

Bridging Genetics and Ophthalmology; The Oculogenetics Patient Pathway

Basamat AlMoallem | MD, PhD

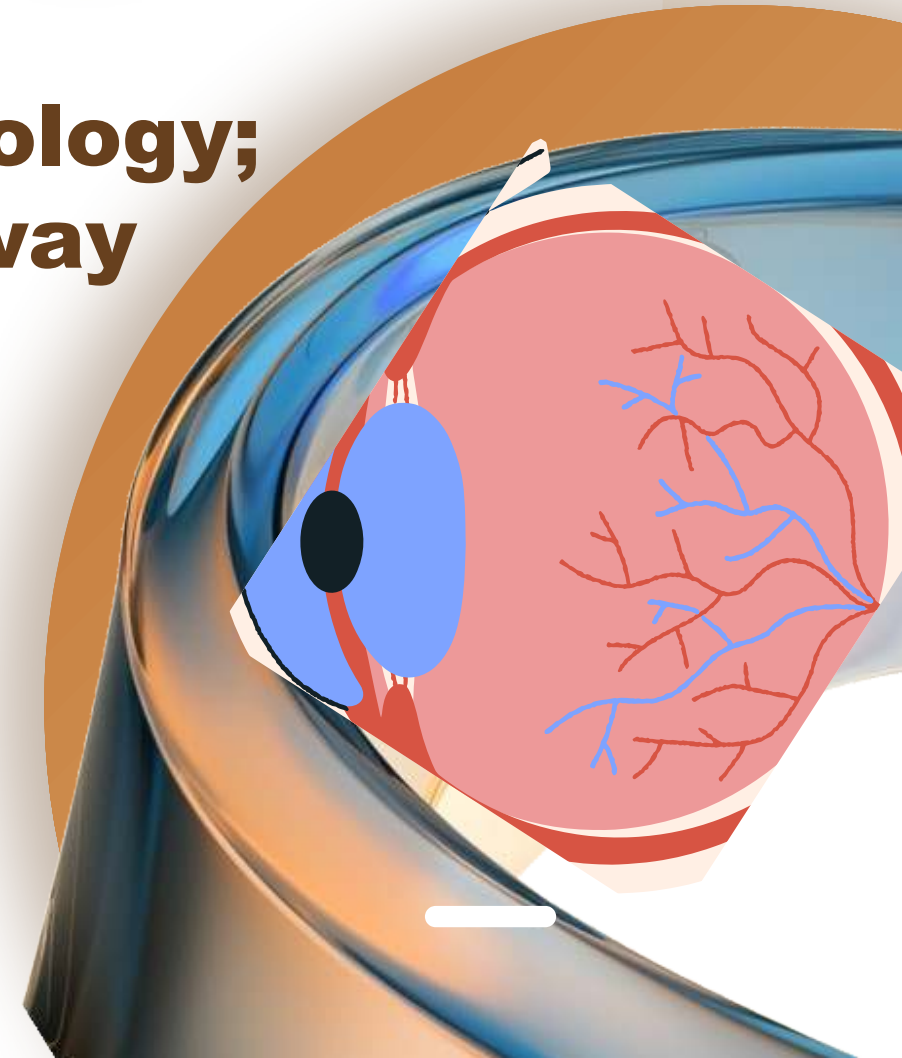
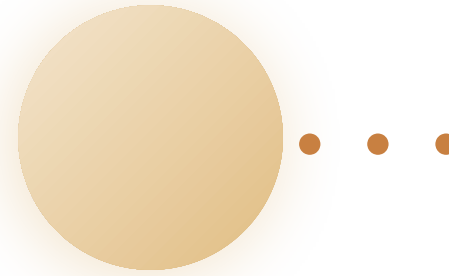
Executive board member of directors | Saudi Ophthalmology
society

Assistant Professor | Consultant Ophthalmic Geneticist
Ophthalmology Department | College of Medicine

King Saud University | King Saud University Medical City

Riyadh | Saudi Arabia

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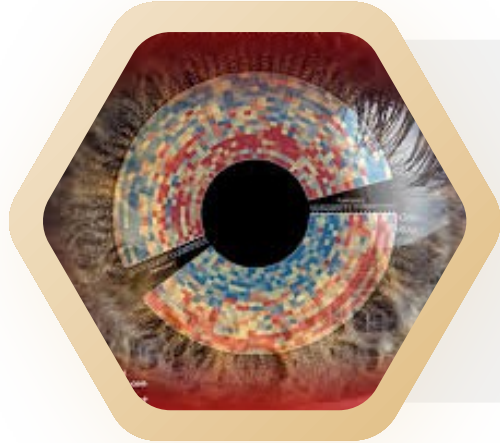


01

Introduction to Oculogenetics



Definition and Scope



Understanding Oculogenetics

Oculogenetics is the study of genetic factors that contribute to eye disorders, encompassing mutations, hereditary patterns, and their molecular mechanisms affecting vision.

