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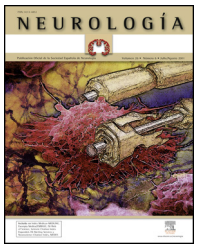

ORIGINAL ARTICLE

Prevalence of sleep disorders in patients with neurofibromatosis type 1☆

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KEYWORDS
Hyperhidrosis;
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Sleep;
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Abstract
Introduction: Neurofibromatosis type 1 (NF1) is frequently associated with neurological disorders unrelated to neurofibromas, including sleep disorders.
Objectives: This article reviews the prevalence of sleep disorders in patients with NF1, compares rates to data reported in the literature, and analyses the relationship between cognitive disorder and attention deficit hyperactivity disorder (ADHD) in these patients.
Material and methods: Comparative retrospective study reviewing data collected between January 2010 and January 2012 from patients diagnosed with NF1 in a tertiary hospital.
Results: We included 95 paediatric patients with NF1 who completed the Bruni Sleep Disturbance Scale in Children (SDSC). The overall prevalence of sleep disorders was 6.3%, which was lower than in the general paediatric population. Patients with NF1 and ADHD had a higher prevalence of sleep onset and maintenance disorders (18% vs 6.3%), sleep-wake transition disorders (12.5% vs 6.3%), and daytime sleepiness (12.5% vs 7.9%); differences were not statistically significant. A statistically significant difference was found in the subdomain of nocturnal hyperhidrosis (21.9% vs 6.3%, $P < .05$). Patients with NF1 and IQ < 85 showed higher prevalence rates of daytime sleepiness (20% vs 6.7%) and of sleep hyperhidrosis (11% vs 0%).
Conclusions: The prevalence of sleep disorders in our cohort of patients with NF1 was no higher than in the general paediatric population, although some of these disorders are more common in cases with cognitive disorders or ADHD.
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PALABRAS CLAVE
Hiperhidrosis;
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Somnolencia;
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Trastorno por déficit de atención e hiperactividad

Prevalencia de trastornos del sueño en pacientes con neurofibromatosis tipo 1

Resumen
Introducción: La neurofibromatosis tipo 1 (NF1) asocia frecuentemente alteraciones neurológicas no relacionadas con neurofibromas, entre las que se encuentran los trastornos del sueño.
Objetivos: Revisión de la prevalencia de trastornos de sueño en pacientes con NF1 y compararla con los datos descritos en la literatura, así como analizar la relación con el trastorno cognitivo y el trastorno por déficit de atención e hiperactividad (TDAH) en estos pacientes.
Material y métodos: Estudio comparativo, retrospectivo, mediante la revisión de los datos recogidos entre enero de 2010 y enero de 2012 de pacientes diagnosticados de NF1 en un hospital de tercer nivel.
Resultados: Se incluyeron 95 pacientes con NF1 pediátricos que respondieron correctamente a la Escala de alteraciones del sueño en la infancia de Bruni, encontrando una prevalencia de trastorno global del sueño del 6,3%, inferior al de la población pediátrica general. Aquellos pacientes con NF1 y TDAH presentaron mayor prevalencia de trastorno de inicio-mantenimiento del sueño (18 vs 6,3%), de transición sueño-vigilia (12,5 vs 6,3%) y somnolencia diurna (12,5 vs 7,9%) sin alcanzar significación estadística, sí encontrándose diferencia estadísticamente significativa en la subescala de hiperhidrosis nocturna (21,9 vs 6,3%; $p < 0,05$). Los pacientes con NF1 y cociente intelectual < 85 presentaron mayor prevalencia de somnolencia diurna (20 vs 6,7%) y mayor hiperhidrosis nocturna (11 vs 0%).
Conclusiones: En nuestra cohorte de pacientes con NF1 no hemos encontrado aumento de la prevalencia de trastornos de sueño con respecto a la población pediátrica general, aunque algunos de estos trastornos son más frecuentes en caso de alteraciones cognitivas o TDAH.
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Introduction

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is a progressive multisystem genetic disorder predominantly affecting the skin and the central nervous system. With an approximate incidence rate of 1 case per 2500 to 3500 live births,[1] NF1 is considered one of the most frequent autosomal dominant genetic conditions. While it shows complete penetrance, its clinical phenotype is variable and unpredictable.[1] Approximately 50% of patients with NF1 present mild forms of the disease, and about a third develop severe complications arising when plexiform neurofibromas affect organs; these neurofibromas present a high risk of malignant transformation.[2] NF1 is frequently associated with neurological and vascular changes which are unrelated to neurofibromas but whose aetiology remains unclear. Some of the neurological diseases described in patients with NF1 include learning disorders, sleep disorders, epilepsy, macrocephaly, and neuroimaging abnormalities.[2]

Sleep is very important as it occupies about one-third of a person's life, and while sleep disorders are frequent during childhood, they may go unnoticed.

In view of the low number of studies reporting an increased incidence of sleep disorders in patients with NF1, we decided to analyse the prevalence of sleep disorders in a cohort of patients with NF1 and compare our results to data provided in the literature. Our study also analyses co-presence of mental retardation and attention deficit hyperactivity disorder (ADHD) in these patients.

Materials and methods

We conducted a retrospective cross-sectional comparative study by reviewing the data gathered between January 2010 and January 2012 from patients diagnosed with NF1 in a paediatric neurology department at a tertiary hospital. Data were collected using a standardised clinical interview, a psychometric assessment, and sleep questionnaires that were completed by the patients' parents.

Sleep disorders were assessed with the Sleep Disturbance Scale for Children (SDSC),[3] which consists of 27 items scored using a Likert-type scale. The SDSC classifies sleep disorders in six categories: disorders of initiating and maintaining sleep, sleep breathing disorders, disorders of arousal/nightmares, sleep-wake transition disorders, disorders of excessive daytime somnolence, and sleep hyperhidrosis. The SDSC evaluates the preceding 6 months and has been validated in children aged 6 to 15 years. Subsequent studies also support use of this scale in preschool-aged children.[4] The cut-off point for global sleep disorder is 39, with a sensitivity of 89% and a specificity of 74%. Additionally, this scale establishes normal ranges for the subscales of specific sleep disorders.

The NF1 database lists a total of 215 patients, 102 of whom had been assessed with the SDSC. Seven patients were excluded from the study: 2 had reached the age of 18 and the remaining 5 had filled in the questionnaire incompletely or incorrectly. A total of 95 patients were included in the study. We analysed the following variables: age, sex, IQ scores, presence and subtype of ADHD, the presence of other

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