Yasunari nodules: specific marker of neurofibromatosis type 1

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Received: April 16st, 2023.
Aproved: May 2nd, 2023.

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Oftalmol Clin Exp (ISSNe 1851-2658)
2023; 16(2): e184-e187.

Funding
The authors did not receive support from any organization for the submitted work and they declare that this research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Abstract

Objective: Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder. Ocular involvement is a common manifestation, with the choroid being one of the structures most commonly affected. The diagnosis of choroidal involvement in NF1 is crucial for appropriate ophthalmological management of these patients.
Case report: A 34-year-old woman presented to our hospital center referred by the dermatology department. On examination, her best-corrected visual acuity was 20/20 and the slit lamp examination was normal in both eyes. Fundus examination revealed multiple nodules in the choroid, which were better visualized on infra-red imaging.
Conclusion: Yasunari nodules are considered a specific marker of NF1, thus their detection is relevant for the diagnosis of choroidal involvement crucial for appropriate management of these patients.
Key words: Yasunari nodules, neurofibromatosis type 1, near-infrared imaging, choroid diseases.

Nódulos de Yasunari: marcador específico de neurofibromatosis tipo 1

Resumen

Objetivo: La neurofibromatosis tipo 1 (NF1) es un trastorno genético autosómico dominante. Su compromiso ocular es una manifestación frecuente, siendo la coroides una de las estructuras más comúnmente afectada. El diagnóstico de la implicación coroidea en la NF1 es crucial para el manejo oftalmológico apropiado de estos pacientes.

Reporte de caso: Una mujer de 34 años se presentó en nuestro centro hospitalario derivada por el servicio de dermatología. Al examinarla, su mejor agudeza visual corregida fue de 20/20 y el examen con lámpara de hendidura fueron normales en ambos ojos. El examen de fondo de ojo reveló múltiples nódulos, que se visualizaron mejor en la imagen de infrarrojo.
Conclusión: Los nódulos de Yasunari se consideran un marcador específico de la NF1, por lo que su detección es relevante para el diagnóstico de la implicación coroidea y el apropiado abordaje en estos pacientes.
Palabras clave: nódulos de Yasunari, neurofibromatosis tipo 1, infrarrojo, enfermedades coroideas.

Nódulos de Yasunari: marcador específico de neurofibromatose tipo 1

Resumo

Objetivo: A neurofibromatose tipo 1 (NF1) é um transtorno genético autossômico dominante. O seu envolvimento ocular é uma manifestação frequente, sendo a coroide uma das estruturas mais comumente afetadas. O diagnóstico de envolvimento da coroide na NF1 é crucial para o manejo oftalmológico adequado desses pacientes.
Relato de caso: Paciente do sexo feminino, 34 anos, atendida em nosso hospital, encaminhada pelo serviço de dermatologia. Ao exame, sua melhor acuidade visual corrigida era 20/20 e o exame de lâmpada de fenda era normal em ambos os olhos. O exame de fundo de olho revelou múltiplos nódulos, que foram melhor visualizados na imagem infravermelha.
Conclusão: Os nódulos de Yasunari são considerados um marcador específico da NF1, pelo que a sua deteção é relevante para o diagnóstico do envolvimento da coroide e para a abordagem adequada nestes doentes.
Palavras-chave: Nódulos de Yasunari, neurofibromatose tipo 1, infravermelho, doenças da coroide.

Introduction

Neurofibromatosis type 1 (NF1), first described by von Recklinghausen, is an autosomal dominant disease caused by a defect in a single gene (NF1) on chromosome 17q11.2[1]. The incidence of NF1 is aroud1/3000 people in general population[2].

The identification of NF1 relies on clinical observations and there are specific diagnostic criteria that have been established. With the advent of novel methods for retinal imaging, there have been modifications to the previously established diagnostic criteria[3].

The high frequency of ocular affection suggests that the choroid is one of the structures most commonly affected by NF1. Choroidal neurofibromatosis was first characterized on histopathology by Wolter *et al*. in 1962 as ovoid bodies of proliferating neoplastic Schwann cells arranged in concentric rings around axons[4].

Choroidal nodules sometimes are inadequately perceptible during clinical examination and traditional color photography. Near-infrared reflectance (NIR) imaging is the optimal technique to visualize these nodules, which appear as numerous luminous patches on this modality of imaging[5].

Further research on this topic supported that NIR is a trustworthy and valid diagnostic imaging technique for detecting choroidal abnormalities in NF1 patients, with an extremely high level of agreement between observers[6].

The aim to present a clinical case that demonstrates the visualization of choroidal nodules using MC and NIR in a patient with NF1 and its implications for the diagnosis of ocular involvement.

Case report

A 34-year-old woman, who was diagnosed with NF1 during childhood, was referred by the dermatology department for an eye exam.

She had no visual symptoms, on examination her best-corrected visual acuity was 20/20 in the right eye and 20/20 in the left eye. Anterior segment examination was unremarkable in both eyes. Fundus examination of the both eyes showed multiple scattered pigmented lesions not defined at the posterior pole, which were better visualized on multicolor (MC) and NIR imaging (Fig. 1).