Importance in Ophthalmology

The integration of oculogenetics into ophthalmology is crucial for accurate diagnosis, personalized treatment approaches, and potential gene therapies for inherited eye conditions.



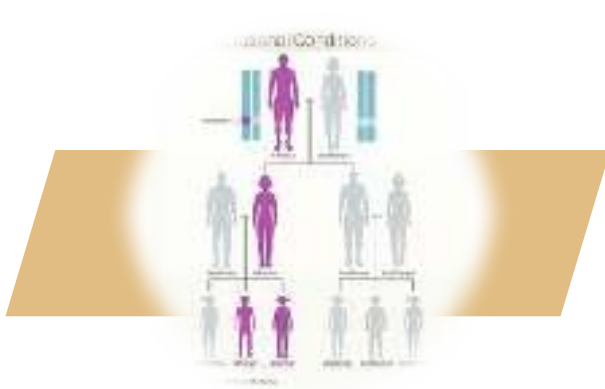


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Genetic Testing in Ophthalmology

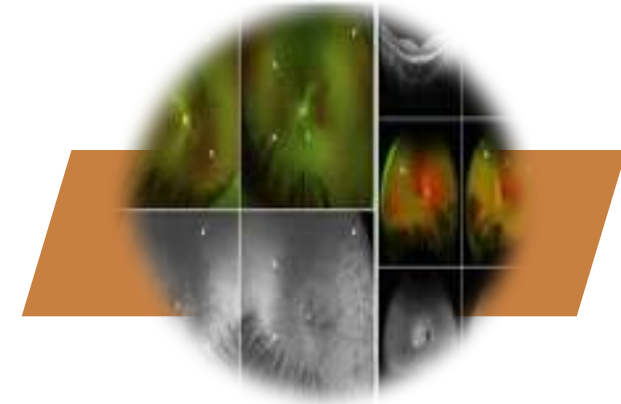


Indications for Genetic Testing



Family History and Genetic Disorders

A detailed family history may indicate a hereditary pattern of eye disorders, prompting genetic testing to identify at-risk individuals and facilitate early detection and management.



Phenotypic Variability in Eye Diseases

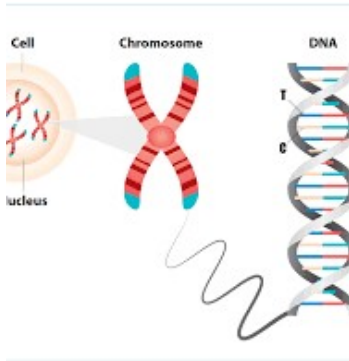
Phenotypic variability in eye diseases may suggest genetic influence; genetic testing can help delineate specific conditions and tailor treatment strategies based on individual patient profiles.

Types of Genetic Tests



Diagnostic Testing

Diagnostic testing helps to confirm or rule out specific genetic conditions, aiding in the identification of the underlying cause of eye diseases and guiding treatment options.



Predictive and Carrier Testing

Predictive and carrier testing assesses an individual's risk of developing eye diseases based on genetic predispositions, providing valuable information for family planning and early intervention.



