

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 57504

**Manuscript Type:** CASE REPORT

**Gene diagnosis of infantile neurofibromatosis type I: A case report**

Li MZ *et al.* Diagnosis of infantile neurofibromatosis type I

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## Abstract

### BACKGROUND

Neurofibromatosis is an autosomal dominant genetic disorder with various manifestations. Systemic multiple neurofibromatosis is rare in infancy. The disease is difficult to identify in the early stage, and it is prone to misdiagnosis and missed diagnosis. In the presence of lower limb swelling with subcutaneous nodules of unknown cause, café-au-lait spots, and axillary freckles, this disease must be

## Match Overview

1	Crossref 51 words Zhen Zhang, Xin Chen, Rui Zhou, Huaixiang Yin, Jiali Xu. "Chinese patient with neurofibromatosis-Noonan syndrome c...	3%
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## Neurofibromatosis type 1 | Genetic and Rare Diseases ...

<https://rarediseases.info.nih.gov/diseases/7866/neurofibromatosis-type-1> ▼

May 06, 2020 · Neurofibromatosis type 1 (NF1) is a genetic condition that affects the skin, the skeleton and the part of the nervous system outside the brain and spinal cord peripheral nervous system). The main signs and symptoms of NF1 include dark colored spots on the skin (café-au-lait spots), benign growths along the nerves (neurofibromas), and freckles in the underarm and groin.

## Bilateral Breast Cancer with Neurofibromatosis Type 1 ...

<https://pubmed.ncbi.nlm.nih.gov/29082380>

Neurofibromatosis type 1 (NF1) is autosomal dominant and it is the most common hereditary disease. This case report is about a woman and her daughter. Both of them are NF1 and mother also has metachronous bilateral breast carcinoma. We analyzed expressions of 84 genes related with DNA Repair by ...

Author: Duygu Dursun, Safiye Aktaş, Zekiye ...

Publish Year: 2017

## Coexistence of Neurofibromatosis Type-1 and MTHFR C677T ...

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3600223>

In neurofibromatosis type-1 (NF1), cerebrovascular disorders are rarely encountered although vasculopathy is a well-known complication. Several mutations seen in methylenetetrahydrofolate reductase (MTHFR) give rise to the formation of hyperhomocysteinemia and homocystinuria, a considerable risk factor for cardiovascular and cerebrovascular disorders, by leading to enzymatic ...

Cited by: 1

Author: Halim Yilmaz, Gulten Erkin, Haluk Gumu...

Publish Year: 2013

### PEOPLE ALSO ASK

Is neurofibromatosis dominant or recessive? ▼

How are neurofibromatosis type 1 diagnosed? ▼

What is the prognosis for neurofibromatosis? ▼

What is inheritance of neurofibromatosis? ▼



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Neurofibromatosis type I: a case report and review of the ...

<https://www.thefreelibrary.com/Neurofibromatosis...>

May 01, 1992 · **Neurofibromatosis type 1** affects 1 in 2500 to 4000 individuals and is related to an abnormality of chromosome 17.[7,8] It is further characterized by systemic, neurologic, cosmetic, and orthopedic manifestations.[4,6,9,10] **Type 2** affects 1 in 50,000 individuals and results from a chromosome 22 abnormality.[11,12] The most prominent **feature** of ...

(PDF) Neurofibromatosis type 1 and infantile spasms

<https://www.researchgate.net/publication/227325466...>

**Gene**-based analysis identifies exome-wide significant ( $P = 2.04 \times 10^{-6}$ ) enrichment of damaging de novo mutations in NF1, a **gene** primarily linked to **neurofibromatosis**, in **infantile** spasm.

PEOPLE ALSO ASK

How is neurofibromatosis type 1 inherited?

Is neurofibromatosis type 1 malignant?

Is neurofibromatosis autosomal dominant?

Feedback

Type I neurofibromatosis with spindle cell sarcoma: A case ...

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6795717>

Oct 06, 2019 · Neurofibromatosis type I (NF1), also known as von Recklinghausen disease, is an **autosomal dominant neurocutaneous disorder caused by a mutation in the NF1 gene**, and it usually presents during the first decade of life.

**Author:** Yu Zhang, Jiao-Jiao Chao, Xiu-Feng Liu,...      **Publish Year:** 2019

Jaffe-Campanacci syndrome or neurofibromatosis type 1: a ...

