

# Case Report

## Usher's syndrome and Von Recklinghausen's neurofibromatosis association: Case Report

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### ABSTRACT

The probability that the same person suffers from Usher's syndrome and Von Recklinghausen's neurofibromatosis is exceptional. This observation was intended to describe the coexistence of these two diseases in a patient born to a consanguineous union. A 40-year-old from Mauritania consulted our department for bilateral blindness. Its review noted an absence of light perception, retinitis pigmentosa typical lesions, bilateral deafness without vestibular damage and neurofibromas covering the entire body surface. Rare genetic diseases can be the result of a mutation. However, inbreeding increases the risk of their occurrence. This practice should be abandoned.

**Keywords:** Consanguinity - Rare genetic diseases - Usher's syndrome-Neurofibromatosis - Association

### INTRODUCTION

Usher's syndrome (US) is defined by the association of congenital sensorineural hearing loss of varying severity, scalable or not and retinitis pigmentosa gradually blinding [1]. There are three clinical types according to the vestibular damage and degree of hearing loss. A gene D1S81 localized in the long arm of chromosome 1 controls type II we are interested in this article. In this form, unlike the other two, the acquisition of language is possible and pigmentary retinopathy appears later, it is progressive and becomes annoying at the age of 30 years [2, 3].

Neurofibromatosis is an autosomal dominant disease [4]. There are two types. Von Recklinghausen's Neurofibromatosis (VRN) described in this work is the type I. It represents about 90% of clinical forms and is due to an abnormality of chromosome 17. The Lisch's iris nodules, plexiform neuroma of the upper eyelid, and the optic glioma represent its most common ocular sign.

The coexistence of both diseases in the same person is exceptional; we found no cases in the literature. This observation has aimed to report an US-VRN combination in a 40-year-old man born of inbreeding, from Mauritania.

### PRESENTATION OF THE CASE

(Mr. M) , a 40 years old admitted for blindness that began at around 21 years old. Bilateral blindness, rapidly progressive, painless, without fever and without impairment of general condition. It was associated with deafness, handicapping quickly, forcing his entourage to speak "hard" for his understanding.

He was born in a family of four children from a consanguineous marriage. His paternal grandfather is suffering from Von Recklinghausen's neurofibromatosis, and her maternal grandmother is suffering from retinitis pigmentosa. One of the sisters is albino and another suffers from retinitis pigmentosa. Figure 1 shows the pedigree of Mr. M.

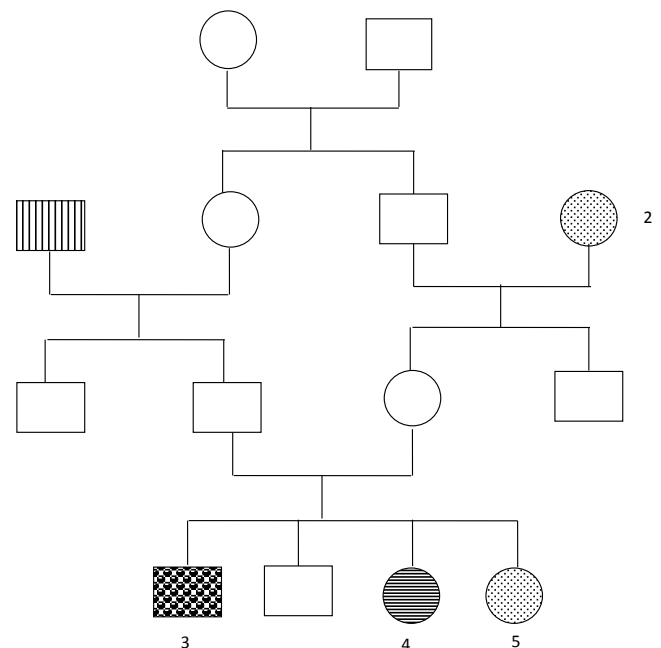


Figure 1 : Mr. M's family pedigree

- Subject 1 : Mr. M's paternal grandfather who suffers from neurofibromatosis
- Subject 2: Mr. M's maternal grandmother who suffers from retinitis pigmentosa
- Subject 3: Mr. M
- Subject 4: Mr. M's Sister who suffers albinism
- Subject 5: Mr. M's Sister who suffers from retinitis pigmentosa

The clinical examen of the patient noted on both sides: an absence of light perception, a retinitis pigmentosa, a medium sensorineural hearing loss (loss of 50 decibels) without vestibular dysfunction. Neurofibromas covered his entire body surface area (Figure 2).

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Figure 2

## DISCUSSION

Von GRAFFE [1] first described US in 1858. Usher [5] was the first to understand the hereditary nature of this disease association and described as a specific syndrome. It is at the origin of 3 to 6% of congenital deafness; and represents 50% of the deafblind population in the United States [3]. Its prevalence (P1) is 1 birth / 1000 [2]. Von Recklinghausen [6] was the first in 1882 to describe the neurofibromatosis that will bear his name. VRN described in this work is the type I. Prevalence (P2) is 1 birth / 4950 [4]. If we considered a simple mathematical model, \* independence in probability theorem ([www.medespace.fr/.../probabilité-conditionnelle-independance](http://www.medespace.fr/.../probabilité-conditionnelle-independance)), the probability (P) for a single individual suffering from these two diseases (US-VRN association) can be calculated by the following formula:  $P(P1 \times P2) = P1 \times P2 = 1 \text{ birth} / 4950000$ . This probability is extremely low. Inbreeding had probably played a role in this association [7]. In Britain, a third of children with rare recessive genetic

diseases result from a consanguineous marriage (<http://www.bivouac-id.com/mariages-consanguins-et-tares-genetiques/>). In Morocco, diseases related to inbreeding occupy an important place in the health system. The worst of these is Duchenne muscular dystrophy, children die around age 15 (<http://www.maghress.com/fr/leconomiste/34435>). Approximately 50% of marriages are consanguineous in Mauritania (<http://www.bivouac-id.com/mariages-consanguins-et-tares-genetiques/>). Some factors could explain inbreeding. The traditional family structure, geographic isolation, economic conditions, poor marry among themselves and the rich do the same, and the religious isolation. It would be interesting to determine the gene at the root of this association. Mr. M refused any blood examen because no treatment would be offered.

\* Theorem of independence in probability: considering two independent events A and B. If the probability of occurrence of event A is P (A), if the probability of occurrence of event B is P (B). The probability that both events A and B can appear simultaneously is  $P(A \cap B) = P(A) \times P(B)$ .

## CONCLUSION

Consanguineous Unions are still common in Mauritania. They are sometimes the source of rare genetic disease, often severe. Doctors, political and religious leaders should unite efforts to prohibit such marriages between members of the same family

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