# RareVision

Turning DNA data
into action with
**whole genome** insights

INOCRAS

FIND ANSWERS – FASTER

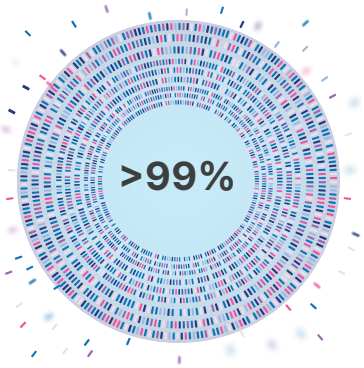
# Identifying your risk can change your future

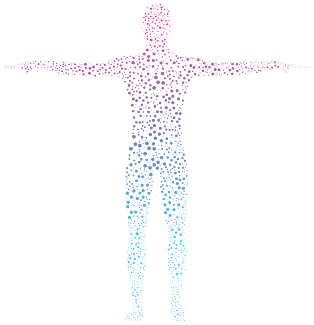
Almost 40% of rare disease patients[1] received a misdiagnosis in their journey. This delays your ability to start effective treatments for rare disease symptoms that may get worse over time.

**RareVision equips you with more information about what might be causing your or your child's symptoms, helping narrow down your diagnosis and may lead to a quicker, more accurate diagnosis.**

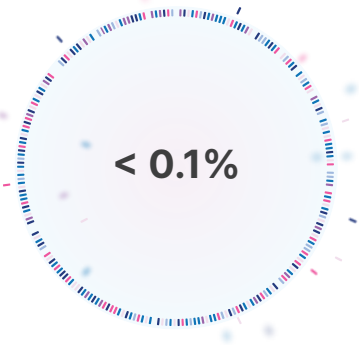
For the same patient

**Whole Genome**



Conventional panel tests




**Whole Genome Sequencing**
>99% genome-wide coverage

We look at the whole picture of your genomic blueprint, not just a small part of it. This allows us to find not only simple changes to your DNA, but complicated ones too. When we see more, we get more insights.

**Targeted Panel Sequencing**
<0.1% genome-wide sequencing

Looking at a much smaller portion of your genetic blueprint could mean missing key changes to your DNA, which could lead to a delayed or missed diagnosis.

RareVision

# Take control of your health to take control of your future

Get the whole picture of your genetic health and more insights about your genes that could contribute to rare diseases, all within two weeks.*

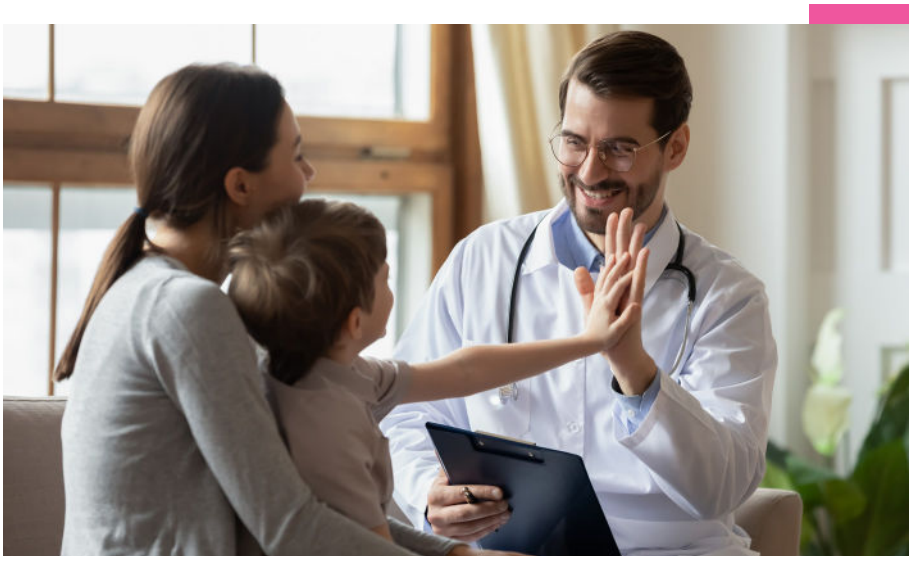
- **Whole genome sequencing** that looks at your genes to assess your risk of rare disease
- **Personalized support from a genetic counselor** to help you understand the test findings and answer your questions
- **Actionable insights** to help you take control of your health
- 2 week turnaround time*

**Price: $2,400†**

*2 weeks from receipt of your blood samples at our lab.
†The listed price is a general estimate. The price may vary depending on your country and the specific scope of services we provide tailored to your needs.

## DID YOU KNOW?

It can take an average of 4-5 years[2] to get a diagnosis of a rare disease. Our whole genome test looks at the bigger picture of your genes for a more holistic understanding of how rare disease could affect your health, so you get the answers you need – faster.



[1] https://rarediseases.org/wp-content/uploads/2022/10/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf
[2] Willmen T, Willmen L, Pankow A, Ronicke S, Gabriel H, Wagner AD. Rare diseases: why is a rapid referral to an expert center so important? BMC Health Serv Res. 2023 Aug 23;23(1):904. doi: 10.1186/s12913-023-09886-7. PMID: 37612679; PMCID: PMC10463573.

