Evidence of genetic heterogeneity in Alberta Hutterites with Usher syndrome type I.

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Abstract: To identify the genetic defect in a Hutterite population from northern Alberta with Usher syndrome type I.

Complete ophthalmic examinations were conducted on two boys and two girls from two related Hutterite families diagnosed with Usher syndrome type I. DNA from patients and their parents was first evaluated for a mutation in exon 10 of the protocadherin-related 15 (PCDH15) gene (c.1471delG), previously reported in southern Alberta Hutterite patients with Usher syndrome (USH1F). Single nucleotide polymorphic linkage analysis was then used to confirm another locus, and DNA was analyzed with the Usher Chip v4.0 platform.

Severe hearing impairment, unintelligible speech, and retinitis pigmentosa with varying degrees of visual acuity and visual field loss established a clinical diagnosis of Usher syndrome type I. The patients did not carry the exon 10 mutation in the PCDH15 gene; however, with microarray analysis, a previously reported mutation (c.52C>T; p.Q18X) in the myosin VIIA (MYO7A) gene was found in the homozygous state in the affected siblings.

The finding of a MYO7A mutation in two related Hutterite families from northern Alberta provides evidence of genetic heterogeneity in Hutterites affected by Usher syndrome type I.

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