Establishing the diagnosis neurofibromatosis type 1: A rare case

Dyatiara Devy, Damayanti
Department of Dermatology and Venereology, Faculty of Medicine, Universitas Airlangga / Dr. Soetomo General Hospital, Surabaya, Indonesia

Abstract

Neurofibromatosis type 1 (NF1) or Von Recklinghausen’s disease is a dominantly inherited genetic, multisystem disorder. Individuals with neurofibromatosis type 1 are prone to develop benign and malignant tumors of the CNS and peripheral nervous system, in addition to malignant diseases affecting other parts of the body. About 50% of individuals with neurofibromatosis type 1 have no family history of the disease and the disease is due to de novo (spontaneous) mutations. Early diagnosis is challenging because of its extremely variable characteristics. Some individuals may be mildly affected showing minimal signs, whereas others are severely afflicted. Individuals with NF-1 are best cared for within a multidisciplinary clinic, which has access to a wide range of subspecialists. The dermatologist has a role not only in the diagnosis of NF1 and differentiating it from other similar disorders, but also in the recognition of rare associated skin manifestations.

Introduction

Neurofibromatosis type 1 (NF1) or Von Recklinghausen’s disease is a rare genetic disorder that affects about one in 2500 to one in 3000 people world wide. Neurofibromatosis type 1 is a dominantly inherited genetic disorder that results from a germline mutation in the NF1 tumour-suppressor gene. NF1 is located on chromosome 17q11.2 and encodes a 220 kDa cytoplasmic protein called neurofibromin. This protein functions, in part, as a negative regulator of the Ras pathway. Neurofibromatosis type 1, typically manifest at birth but can continue to grow during adolescence and early adulthood, plexiform neurofibromas may manifest as diffuse appearance and/or a tendency to expand along large segments of affected nerves, causing disfigurement and nerve dysfunction. In this case there were plexiform neurofibromas with no disfigurement of the other structure. Yearly eye examinations by an ophthalmologist are essential in children under the age of 10 and may be necessary in older children. It is important for the health care provider to detect any visual changes. The common finding of ophthalmic abnormalities is lisch nodules, which are small, domeshaped hyperpigmented macules of the iris that cause no impairment of vision. They are a

Case Report

A 5-year-old girl presented with multiple hyperpigmented macule since birth. The macule became wider and multiplied in growth. No history with the same disease in her family (Figure 1). From physical examination, there were multiple café-au-lait spots with diameter more than 0.5 cm all over the body, plexiform neurofibroma, and axillary freckling (Figure 2). At this moment, this patient was diagnosed with neurofibromatosis type 1. We consult this patient to ophthalmologist, pediatricians and do the routine blood examination. The standard laboratory tests values were in the normal range. Lisch’s nodules on the iris of both eyes were found without clinical visual involvement, and from the CT Scan result showed no tumor in optic pathway. The pediatricians did not detect alterations in the central and peripheral nervous system.

Discussion

In childhood or early adolescence, the characteristic clinical features neurofibromatosis type 1 would be apparent. The diagnosis NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference (Table 1). In this case, we found 4 point indicates as neurofibromatosis. The café-au-lait macule with more than 0.5 cm in diameter is one of the seven cardinal diagnostic criteria of NF1 was found in this case more than six.

Café-au-lait spots, which are flat, pigmented macules, are often the first manifestation of NF-1 to appear. Frequently present at birth, they become more numerous as the infant grows. About 95-99% individu with neurofibromatosis shows café-au-lait spots. Axillary and inguinal freckling is another common clinical feature of neurofibromatosis type 1 and is usually detected in affected individuals by age 5–8 years. These pigmented abnormalities are typically the second diagnostic characteristic seen in children with the disorder, generally arising after development of cafe au-lait macules.

Correspondence: Dyatiara Devy, Department of Dermatology and Venerology, Faculty of Medicine, Universitas Airlangga/Dr. Soetomo General Hospital, Jl. Prof. Dr. Moestopo no. 47, Surabaya, Indonesia. Telephone/fax: 081216390735 E-mail: devy_doctor@yahoo.com

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conclusion by more than 3 years of age of children and are included as one of the cardinal NIH diagnostic criteria.2,6

Conclusions
Neurofibromatosis type 1 is a multisystem disorder requiring management by multiple disciplines. However, the diagnosis is missed on many individuals. The dermatologist has a role to play in the diagnosis of NF1, differentiating it from other similar disorders, and also recognition of rare but associated skin manifestations.

References