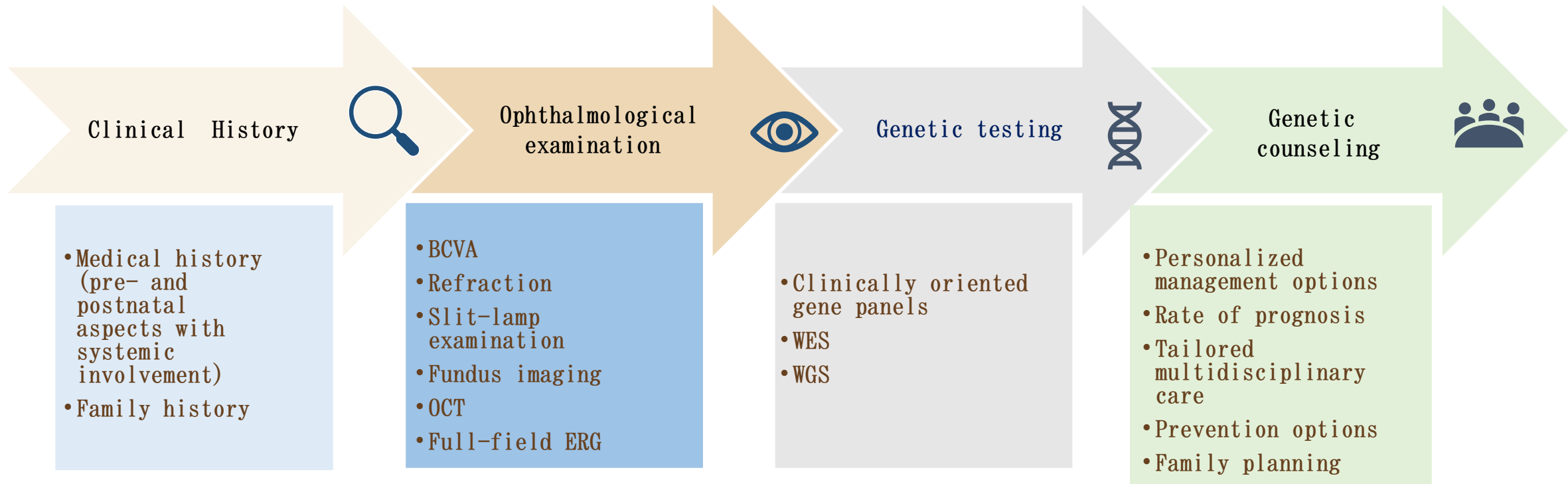


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The Oculogenetics Patient Pathway



Ophthalmic Genetics Clinic workflow



Important to resolve cases with inconclusive data, such as VUS, missing heritability, or novel findings



