

# The Other Half of the Story: High Rates of Unique Parental Secondary Findings

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## BACKGROUND

- Secondary findings (SF) are an optional result type from exome and genome testing for patients and their families.
- The purpose of SF is to give individuals the choice to receive clinically actionable information unrelated to the primary testing indication, supporting proactive health management.
- The ACMG maintains a recommended SF gene list; version 3.3, published in 2025, includes 84 actionable genes (1).
- Individualized SF results play an important role in providing each family member with personalized, clinically relevant information, though availability of individual reports varies across laboratories.

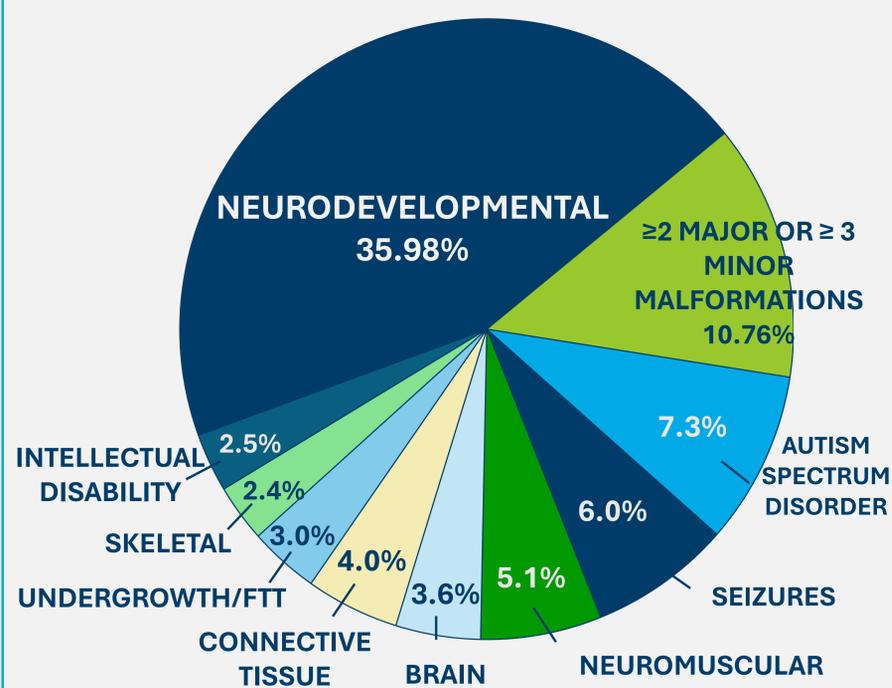
## METHODS

Retrospective review of probands and their parents who underwent exome testing at a single clinical laboratory between 2016–2024.

Assessed trends in ordering of secondary findings and the results of secondary findings in probands and their family members

## RESULTS

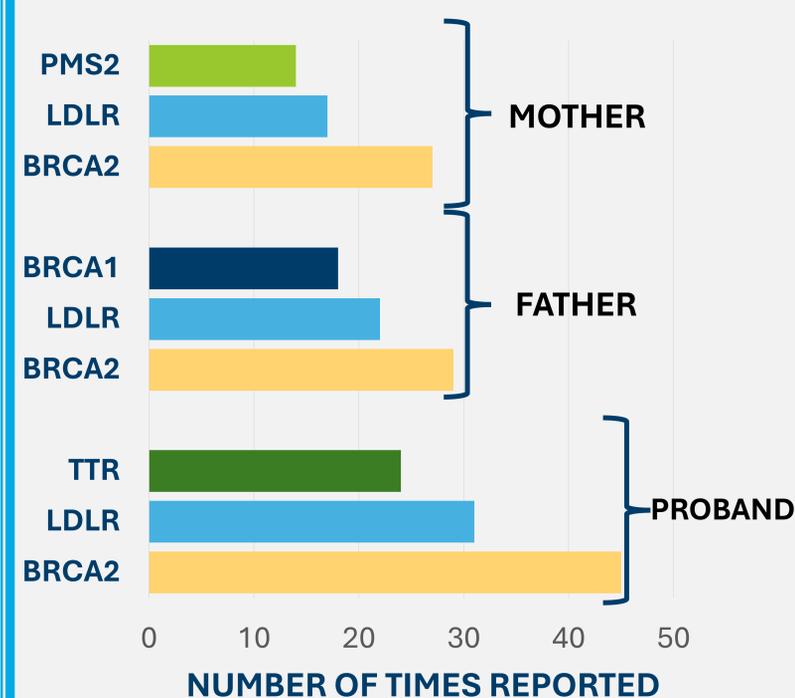
### TOP TEN INDICATIONS FOR EXOME TESTING FOR PROBANDS WHO OPTED-IN TO SECONDARY FINDINGS



