High prevalence of m.1555A > G in patients with hearing loss in the Baikal Lake region of Russia as a result of founder effect

MT-RNR1

Article: High prevalence of m.1555A > G in patients with hearing loss in the Baikal Lake region of Russia as a result of founder effect
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Authors: Tuyara V. Borisova et al. **Date:** July 2024 **Journal:** Scientific Reports

Key Findings:

- **Prevalence of m.1555A > G:** Among 165 individuals studied, the m.1555A > G variant was present in 12.7% (21 patients), significantly higher than the global prevalence of around 1 in 500.
- **Ethnic disparities:** The variant was more prevalent in Buryat patients (20.2%) compared to Russian patients (1.3%).
- **Lineages:** Among the Buryat families, 92.9% belonged to a single maternal lineage cluster (A5b sub-haplogroup), indicating a founder effect.
- **Global prevalence:** The m.1555A > G variant shows notable variation worldwide, with the highest prevalence in Spain (36.3%), likely due to founder effects associated with the mitochondrial sub-haplogroup H3, followed by Buryatia (20.2%).
- **Aminoglycoside use:** Widespread aminoglycoside antibiotic use in some regions, such as Asia, may contribute to increased hearing loss in variant carriers.

Study Focus

This study investigates the prevalence of the MT-RNR1 m.1555A > G variant among patients with hearing loss in the Republic of Buryatia, located in the Baikal Lake region of Russia. The region presents a unique genetic landscape due to its population's history and demographics.

Background

The A > G change at position 1555 of the MT-RNR1 mitochondrial gene creates a new base pair in human 12S rRNA, making it look more like a bacterial structure that aminoglycoside antibiotics target, potentially leading to hearing loss.

Definitions

Mitochondrial Haplogroup:

A group of genetic variants traced through mitochondrial DNA (mtDNA) to a common ancestor, used to study maternal lineage and migration patterns.

Founder Effect (in mtDNA):

Occurs when a small group with limited mitochondrial genetic diversity establishes a population, leading to a higher frequency of certain mitochondrial traits or mutations in that group. This can result in the formation of a distinct mitochondrial haplogroup.

Conclusion

The study highlights the importance of including the m.1555A > G variant in genetic testing protocols for hearing loss, especially in regions with a known high prevalence. The findings underscore the impact of founder effects in genetic epidemiology, influencing the distribution and frequency of specific pathogenic variants.

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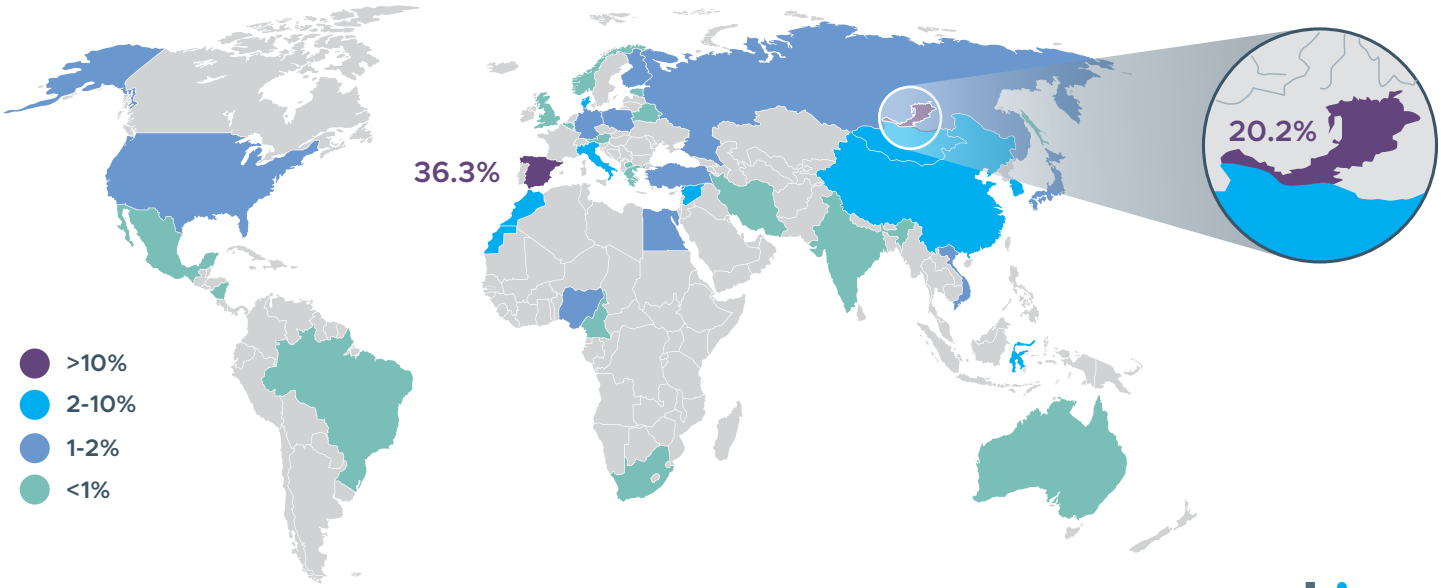
The worldwide prevalence of the **m.1555A > G variant** of the **MT-RNR1 gene among 47,328 patients** with hearing loss.



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