

# Inherited Retinal Degenerations (IRD) Survey

EVICR.net is conducting an International Survey entitled “*Current management of Inherited Retinal Degenerations (IRD) subjects in Europe*” in a research collaboration with Novartis.

This IRD Survey is coordinated by EVICR.net Coordinating Centre (AIBILI) and is developed by the IRD Survey Expert Committee: Brigit Lorenz (BL), MD PhD, from CS065, Scientific Coordinator and the following members: Hendrik Scholl, MD PhD, from CS110; Isabelle Audo, MD PhD, from CS006; Ingeborgh van den Borg, MD PhD, from CS040, and João Pedro Marques, MD PhD, from CS001.

Please note that all information provided is confidential and will only be used in the EVICR.net network with regard to Retinal Dystrophies.

## ***What is the purpose of this IRD Survey?***

The purpose of the IRD Survey is to conduct the first international survey to understand the management of IRD cases (including on the genotype and phenotype experiences of bi-allelic RPE65 patients) across Europe.

## ***Why to participate in this IRD Survey?***

- Even if your centre does not manage IRD patients, please reply only to question 1 of Section 1- IRD Demographics, as we need to have also this information to understand the management of IRD cases in Europe.

- If your centre manages IRD patients, replying to this survey has the following benefits:

1. Co-authorship in the IRD Survey results Publication as part of the Study Group.

2. If you complete only Sections 1 and Section 2 of the survey, your centre will benefit of 100 € discount in the next EVICR.net annual Membership fee.

3. If you complete all Sections (1 to 5) of the survey, your centre will benefit of 200 € discount in the next EVICR.net annual Membership fee.

## ***Until when to participate in the IRD Survey?***

We ask your collaboration by **filling out this questionnaire before June 7, 2019**.

**Counting on your collaboration with this survey. Thank you in advance,**

Birgit Lorenz  
Scientific Coordinator of the IRD Survey Expert Committee  
There are 112 questions in this survey.

**EVICR.net Clinical Site no.:**

See list of member available  
at <https://www.evicr.net/clinical/member-list/>  
(<https://www.evicr.net/clinical/member-list/>).

\*

Please write your answer here:

**Name: \***

Please write your answer here:

**Function: \***

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Ophthalmologist

Pediatric ophthalmologist

Medical retina specialist

Other

**Phone: \***

❗ Only numbers may be entered in this field.

Please write your answer here:

Email address: \*

Please write your answer here:

## IRD Demographics

Please select the option(s) that reflects your Clinical Site experience with IRD patient's management. Note that your answers should be estimations (no database or patients clinical history consultation).

1. How many IRD patients are currently managed by your centre?

\*

**i** Choose one of the following answers

Please choose **only one** of the following:

- None
- <10
- 10 - 19
- 20 - 49
- 50 - 99
- 100 - 199
- 200 - 499
- 500 - 999
- > 1000
- Don't know

## 2. How do you manage IRD patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

📌 Check all that apply

Please choose **all** that apply:

- Refer IRD patients to expert centres
- Refer IRD patients to ERN-EYE Health Care Providers (HCP)
- Manage IRD patients at our centre

## 3. Do you have a database for IRD patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

Please choose **only one** of the following:

- Yes
- No

## 3a. What type of database do you have for IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '8 [H]' (3. Do you have a database for IRD patients?)

📌 Check all that apply

Please choose **all** that apply:

- Local webbased database
- National webbased database
- Local files (e.g. Excel)

Other:

### 3b. How many IRD patients are in your database? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '8 [H]' (3. Do you have a database for IRD patients?)

🗨️ Choose one of the following answers

Please choose **only one** of the following:

- < 100
- 100 - 499
- 500 - 1999
- 2000 - 4999
- > 5000
- Don't know

## 4. Who refers IRD patients to your centre?

Please provide an estimate of fractions in %.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

❗ Only numbers may be entered in these fields.

❗ The sum must be between 0 and 100

Please write your answer(s) here:

Patient self-referral

General practitioners

Pediatricians

Mother and child care centres

Ophthalmologists

Pediatric ophthalmologists

Medical retina specialists

Genetic counselors

## 5. What is the age range at IRD patient's first visit to your centre?

Please provide an estimate of fractions in %.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

- ❗ The sum must be between 0 and 100
- ❗ Only integer values may be entered in these fields.

Please write your answer(s) here:

< 3 years old

Pre-school (3 - 5 years old)

School (6 - 17 years old)

Young adult (18 – 30 years old)

Adult (> 30 years old)

## 6. What are the IRD patient's signs/symptoms implicating a visit at your centre?

Please provide an estimate of fractions in %.

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

❗ Only numbers may be entered in these fields.

	< 3 years old	3-5 years old	6-17 years old	18-30 years old	> 30 years old
<b>Reduced visual acuity</b>	<input type="text"/>				
<b>Nystagmus</b>	<input type="text"/>				
<b>Squint</b>	<input type="text"/>				
<b>Reduced visual field</b>	<input type="text"/>				
<b>Night blindness</b>	<input type="text"/>				
<b>Colour blindness</b>	<input type="text"/>				
<b>Metamorphopsia</b>	<input type="text"/>				
<b>Extra ocular abnormalities/suspicion syndrome</b>	<input type="text"/>				
<b>Familiar history</b>	<input type="text"/>				

## 7. What is the mean time between inquiry of appointment and first contact with a retina expert for IRD patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

🗨️ Choose one of the following answers

Please choose **only one** of the following:

- < 4 weeks
- Months
- Don't know

## 7a. How many months? \*

Only answer this question if the following conditions are met:

Answer was 'Months' at question '14 [N]' (7. What is the mean time between inquiry of appointment and first contact with a retina expert for IRD patients?)

