence of Lisch’s nodules (iris hamartomas) 2 or more in numbers, The occurrence of a distinctive osseous lesion such as pseudoarthrosis of the tibia or sphenoid wing dysplasia, History of a first-degree relative, who has been diagnosed with neurofibromatosis type 1 ("National Institutes of Health Consensus Development Conference," 1981).

Clinically, the patients can be categorized into four classes, as under:
- Patients presenting with only pigmentary changes,
- Patients presenting with only neurofibromas,
- Patients presenting with both pigmentary changes and neurofibromas,
- Patients presenting with isolated plexiform neurofibromas.

2. CASE REPORT

A 7-year-old male child presented with the chief complaint of lower back pain for the past 2 months. He experienced constant excruciating pain and hyperesthesia over the lower back. The pain was not associated with any movement restrictions. He presented with a diffused nodular swelling over his lower back, which advanced gradually in the past three years. The patient also complained of a headache for the past 1 year, with the frequency of 2-3 times per month. As per the patient, it usually involved the frontal region and due to its high severity, he had vomiting.

The child was born full-term without complications by normal vaginal delivery, second in order of nonconsanguineous marriage, weighing 2.8 kg during birth. No significant history was present during birth. Immunization of the child was done appropriately for age. The patient’s developmental milestones were normal. On General examination, he was vitally stable. His body was demarcated by generalized hyperpigmentation from below the chest to the mid-thigh since birth. Coffee-coloured hyperpigmented macules were present on the face, neck, shoulders, arm, legs and abdomen. In the lumbar region, the skin was hyperpigmented with an increased amount of hair and appeared like a Nevus. In his left upper thigh near the ASIS, he had a black hairy nevus. Greenish patches were also present in the lumbar region. On Central Nervous System (CNS) examination, the tone, reflexes and power were normal. There were no focal neurologic deficits. Hyperesthesia of the skin over the lumbar region was present. Rests of the systems were normal.
Routine blood investigation was performed. Complete Blood count (CBC), Urine examination Kidney Function Test (KFT), and Liver Function Test (LFT) was normal. Ultrasonography (USG) of the lumbar region was suggestive of multiple subcutaneous neurofibromas. USG abdomen and USG pelvis were not significant. Magnetic resonance imaging (MRI) spine revealed well-defined dumbbell-shaped extra-axial lesion, which was homogenous in enhancement, at the level of the body of the L1 vertebra. It was suggestive of either spinal neurofibroma. MRI brain revealed a lesion of cerebrospinal fluid (CSF) signal-intensity in the suprasellar cistern and medial left temporal lobe. The probable diagnosis was left arachnoid cyst. The ophthalmic examination confirmed Lisch nodules in both the iris. The pupils of the patient were normal-3mm bilaterally reacting to light. After performing the refraction testing, he was advised to use spectacles. The patient did not have complaints of hearing difficulty, pain anywhere else in the body, or pins and needles sensation. From the examination or patient’s findings, the diagnosis was neurofibromatosis type 1 (figure 1 – 7).

The intervention included Carbamazepine for lower back pain. Soon after starting Carbamazepine, there was a reduction in pain. Counselling of his parents was done regarding neurofibromatosis. No surgical interventions were advised by the neurosurgeon for the neurofibromas. The patient was recommended six months follow up for the assessment for ophthalmologic examination, neurologic assessment, monitoring of the blood pressure and scoliosis evaluation.
3. DISCUSSION

Patients with neurofibromatosis type 1 start developing in early childhood, multiple café-au-lait spots that are flat dark patches on the skin (Crowe et al., n.d.). As the age advances, these dark spots enlarge in size and increase in number, as is present in our case. Usually, the freckles in the groin and underarms (Crowe’s sign) develop later in childhood (Crowe et al., n.d.). They are also present diffusely over the base of the neck, trunk, extremities and upper eyelids. The pigmenitary changes, in the majority of cases, follow the lines of Blaschko. One of the most frequent findings is optic pathway tumours (OPT). Optic tract lesions are typically localized to prechiasmal, chiasmal, and postchiasmal regions. Skeletal involvement in NF-1 is present in 40% of the cases, scoliosis being the prevailing deformity. A study conducted by Shapiro et al. revealed that 72% of the patients with NF-1 presented with oral manifestations (Shapiro et al., 1984).
Histologically, neurofibromas include a combination of perineurial cells, Schwann cells and endoneurial fibroblasts, which are non-capsulated. In the oral cavity, they can develop in both soft tissue and hard tissue. The tongue is the most frequently involved site in the oral cavity. The neurofibromas are typically asymptomatic and vary in size from 0.1 cm to various centimetres in diameter. They tend to emerge in a dermatomal distribution, most frequently the cervical region, followed by thoracic, lumbar, and sacral regions. They mostly develop on the skin (Gabhane et al., 2010). The plexiform neurofibroma expands along the peripheral nerve and it can involve certain nervous rami. Commonly, there is the involvement of 5th, 9th, and 10th cranial nerves. Neurological pathologies like Iris hamartomas, tumours of the central nervous system (such as gliomas and glioblastomas), acoustic nerve neurinoma, macrocephalies, and mental retardation (in 40% of cases) may also be present. There is rarely an occurrence of bilateral temporal arachnoid cysts. In a few instances, neurofibromatosis has been reported to involve unilateral arachnoid pouches (Martínez-Lage et al., 1993).

4. CONCLUSION
Neurofibromatosis is a progressive disorder. There is no specific treatment for NF. Therapy is designed to prevent or manage complications. Although there is a lack of available medical interventions, the ongoing trials are likely to be effective in treating both the cutaneous and noncutaneous manifestations of NF1. It is necessary to put these patients on the long-term follow-up to detect any possible signs of malignant transformation at the earliest.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal.

Conflict of Interest
There is no conflict of interest among the authors.

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Ethical Approval
Necessary approval was taken from the Institution and the patients for carrying out this work.

REFERENCE