

Voice-related quality of life in adults with neurofibromatosis type 1

Marjan Cosyns¹, Geert Mortier^{2,3}, Sandra Janssens² and John Van Borsel¹

¹Department of Otorhinolaryngology & Logopaedic and Audiologic Sciences, Ghent University

²Center for Medical Genetics, Ghent University Hospital

³Center for Medical Genetics, Antwerp University Hospital

Introduction

NF1 is an autosomal dominant neurocutaneous disorder occurring in 1/3000 to 4000 individuals. Its hallmark features include café-au-lait spots, neurofibromas, skinfold freckling and Lisch nodules (Figure 1). NF1 is caused by mutations in the NF1 gene, which is located at the long arm of chromosome 17 (17q11.2).

Voice characteristics of NF1 patients have been documented using both subjective and objective evaluations.

- Subjective: The voice of NF1 patients has been described as breathy, weak, hoarse, harsh, creaky, monotone, tremulous, atypically loud and monoloud.
- Objective: Findings included a reduced VC, narrower frequency and intensity range, smaller standard deviation of F_0 and lower DSI.

As long as these voice characteristics pose no adversities in everyday life, they may not be considered 'voice problems' as such. Therefore, **the aim was to measure the psychosocial handicapping effects of the voice characteristics associated with NF1.**

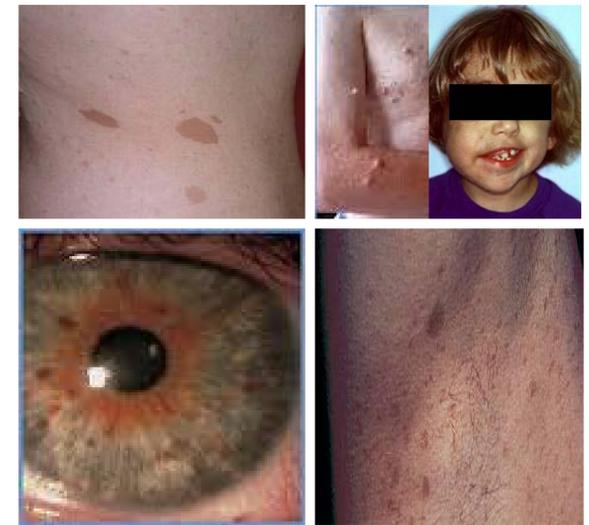


Figure 1. Hallmark features of NF1

Methods

The **patient group** consisted of 30 adults (15 males and 15 females) with NF1 ranging in age from 17 to 64 years old (mean = 35y 6m, SD = 10y 11m). They all fulfilled the diagnostic criteria for NF1 and none had been diagnosed with laryngeal or pharyngeal neurofibromas.

The **control group** was frequency matched for age, gender, smoking behavior and vocal usage. No one reported a history of voice problems.

Participants were asked to complete the Flemish version of the **Voice Handicap Index (VHI)**. Additionally, the level of severity of NF1 was rated and DSIs, calculated in a previous study, were collected.

Results

NF1 patients obtained significantly higher values than controls for total, functional and emotional VHI (sub)score ($p < 0.001$). For the physical subscale, a trend was observed ($p = 0.075$) (Figure 2).

In the patient group, VHI (sub)scores of males and females were compared for possible gender differences, and possible correlations between total VHI score and either age, severity of NF1 or DSI were explored. Only a statistically significant correlation between total VHI score and age could be demonstrated ($r = 0.402$, $r^2 = 0.16$, $p = 0.031$) (Figure 3-5).

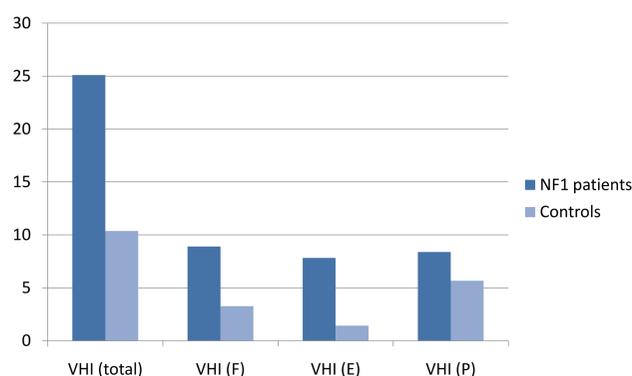


Figure 2. Mean VHI scores of NF1 patients compared with controls

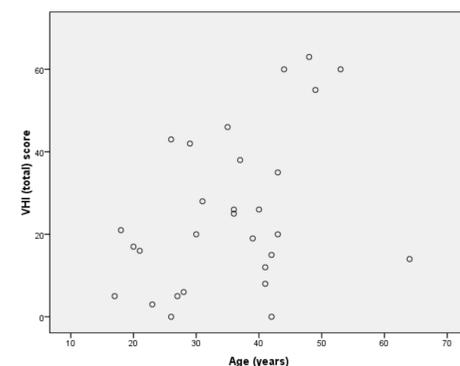


Figure 3. Total VHI score in function of age

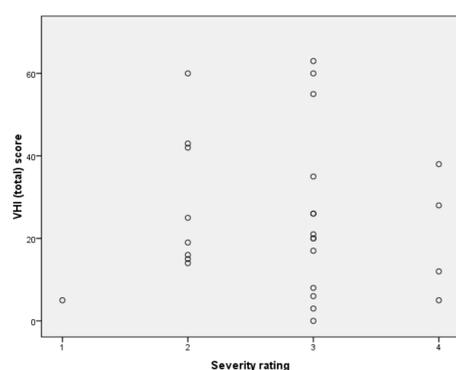


Figure 4. Total VHI score in function of severity of NF1

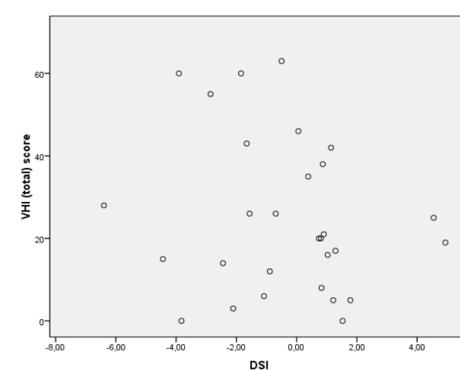


Figure 5. Total VHI score in function of DSI

Conclusion

NF1 patients scored significantly higher on the VHI compared with controls, suggesting that NF1 patients present with voice characteristics that are sufficiently different to experience psychosocial consequences. However, it is likely that the elevated VHI scores were not merely related to the voice itself. A flow-over from other psycho-physical issues inherent to NF1 might have played a role.

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