STORIES WITH IMPACT

# Andrew's story

Andrew, a 1-year-old boy, was experiencing a myriad of perplexing symptoms, including decreased muscle tone, facial abnormalities, and developmental delays. With the hope of finding answers, Andrew's provider ordered the RareVision test. The results revealed a change to his DNA that supported a definitive diagnosis of a rare genetic disorder called Noonan syndrome.

**This finding provided a roadmap for his future care that allowed his family to take proactive steps in managing his health and to make empowered family planning decisions.**

# Chloe's story

Chloe, 17, had severe symptoms that affected her intestines. Doctors suspected inflammatory bowel disease (IBD), but finding the exact cause remained a challenge.

Her results from RareVision revealed two disease-causing changes to her DNA that confirmed her diagnosis of early-onset IBD. **She learned that IBD caused by her mutations can be treated, and even cured, through hematopoietic stem cell transplantation. She is now actively preparing for transplantation, a potential treatment for her condition.**



*While all patient names provided are fictionalized to ensure privacy and confidentiality, the age, disease information, and diagnostic process in the testimonials are based on real cases. Please note that clinical outcomes may vary for each individual. These patient stories are based on collaborative research funded by Inocras.*

# How can I order Rare**Vision**?

## Sign up

Fill out the online order request form and consent form at **inocras.com**.

## Clinician approval

An external clinician will review and order your test request.

## Blood sample collection

Once your test order is made, we'll send you a link to make an appointment with a phlebotomist, who will visit your home and draw your blood.

## Genetic counselor appointment

When your report is ready, you will receive a notification with a link to make an appointment with a genetic counselor who will review your results with you.

## Result review

A genetic counselor will review your Rare**Vision** report with you.

**Scan the QR code to get started**

RareVision

# What you'll receive

**An comprehensive, easy-to-understand testing report** that provides an overview of your genetic risk for rare disease

**Personalized support from a genetic counselor** to help you understand your test findings and answer your questions

**Actionable insights** to help you take control of your health

RareVision

John Smith
Patient ID: H23/028662

**Patient**
Name: John Smith
Patient ID: H23/028662
Sex at birth: Male
Date of birth: Nov 20, 1987

**Physician**
Name: John Doe
Institution: Heritage Medical Center
Contact: +82-10-0000-0000
Address: 1600 Amphitheatre Parkway, Mountain View, CA 94043

**Specimen**
Specimen ID: C346399(9)
Specimen type: Blood
Specimen collection: Blood draw
Collected: Nov 20, 2023, 11:23
Received: Nov 24, 2023, 13:20
Clinical diagnosis: Autism spectrum disorder

**Test Information**
Test name: Whole genome analysis and interpretation
Quality: Satisfactory
Sequencing mean depth: 48.2x

**RESULT SUMMARY**

| | |
|---|---|
| Positive variants detected | *PTEN* |
| Inconclusive variants detected | *SCN2A, TCF7L2* |
| Secondary (incidental) findings detected | *BRCA1* |

**A. POSITIVE VARIANTS**

| Gene | Variant type | Zygosity | ACMG classification | Related diseases |
|---|---|---|---|---|
| *PTEN* | SNV | Heterozygosity | Likely pathogenic | Lhermitte-Duclos disease |

**B. INCONCLUSIVE VARIANTS**

| Gene | Variant type | Zygosity | ACMG classification | Related diseases |
|---|---|---|---|---|
| *SCN2A* | SNV | Heterozygosity | Pathogenic | Developmental and epileptic encephalopathy 11 |
| *TCF7L2* | SV | Heterozygosity | - | Diabetes mellitus, type 2, susceptibility to |

**C. SECONDARY (INCIDENTAL) FINDING**

| Gene | Variant type | Zygosity | ACMG classification | Related diseases |
|---|---|---|---|---|
| *BRCA2* | SNV | Heterozygosity | Likely pathogenic | Breast-ovarian cancer, familial, 2 |

**D. INTERPRETATION**

- In *PTEN*, variant ENSP00000278317.6:p.R234H is detected as likely pathogenic, and the variant is identified as heterozygous. And the variant is identified as de novo.
- *PTEN* is known to be associated with the following diseases.
  Lhermitte-Duclos disease (Autosomal dominant) (PMID : 24102544, 21926107).
- Incidentally, in *BRCA2*, variant ENSP00000262426.4:p.F85L is detected as likely pathogenic, and the variant is identified as heterozygous. And the variant is identified as de novo.
- *BRCA2* is known to be associated with the following diseases.
  Breast-ovarian cancer, familial, 2 (Autosomal dominant) (PMID: 17924331).
- Geneticist evaluation for clinical correlation of these results is recommended.

*SNV; Single nucleotide variant, INDEL; Insertion and deletion, SV; Structural variation, TE; Transposable elements, DEL; Deletion, DUP; Duplication, INV; Inversion, BND; Translocation, Chr; Chromosome, AD; Autosomal dominant, AR;Autosomal recessive; XR, X-linked recessive

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Request an order for **RareVision** testing today

at **inocras.com**

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Clinical Laboratory Improvement Amendments

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COLLEGE of AMERICAN PATHOLOGISTS

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