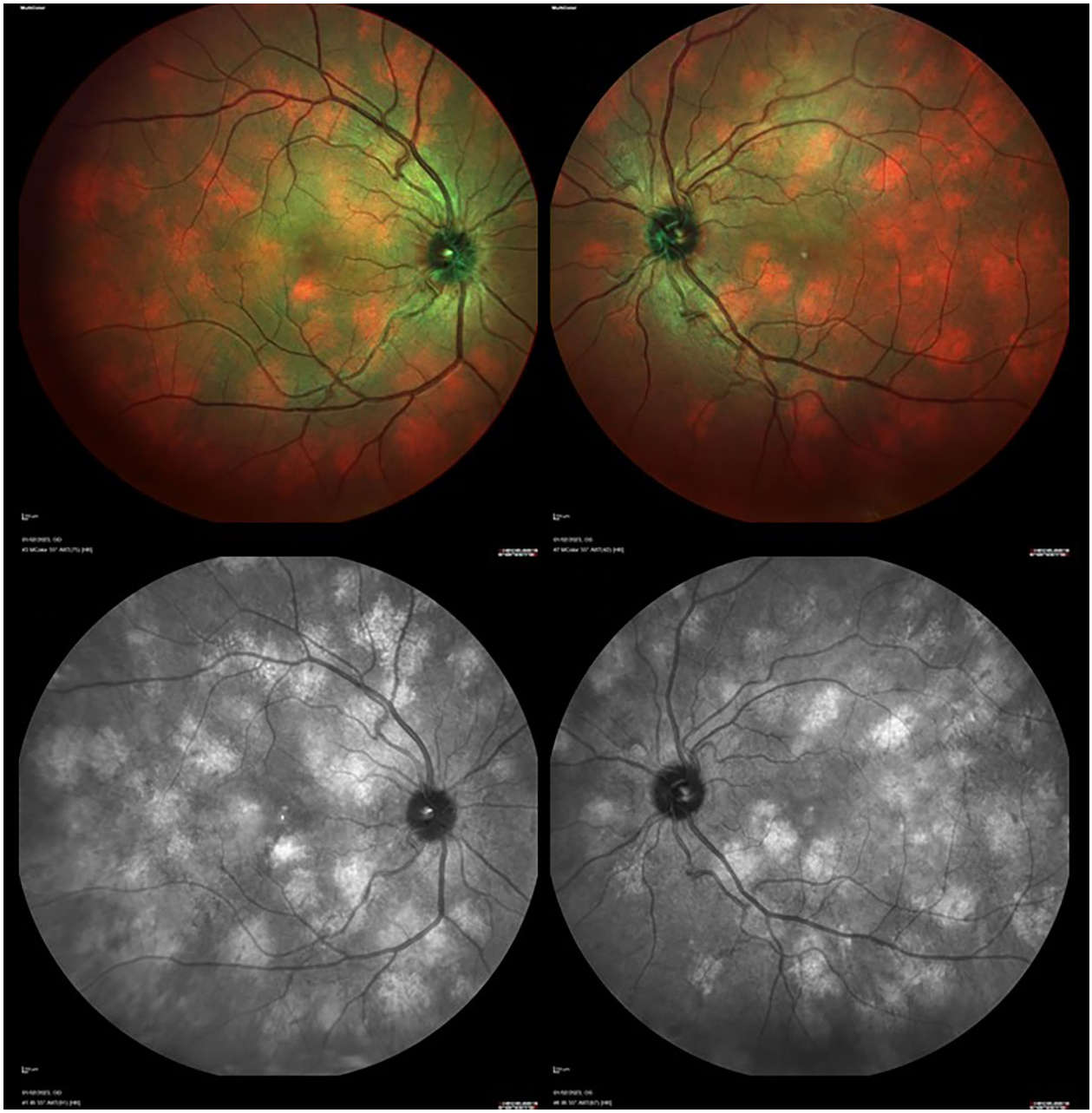

Figure 1. Multicolor scanning laser images of both eyes (A and B, respectively) exhibit choroidal nodules as red lesions, which are well-supported by the luminous patches seen on NIR imaging (C and D). Images were acquired using Spectralis (Spectralis™ HRA, Heidelberg Engineering Inc, Germany).

The MC images from right eye (Fig. 1a) and left eye (Fig. 1b) was performed by Spectralis, MultiColor module (Spectralis™ HRA, Heidelberg Engineering Inc, Germany) showed multiple scattered red lesions of varying size and number with blurred margins.

NIR imaging in right eyes (Fig. 1c) and left eyes (Fig. 1d) by Spectralis revealed the presence of multiple hyperreflective lesions.

The bright patches seen on NIR images correlated well with the areas of red pigmentation visible in the MC images. The patient was informed

of the situation and referred to the dermatology service notifying choroid involvement.

Discussion

Choroidal involvement is a common manifestation of NF1 with the reported prevalence ranging from in nearly 80% and can have significant implications for visual function[7-8].

These choroidal nodules visible in IR light was previously described in 2000 by Yasunari *et al.*[9]. The authors were the first to image choroidal neurofibromatosis in vivo, and they disclosed it as bright patchy regions on IR fundus examination and they called it as a Yasunari nodule. Yasunari nodules were described as benign, asymptomatic and tending to increase with age[10].

In 2012, Viola *et al.* postulated NIR examination to detect Yasunari nodules as a new diagnostic criterion for NF1 with choroidal involvement[8]. Further, in the 2021 recommendation, the presence of either two choroidal abnormalities or Lisch nodules was assigned diagnostic criteria point for NF1[11].

After the introduction of the mentioned criteria, the diagnostic scope of ocular findings in patients suspected of having NF1 was expanded[7].

In the actuality NIR imaging is being increasingly used to identify these choroidal abnormalities and should be performed, when possible, for the diagnosis of choroidal involvement in cases where the nodules are not visible on fundus examination[12].

Regular ophthalmic follow-up is recommended to monitor for any changes in the choroid that may require intervention.

In conclusion, the diagnosis of choroidal involvement in NF1 is crucial for appropriate management of these patients, in our case, NIR imaging was helpful in visualizing the choroidal nodules and confirming the diagnosis of NF1.

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