Inherited Eye Diseases: Evaluation of current approaches and future needs of ophthalmologists in Turkey

Dear Participant,
This survey aims to evaluate the clinical practices of ophthalmologists in Turkey for inherited eye diseases, their level of knowledge about genetic diagnostic and treatment options, and their needs for approaching inherited eye diseases.

The information you provide during your participation in this study will be kept strictly confidential. The information you give personally will be collected and processed for research purposes only. The accuracy of the information you provide is essential for the quality of the research. The survey duration is, on average, 6-8 minutes. Thank you for your participation and candid responses.

I. Demographic features and occupational characteristics

1. Please indicate the year you started working as an ophthalmologist.

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2. Please indicate your academic degree.

- Specialist
- Instructor
- Assistant professor
- Associate professor
- Professor

3. Please tick your main practice site.

- Private clinic
- Private hospital
- State hospital
- Training and research hospital
- University-foundation hospital (Public)
- University-foundation hospital (Private)

4. Please indicate the city you are practicing

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5. Please select if there is a subspecialty unit in the institution you work for (Select all that apply)

- There is no subspecialty unit in my institution, I am practicing in general ophthalmology.
- Posterior segment (Medical retina, vitreoretinal surgery, uvea)

- Glaucoma
- Anterior segment (Cataract, refractive surgery, cornea)
- Pediatric ophthalmology
- Neurophthalmology
- Oculoplastic surgery
- Low-vision rehabilitation
- Electrodiagnostic unit
- Other:...

6. Is there a medical genetics specialist in your facility?

- Yes
- No
- I do not know

II. Current clinical practices

7. How often do you take a family history?

- Always
- Usually
- Sometimes
- Rarely
- Never

8. How many patients do you see per week with a suspected inherited eye disease?

- 0
- 1-2
- 3-5
- 5-10
- >10

9. Do you have a major role in managing patients with an inherited eye disease?

- Yes
- No (Go to question 20)

10. Is there a standard approach you use in your clinic for a patient with an inherited eye disease?

- Yes
- No

11. Which of the following professionals or organizations do you receive support from during management of patients with inherited eye diseases? (Select all that apply)

- Subspecialists in ophthalmology
- Ophthalmologists interested in ocular genetics
- Low vision rehabilitation specialists
- Social services
- Patient organizations
- International researchers or specialists
- I do not ask for support

12. Do you question and try to reveal the inheritance pattern in patients with inherited eye diseases?

- Yes (Go to question 14)
- No

13. If your answer to the previous question is No, which of the following is the reason(s)? (Select all that apply)

- This information does not affect the diagnosis, treatment, and follow-up method I use for the patient
- I do not have enough time to allocate to this information
- I do not think the patient's answers are sufficient or reliable for this information
- I do not feel competent enough to reach this information.
- Other:...

14. Do you recommend genetic diagnostic tests to the patients with inherited eye diseases?

- Yes
- No

Please branch based on the response to #14

14a. If your answer is "Yes", how do you direct the patient to genetic diagnostic tests? (Select all that apply)

- I order the genetic diagnostic tests myself and do not get help.
- I order the genetic diagnostic tests myself, but I get help from a medical geneticist or another ophthalmologist.
- I refer the patient to a medical geneticist, and they do the testing.
- I refer the patient to an ophthalmologist for genetic testing who deals with hereditary eye diseases.
- I refer the patient to a private genetic diagnostic center.
- Other:....

14b. If your answer is "No", which of the following is the reason(s)? (Select all that apply)

- Genetic diagnostic tests do not change my patient management strategy, so I follow up without getting tested.
- There is no center near my vicinity where genetic diagnostic tests are performed
- I think genetic diagnostic tests are too expensive
- I do not have enough information about genetic diagnostic tests
- My facility does not have a medical geneticist that I can cooperate for genetic testing
- Other: ....

