Nationwide real-world *ABCA4* cohort highlights the complex clinical and genetic landscape of **Stargardt** disease

Purpose

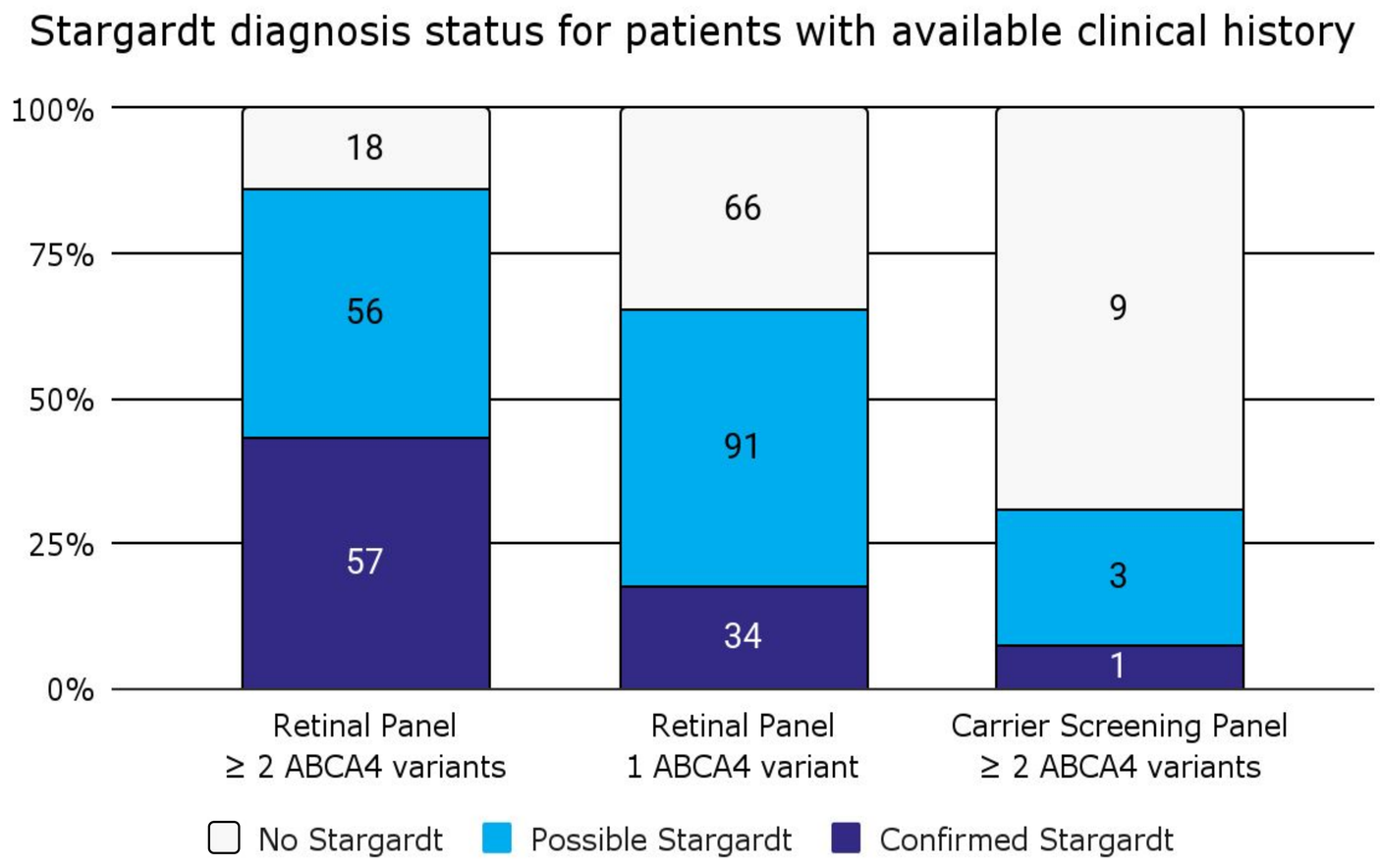
- Stargardt disease, the most common inherited macular dystrophy, is caused by biallelic *ABCA4* variants.
- Genetic testing is complicated by complex alleles, hypomorphic variants, and second alleles that can be difficult to detect in many affected individuals.
- To date, published *ABCA4* cohorts have been drawn solely from academic medical centers or clinical trials.
- We aimed to describe a real-world *ABCA4* cohort from a nationwide telehealth genetic counseling practice using test results and available patient characteristics.

Methods

Retrospective chart review of individuals with ≥1 pathogenic or likely pathogenic *ABCA4* variant referred to a nationwide telehealth genetic counseling

Study timeframe: 2019–2025

Phenotype classified based on available clinical history as confirmed Stargardt, possible Stargardt, or no Stargardt



***ABCA4* Genetic Testing in a Nationwide Real-World Genetics Services Cohort**

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Results

N	Median Age	Sex
1174 patients	**35.2y** (IQR 30.6-41.9y)	**66.4%** Female (779/1174)

Patients from **48** US states, most frequently:

- CA: 15.6% (183/1174)
- NY: 12.5% (147/1174)
- FL: 11.1% (130/1174)
- WA: 8.1% (95/1174)
- TX: 6.0% (71/1174)

Genetic test type:

- Reproductive Carrier Panel: 68% (800/1174)
- Retinal Panel: 29% (339/1174)
- Other: 3% (35/1174)

Most frequent variants in patients with ≥2 variants):
p.Gly863Ala: 26% (40/155)
p.Gly1961Glu: 23% (35/155)

Conclusion

- This nationwide real-world cohort is unselected and may provide less-biased data than previously published cohorts derived from academic centers and clinical trials.
- Confirmed or possible Stargardt occurred in patients with one variant (18%) and patients with two variants tested for carrier screening (12%).