<https://ijponline.biomedcentral.com/articles/10.1186/s13052-020-0813-9>

May 11, 2020 · The clinical diagnosis of NF1 is based on the **presence of six or more CALMs > 5 mm** in prepubertal and > 15 mm in postpuberal individuals, two or more neurofibromas of any type or one plexiform neurofibroma, freckling in the axillary or inguinal regions, optic glioma, two or more Lisch nodules, distinctive osseous lesion (such as sphenoid dysplasia or tibial pseudarthrosis), a first-degree relative harboring **NF1 gene mutation** ...

**Cited by:** 1      **Author:** Silvia Vannelli, Raffaele Buganza, Federica ...  
**Publish Year:** 2020

Painful tumors in a patient with neurofibromatosis type 1 ...

<https://jmedicalcasereports.biomedcentral.com/...>

Oct 19, 2018 · Herein, we **report** an unusual **case** of multifocal glomus tumors in the same hand in a patient suffering from **neurofibromatosis type 1**. The patient was a 37-year-old Moroccan woman, suffering from **neurofibromatosis type 1**, with intense pain in the fingers, successfully treated with the excision of the tumors. Histology of the lesions confirmed the **diagnosis** of glomus tumor.

**Author:** Niema Aqil, Salim Gallouj, Kaoutar Mou...      **Publish Year:** 2018

Neurofibromatosis type 1 | Genetic and Rare Diseases ...

<https://rarediseases.info.nih.gov/diseases/7866/neurofibromatosis-type-1>

May 06, 2020 · **Neurofibromatosis type 1 (NF1) is inherited in an autosomal dominant pattern. All individuals inherit two copies of each gene. Autosomal means the gene is found on one of the numbered chromosomes found in both sexes. Dominant means that only one altered copy of a gene is necessary** to have the condition.

Lipofibromatosis-like neural tumor: Case report of a ...

<https://www.sciencedirect.com/science/article/pii/S2352512617302102>

Mar 01, 2018 · LPF-NT is rare, with 14 cases previously reported, and our patient is the first **report** of this **diagnosis** in infancy. This **case report** and literature review includes comparison of similar diagnoses including lipofibromatosis, low-grade malignant peripheral nerve sheath tumor, **infantile** fibrosarcoma, and dermatofibrosarcoma protuberans and serves ...

[PDF] Lipofibromatosis-like neural tumor: Case report of a ...

[https://www.jaadcasereports.org/article/S2352-5126\(17\)30210-2/pdf](https://www.jaadcasereports.org/article/S2352-5126(17)30210-2/pdf)

pleomorphism, or necrosis; **genetic** workup found LMNA-NTRK1 **gene** fusion, overall consistent with lipofibromatosis-like neural tumor (LPF-NT). LPF-NT is rare, with 14 cases previously reported, and our patient is the first **report** of this **diagnosis** in infancy. This **case report** and literature review includes

A novel mutation of the IL12RB1 gene in a child with ...

<https://www.ncbi.nlm.nih.gov/pubmed/19839503>

**A novel mutation of the IL12RB1 gene** in a child with nocardiosis, recurrent salmonellosis and neurofibromatosis type I: first case report from Thailand. Luangwedchakarn V(1), Jirapongsaranuruk O, NiemeLa JE, Thepthai C, Chokephaibulkit K, Sukpanichnant S, Pacharn P, Visitsunthorn N, Vichyanond P, Piboonpocanun S, Fleisher TA.

SEGMENTAL NEUROFIBROMATOSIS: A REPORT OF 3 CASES

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2856359>

**Neurofibromatosis type 1** is the most common of these disorders, affecting approximately 1 in 3500 individuals worldwide and with nearly 100% penetrance of the disease.[1,2] Segmental **neurofibromatosis** (SNF) was first described by Crowe et al. in 1956 and the authors termed it Sectorial **neurofibromatosis**.

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Neurofibronatosis type I

Complex Multi-System Human Disorder

Neurofibromatosis type I is a complex multi-system human disorder caused by the mutation of a gene on chromosome 17 that is responsible for production of a protein, called neurofibromin, which is needed for normal function in many human cell types. NF-1 causes tumors along the nervous system which can grow anywhere on the body. NF-1 is one of the most common genetic disorders and is not limited to any person's race or sex. NF-1 is an autosomal dominant disorder, which means that mutation or deletion of one copy of the NF-1 gene is sufficient for the development of NF-1, although presentation varies widely and is often different even between relatives affected by NF-1.

Wikipedia

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Neurofibromatosis

McCune–Albright syndrome

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