Alezzandrini Syndrome – A forme fruste?

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Introduction

Alezzandrini syndrome is a rare group of ipsilateral pigmentary changes characterised by poliosis, facial vitiligo, unilateral degenerative retinitis, an atrophic iris, reduced visual acuity and occasionally deafness.(1, 2)

Our case demonstrates 2 of the four major features and a strong family history of each of these features. It is especially interesting as there may be a genetic basis for a rare condition previously thought to be idiopathic. Alternative diagnoses are also suggested in the discussion below.

Case History

A 38 year old gentleman presented to our dermatology clinic with new-onset peri-ocular vitiligo (Figure 1) noticed a few months after recovery from a kidney infection. It then transpired that from childhood he has had hearing loss that was worse on the left. His ophthalmologist also recently diagnosed him with bilateral cataracts also worse on the left that were likely to have been present since childhood. He is otherwise well and of normal stature with no history of associated autoimmune disorders such as hypo- or hyperthyroidism, diabetes mellitus, pernicious anaemia or gonadal failure.

In his family history, his sister has epilepsy and hearing loss and his brother possibly has alopecia areata. His mother also has hearing loss and degenerative retinitis. His maternal grandmother, her three sisters and brother all have hearing loss as well as two nephews. Our patient’s family tree is illustrated in Figure 2. This tree is only partially complete as he is unaware of the medical histories of his great-aunts’ children and his mother’s cousins.

Discussion

There are a number of dominantly inherited causes of deafness including Muckle-Wells Syndrome, an auto-inflammatory disorder (3). In these scenarios, auditory disturbances tend to occur either in isolation or with a different constellation of symptoms. Other syndromes that include deafness and degenerative retinitis. His maternal grandmother, her three sisters and brother all have hearing loss as well as two nephews. Our patient’s family tree is illustrated in Figure 2. This tree is only partially complete as he is unaware of the medical histories of his great-aunts’ children and his mother’s cousins.

Discussion Cont’d.

HLADRB1*0405 has been identified as a susceptibility allele for VKH syndrome (4) however, there is no mention of a clear pattern of inheritance. Alezzandrini syndrome is thought to be a mild variant of VKH syndrome without meningitis and severe uveitis.

In our literature review, less than 10 cases have been reported and none of them have had a clinical history suggestive of a Mendelian autosomal dominant inheritance pattern. Given the bilateral involvement with unilateral accentuation and clear inheritance pattern, we suggest that this case may represent a forme fruste of Alezzandrini syndrome and warrants further exploration.

References


*Clinical image used with consent for this poster.

Disclosures: None