04

Management of Genetic Eye Diseases



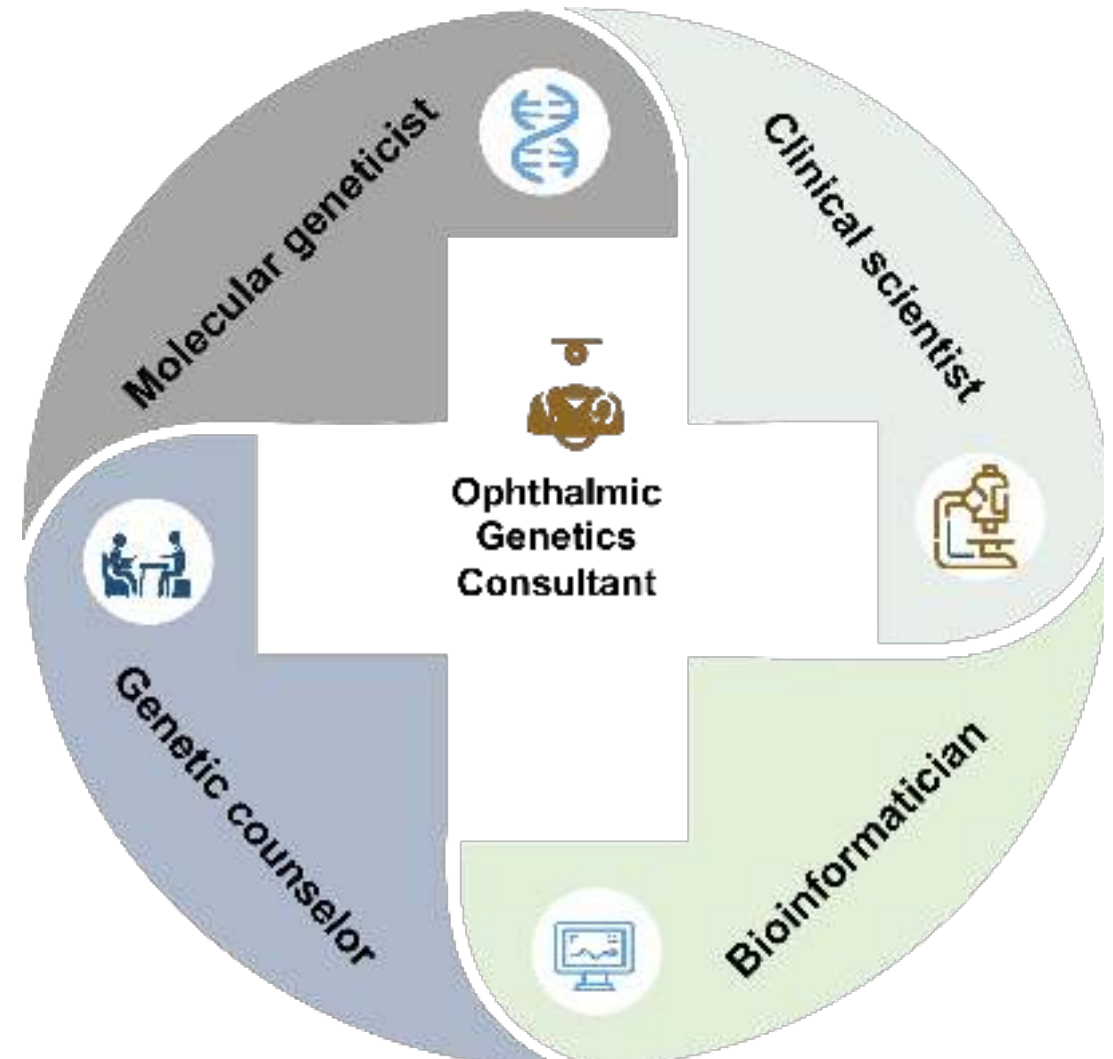
Multidisciplinary Approach

Collaboration Between Geneticists and Ophthalmologists

Effective management of genetic eye diseases requires a seamless collaboration between geneticists and ophthalmologists to ensure comprehensive care and tailored treatment strategies for patients.

Role of Allied Health Professionals

Allied health professionals play a vital role in supporting patient care by providing rehabilitation services, counseling, and educational resources to enhance the overall treatment experience for those with genetic eye diseases.





05

Clinical case and Ethical Considerations in



Clinical case

5 years old girl who is referred for Ophthalmic Genetic Assessment as case of sensory Nystagmus

History of Present Illness

Nystagmus and low vision

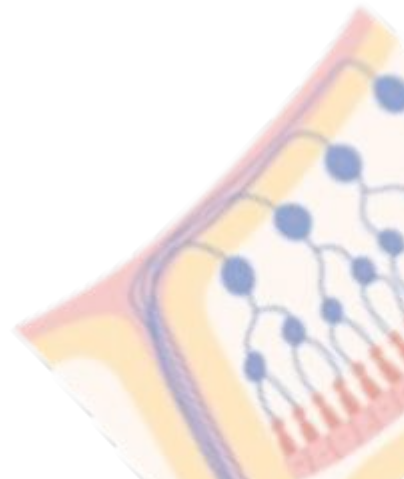
Systemic: Normal

Family pedigree:

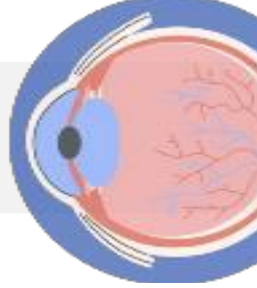
- Parents are first degree relatives
- Affected brother and maternally related cousin

She was booked for MRI brain + orbit + MRV

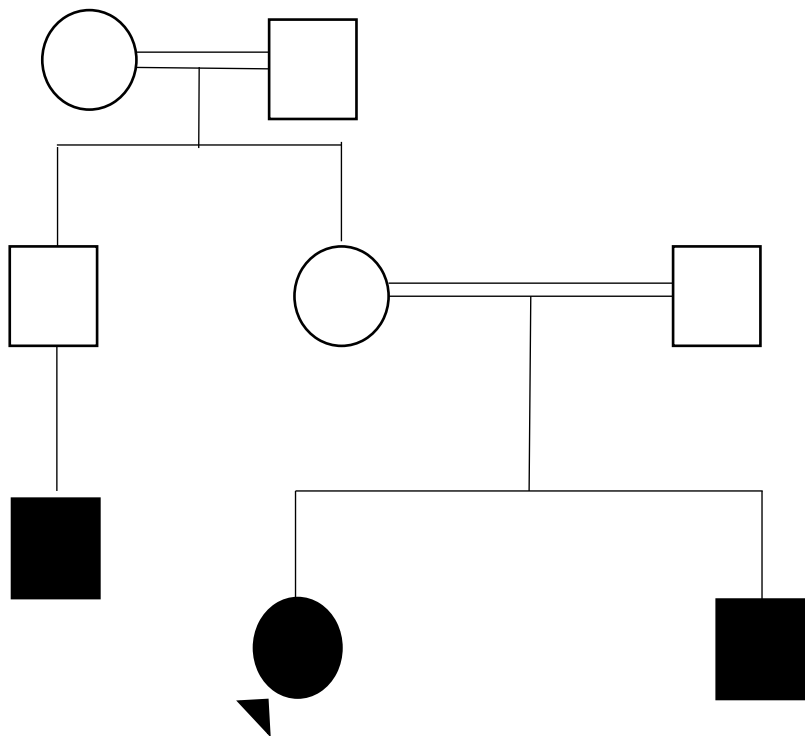
She was then referred as a case of sensory Nystagmus to the ophthalmic genetic clinic at the Department of Ophthalmology, KAUH, Riyadh, Saudi Arabia