### PROBAND SF OPT-IN RATE CORRELATES WITH AGE GROUP

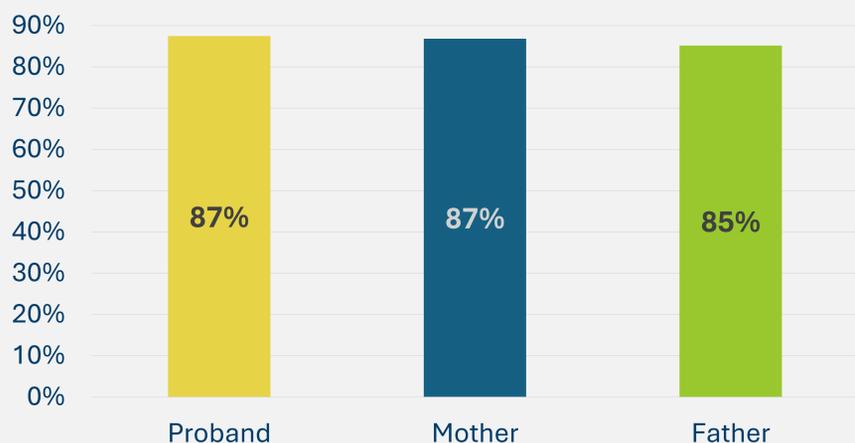


### BRCA2 AND LDLR ARE THE TWO MOST COMMONLY REPORTED SF GENES



### SF OPT-IN RATE FOR PROBANDS AND THEIR PARENTS

FOR PROBANDS AND THEIR PARENTS

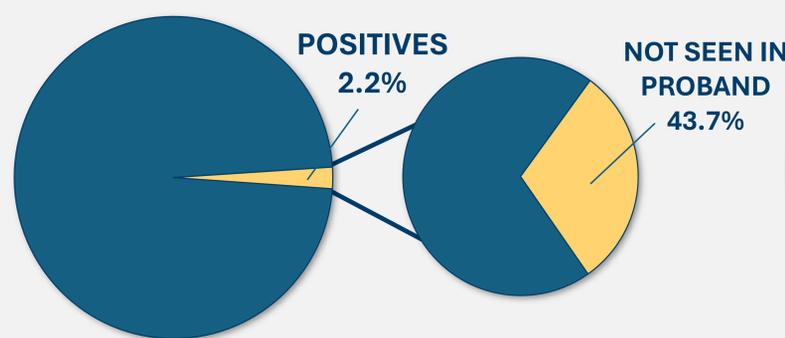


### NEARLY HALF OF REPORTED SF IN PARENTS WERE NOT PRESENT IN PROBAND

#### MOTHER OF PROBAND

Maternal Positive SF Rate

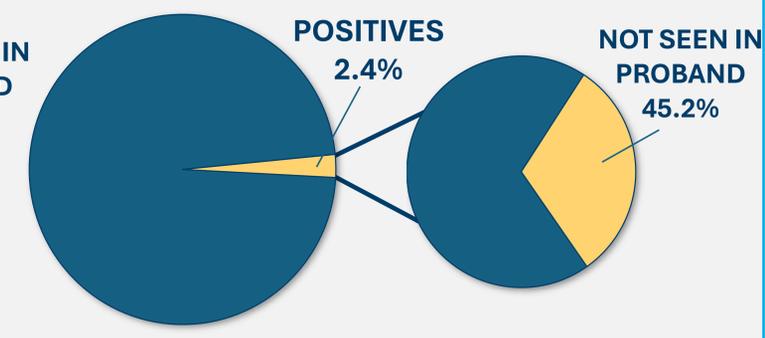
% of Positives Not Seen in Proband



#### FATHER OF PROBAND

Paternal Positive SF Rate

% of Positives Not Seen in Proband



## TAKE HOME POINTS

- Many parental secondary findings are not present in the proband, underscoring the importance of reporting results for each individual rather than relying solely on the proband's findings.
- Independent secondary finding tests and reports ensure parents do not miss actionable information and supports early detection and prevention.

## REFERENCES

(1) Miller, D. T., Lee, K., Abul-Husn, N. S., Amendola, L. M., Brothers, K., et al., & ACMG Secondary Findings Working Group. Electronic address: documents@acmg.net (2023). ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in medicine : official journal of the American College of Medical Genetics*, 25(8), 100866. <https://doi.org/10.1016/j.gim.2023.100866>