15. Do you communicate the genetic test results to your patients?

- Yes
- No

Please branch based on the response to #15

15a. If your answer is "Yes", to what extent do you feel competent in the face of the reactions and questions you will encounter during this statement?

- Not at all
- A little
- Moderately
- Very comfortable
- Completely comfortable

15b. If your answer is "No", who shares the genetic diagnosis test results with the patient?

- The medical geneticist or the staff at the center where the test is performed explains the test results to the patient.
- The ophthalmologist I cooperate with explains the results to the patient.
- Patients learn the results from the result document in their hands.
- Other:....

16. Do your patient or patient relatives request genetic counseling?

- Yes
- No

17. Do you refer your patient for genetic consultation?

- Yes (Go to question 19)
- No

18. If your answer to the previous question is No, which of the following is the reason(s)? (Select all that apply)

- My patients do not request genetic counseling.

- I am competent to provide genetic counseling services, and I provide this service when necessary.
- I do not know who provided the genetic counseling service.
- There is no specialist in our center or the immediate vicinity that I can refer my patients to for this service.
- I find genetic counseling services unnecessary.
- Other:....

19. After diagnosing inherited eye disease, which of the following treatment and follow-up options do you apply? (Select all that apply)

- I recommend the patient the appropriate standard medical or surgical treatment options.
- I search for treatment options in the clinical trial phase and refer the patient if I find a suitable trial.
- I regularly follow up with patients who do not have a standard treatment option.
- I save the patient contact information and inform them that if there is any development in treating their diseases
- I offer suitable patients the low vision rehabilitation service in my clinic
- I refer the patients eligible for the low vision rehabilitation option to centers where this service is provided.
- Other:.....

III. Knowledge about genetics and available healthcare services

20. How have you learned about genetics? (Select all that apply)

- Medical school
- Residency
- Continuing education
- Study abroad
- Master or PhD in genetics
- Other...

21. Which of the following eye diseases should be referred to genetic diagnostic tests? (Select all that apply)

- Childhood nystagmus
- Congenital glaucoma
- Congenital/juvenile cataract
- Corneal dystrophies
- Retinopathies
- Age-related macular degeneration
- Diabetic retinopathy
- Retinablastoma
- Eye diseases with malformations or syndromes

22. Which of the following genetic diagnostic tests do you have information about? (Select all that apply)

- Targeted mutation (allele-specific) analysis
- Next generation sequencing
- Sanger sequencing
- Microarray
- Deletion/dupplication analysis
- Whole exome sequencing
- Whole genome sequencing
- I have no understanding of any of the following types of genetic testing

23. Do you know which genetic tests are available in your facility or province?

- Yes
- No

24. Do you know which genetic diagnostic tests the health insurance system covers?

- Yes
- No

25. To what extent can you follow scientific developments and clinical studies in hereditary eye diseases or ocular genetics?

- Never
- Rarely
- Sometimes
- Usually
- Always

IV. Education activities and future needs for better healthcare

26. Do you find the training after residency (continuing education) in inherited eye diseases sufficient?

- Not enough at all
- Not enough
- A little
- Sufficient
- Very sufficient

27. Which of the following do you prefer for continuing education in inherited eye diseases? (Select all that apply)

- It should include up-to-date information and new developments.
- There should be training sessions that are repeated and updated at regular intervals
- It should consist of approaches that are currently available in Turkey
- It should encourage and guide cooperation
- It should contain basic information on the field of genetics
- It should include clinical studies and practices in other countries
- Other:....

28. Is it necessary to establish a sub-unit within the Turkish Ophthalmology Association and Medical Genetics Association that organizes training activities in ophthalmic genetics, closely following the developments in this field and encouraging cooperation and research in this field?

- Yes
- No

29. If a team was formed to improve the service provided to patients and their relatives with inherited eye diseases, who should be included in this team? (Select all that apply)

- Ophthalmologist
- Medical geneticist
- Low vision rehabilitation specialist
- Social services specialist
- Nurse
- An ophthalmologist trained in ocular genetics
- Other:....