Clinical case

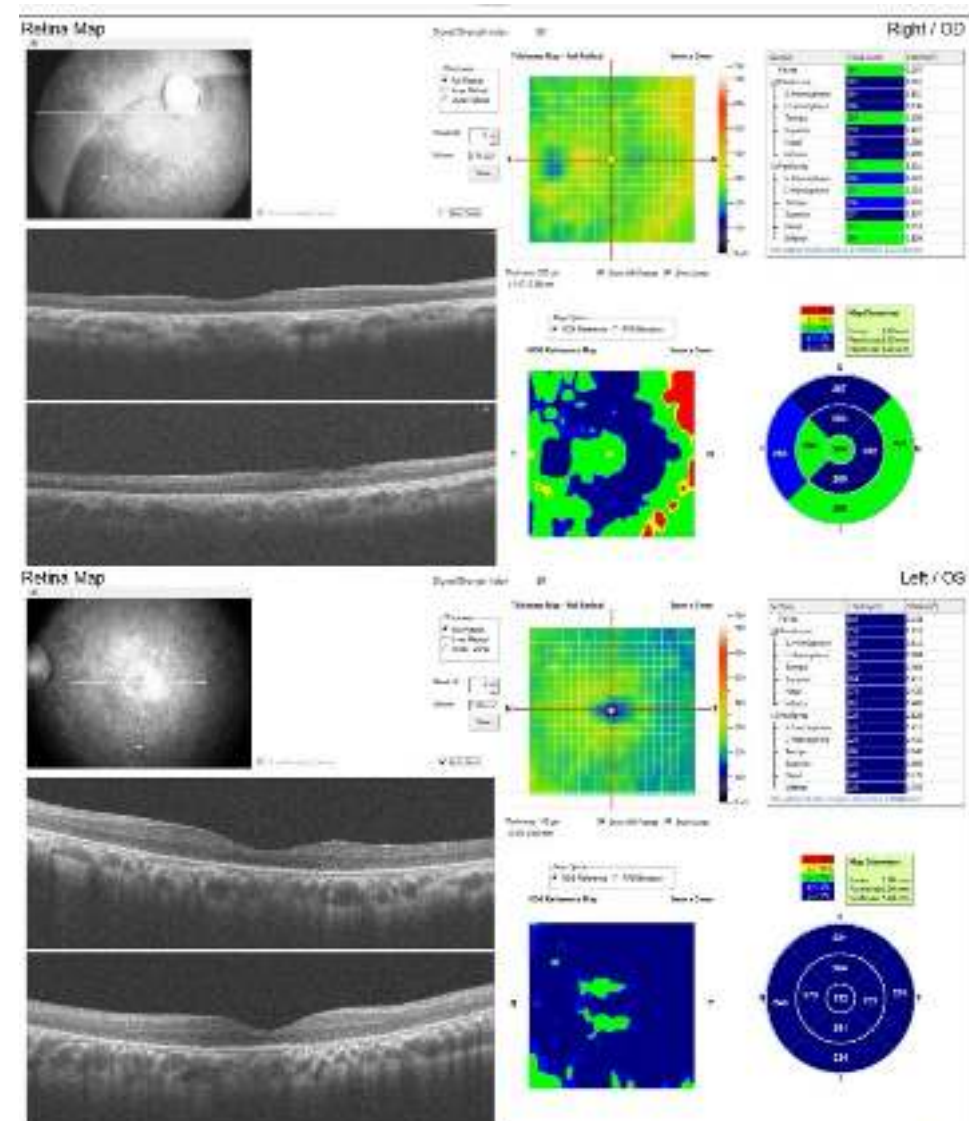
