Family-based exome sequencing disclose associated genes in primary headaches

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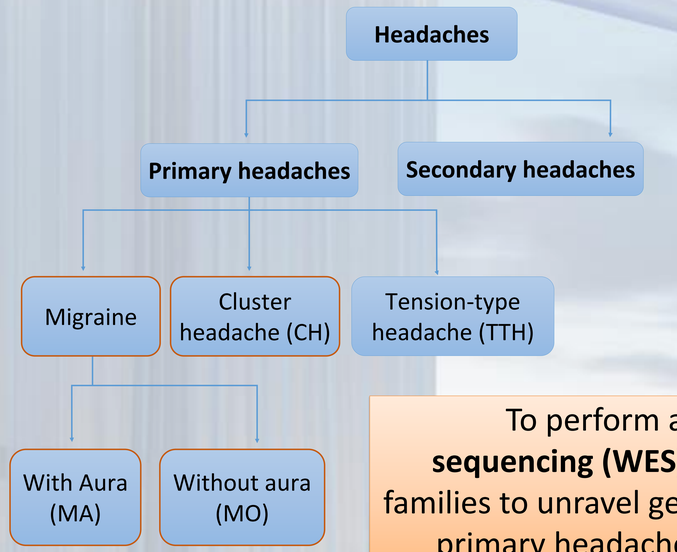
INTRODUCTION



AIM

To perform a **whole-exome sequencing (WES)** in Portuguese families to unravel genetic factors in primary headache susceptibility.

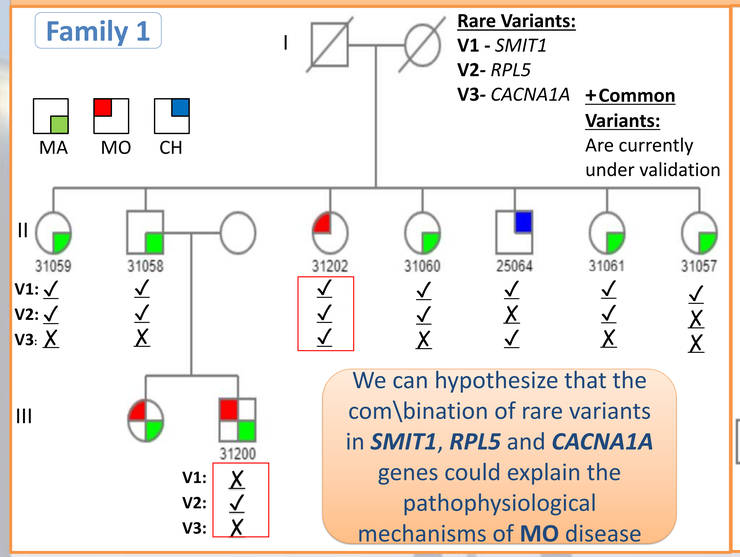
SUBJECTS/METHODS

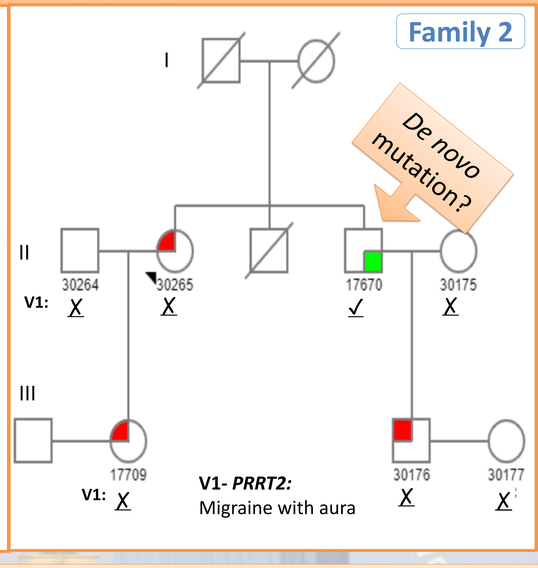
- DNA samples from 3 families (20 individuals) with migraine and CH (as well as controls).
- **WES (Novogene corporation)**
- ✓ Data analysis: VarAFT (Variant Annotation and Filtration Tool)

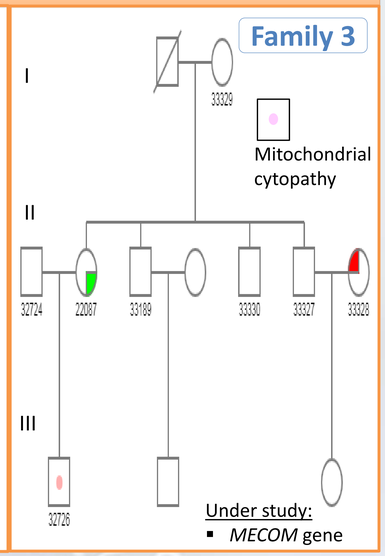
Criteria filters

Rare variants:	Common variants:
Exonic and splicing variants; Frequency – gnomAD ≤ 0,01; UMD Predictor	Exonic and splicing variants; UMD Predictor; HPO: Migraine

RESULTS







CONCLUSIONS

❖ For **Family 1, we have 2 hypothesis**.

1st hypothesis	2nd hypothesis
"Common disease-common variant" ?	"Common disease-rare variant" - *SMIT1* - *RPL5* - *CACNA1A*

The two types of variants may interplay in migraine susceptibility

❖ **Family 2:** A mutation in ***PRRT2*** gene was found associated with **MA** susceptibility. It was not possible to conclude if this is a *de novo* mutation or if it was inherited by any of the parents.

❖ **Family 3:** results need to be further deepened to draw some conclusions.

Acknowledgments

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