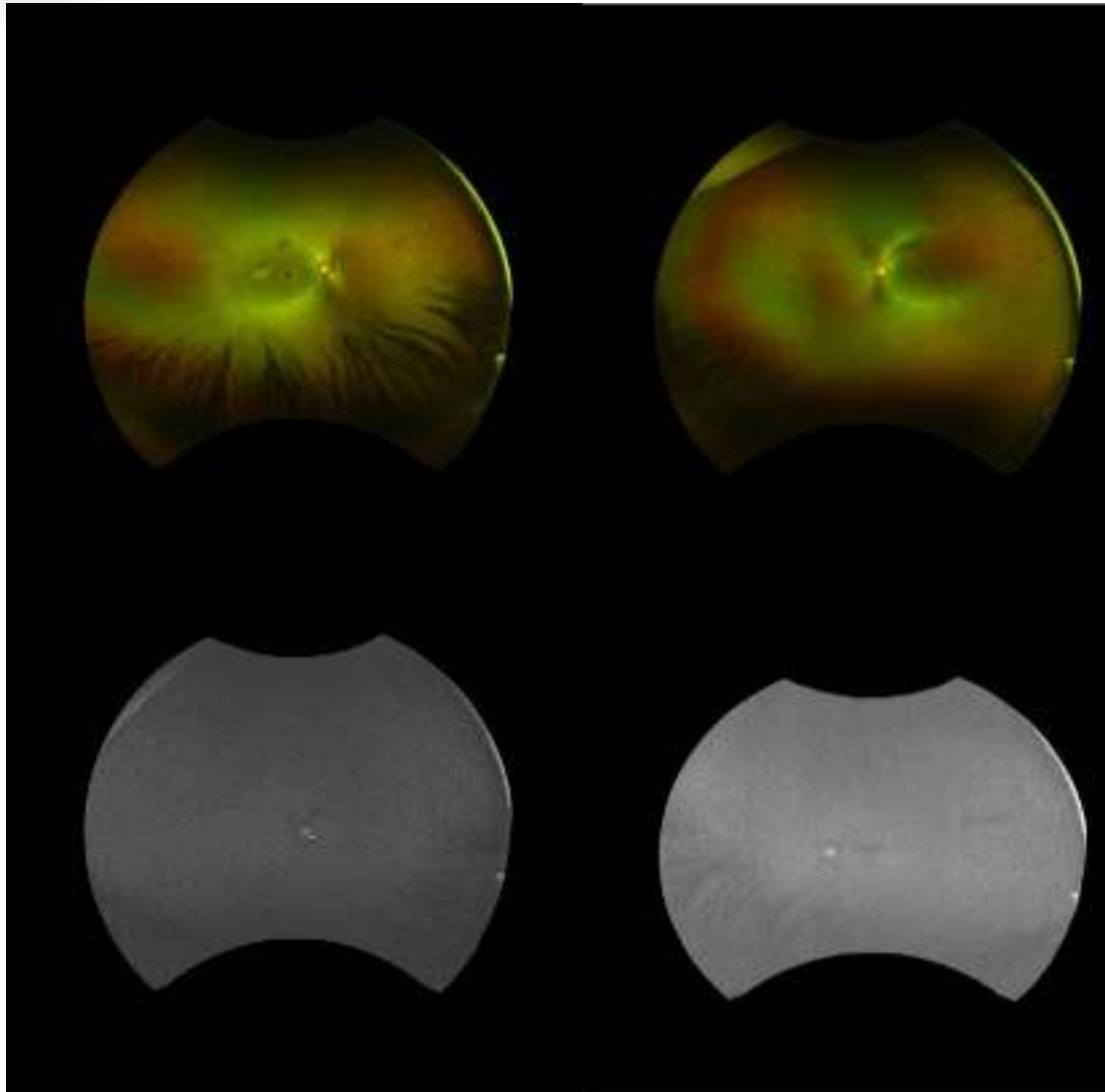
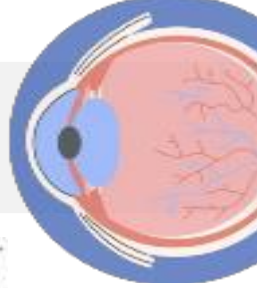


Slit-lamp examination

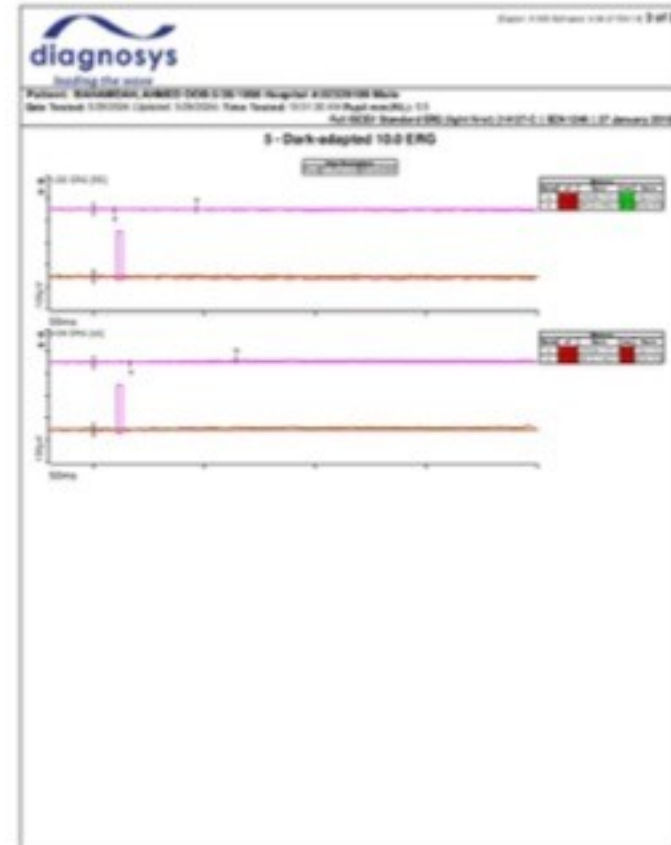
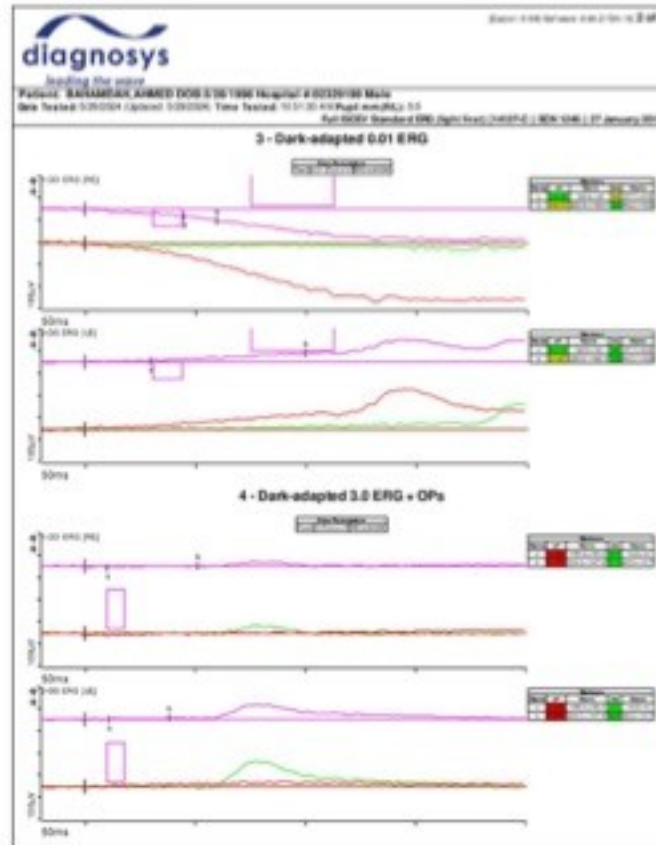
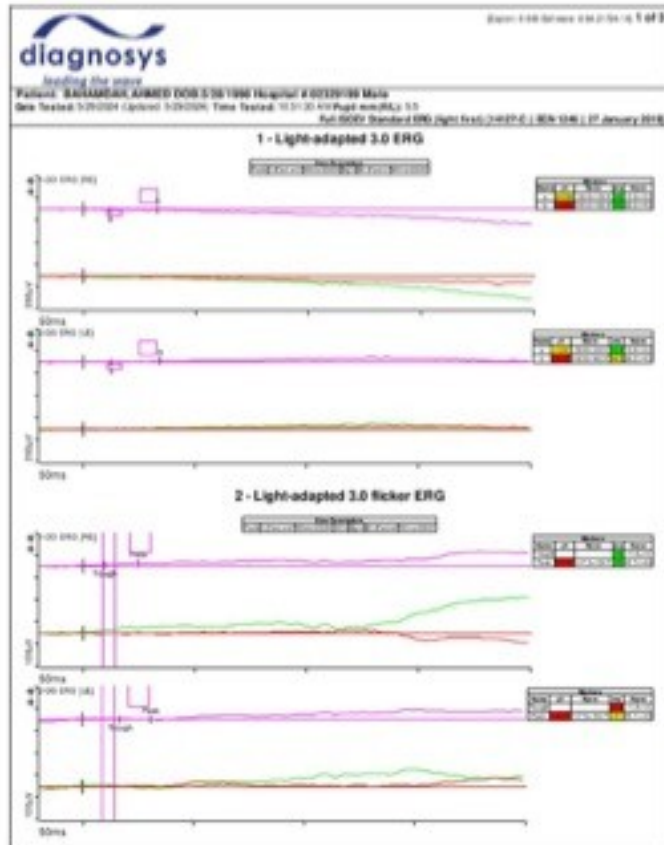
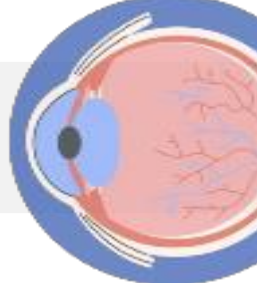


Right eye	THE STUCTURE	Left eye
F&F	BCVA	F&F
-9. 00-4. 00x10	Refractive	-10. 00-4. 00x150
NORMAL	Red reflex	NORMAL
NORMAL	LID	NORMAL
NORMAL	CONJECTIVA	NORMAL
CLEAR	LENS	CLEAR
CLEAR	CORNEA	CLEAR
Horizontal jerky with torticollis	Nystagmus	Horizontal jerky with torticollis

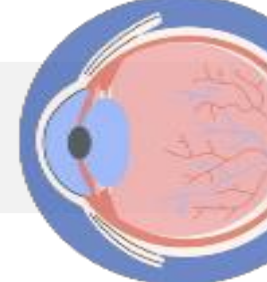
Clinical case



Clinical case



Clinical case



Additional report recipient(s): Dr. Basamat Al Moallem, King Abdullah University Hospital, Genetics Ophtalmology, Rlyadh, Saudi Arabia

Test(s) requested: CentoXome® Solo

CLINICAL INFORMATION

Nystagmus; Reduced visual acuity; Retinal degeneration; Rod-cone dystrophy
(Clinical information indicated above follows HPO nomenclature.)

Age of onset: 1 month(s).

Family history: Yes.

Siblings affected.

Consanguineous parents: Yes.

Clinician suspects: Leber congenital amaurosis 2. Rule out request(s): rod-cone dystrophy.

Targeted gene request(s): RPE65.

Reason for this additional report: the previous report was issued with the incorrect requested test name and methods section. This information is hereby adjusted. The final diagnosis remains unchanged. This report supersedes the report from 28 Sep. 2022.



POSITIVE RESULT
Pathogenic variant identified

INTERPRETATION

A homozygous pathogenic variant was identified in the **RPE65 gene**. This finding confirms the genetic diagnosis of autosomal recessive Leber congenital amaurosis type 2.

No further clinically relevant variants related to the described phenotype were detected.

Data Privacy and Genetic Information



Confidentiality of Genetic Data

Protecting the confidentiality of genetic information is paramount, as unauthorized access can lead to misuse and discrimination, jeopardizing patients' trust in healthcare systems.



Implications for Family Members

The results of genetic testing not only affect the individual but can also have significant implications for family members, including risk assessments and the potential for hereditary conditions.

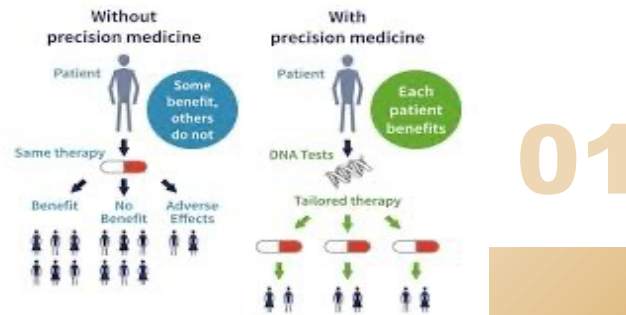


06

Future Perspectives in Oculogenetics



Implications for Personalized Medicine




Tailoring Treatments Based on Genetic Makeup

Personalized medicine leverages genetic information to customize treatment plans for individuals, optimizing efficacy and minimizing adverse effects in managing ocular conditions.



The Role of Artificial Intelligence in Oculogenetics

Artificial Intelligence (AI) assists in analyzing vast genetic data, identifying patterns, and predicting disease progression, thereby enhancing diagnostic accuracy and treatment strategies in oculogenetics.



Genetics is now central to IRD care.
From diagnosis to therapy, oculogenetics offers;
PRECISION, HOPE, AND PROGRESS.
Let's bridge the gap together.



Thanks