🗨️ Only numbers may be entered in this field.

Please write your answer here:

## 8. What is the mean time between inquiry of appointment and final ophthalmological diagnosis for IRD patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

🗨️ Choose one of the following answers

Please choose **only one** of the following:

- < 4 weeks
- Months
- Years
- Don't know

### 8a. How many months? \*

Only answer this question if the following conditions are met:

Answer was 'Months' at question '16 [O]' (8. What is the mean time between inquiry of appointment and final ophthalmological diagnosis for IRD patients?)

❗ Only numbers may be entered in this field.

Please write your answer here:

### 8a. How many years? \*

Only answer this question if the following conditions are met:

Answer was 'Years' at question '16 [O]' (8. What is the mean time between inquiry of appointment and final ophthalmological diagnosis for IRD patients?)

❗ Only numbers may be entered in this field.

Please write your answer here:

### 9. What is the mean time between inquiry of appointment and final ophthalmological and genetic diagnosis for IRD patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

❗ Choose one of the following answers

Please choose **only one** of the following:

- Months
- Years
- Don't know

## 9a. How many months? \*

Only answer this question if the following conditions are met:

Answer was 'Months' at question '19 [P]' (9. What is the mean time between inquiry of appointment and final ophthalmological and genetic diagnosis for IRD patients?)

❗ Only numbers may be entered in this field.

Please write your answer here:

## 9a. How many years? \*

Only answer this question if the following conditions are met:

Answer was 'Years' at question '19 [P]' (9. What is the mean time between inquiry of appointment and final ophthalmological and genetic diagnosis for IRD patients?)

❗ Only numbers may be entered in this field.

Please write your answer here:

## 10. What is the current age range of the majority of the diagnosed IRD cases? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? )

❗ Choose one of the following answers

Please choose **only one** of the following:

- < 3 years old
- Pre-school (3 - 5 years old)
- School (6 - 17 years old)
- Young adult (18 – 30 years old)
- Adult (> 30 years old)
- Don't know

# Local Setting

## Section 2

## 1. What basic tests do you perform at your centre in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Refer IRD patients to ERN-EYE Health Care Providers (HCP)' or 'Refer IRD patients to expert centres' at question '7 [G]' (2. How do you manage IRD patients?)

🚫 Check all that apply

Please choose **all** that apply:

- Visual acuity
- Visual fields
- Fundus imaging
- Fundus autofluorescence (FAF)
- Retinal stratification (OCT)
- Electrophysiology (ERG, EOG, VEP, FST)

Other:

## 1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre? \*

Only answer this question if the following conditions are met:

Answer was 'Manage IRD patients at our centre' at question '7 [G]' (2. How do you manage IRD patients?)

🚫 Check all that apply

Please choose **all** that apply:

- Visual acuity
- Refraction
- Retinal stratification (OCT)
- Fundus autofluorescence (FAF)
- Fundus photography
- Electrophysiology (ERG, EOG, VEP, FST)
- Perimetry

Other:

## Which methods for visual acuity testing do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Visual acuity' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

🚫 Check all that apply

Please choose **all** that apply:

- ETDRS charts
- Snellen charts
- Number charts
- Landolt rings
- Tumbling 'E' charts
- Berkeley Rudimentary Vision Test (BRVT)
- Teller acuity cards
- Lea Symbols®
- Lea Symbols® low contrast test
- Pelli Robson contrast test

## Which devices / test for refractometry do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Refraction' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

🚫 Check all that apply

Please choose **all** that apply:

- Retinoscopy
- Autorefractometer

Other:

## Please specify the autorefractometer used in IRD patients. \*

Only answer this question if the following conditions are met:

Answer was 'Autorefractometer' at question '26 [U]' ( Which devices / test for refractometry do you use in IRD patients?)

❗ Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Nidek - Device:

Topcon - Device:

Reichert - Device:

## Which device for Optical Coherence Tomography (OCT) do you use in IRD patients?

\*

Only answer this question if the following conditions are met:

Answer was 'Retinal stratification (OCT)' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

❗ Check all that apply

Please choose **all** that apply:

Spectralis, Heidelberg Engineering

Cirrus HD-OCT, Zeiss

DRI OCT Triton, Topcon

OCT1 Maestro, Topcon

Other:

## Which Spectralis, Heidelberg Engineering do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Spectralis, Heidelberg Engineering' at question '28 [V]' (Which device for Optical Coherence Tomography (OCT) do you use in IRD patients? )

🚫 Check all that apply

Please choose **all** that apply:

- OCT
- ECT EDI
- OCT Angio

## Which device for Fundus Autofluorescence (FAF) recording do you use in IRD patients?

\*

Only answer this question if the following conditions are met:

Answer was 'Fundus autofluorescence (FAF)' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

🚫 Check all that apply

Please choose **all** that apply:

- Spectralis, Heidelberg Engineering
- F-10, Nidek
- Panoramix 200 Tx, Optomap, OPTOS

Other:

## Which device for ocular image recording do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Fundus photography' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

🚫 Check all that apply

Please choose **all** that apply:

Fundus camera

Wide-field fundus camera

## Please specify the fundus camera used in IRD patients. \*

Only answer this question if the following conditions are met:

Please indicate the device:

🚫 Comment only when you choose an answer.

🚫 If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose all that apply and provide a comment:

Zeiss - Device:

Nidek - Device:

Kowa - Device:

Topcon - Device:

Other:

## Please specify the Wide-field fundus camera used in IRD patients. \*

Only answer this question if the following conditions are met:

Answer was 'Wide-field fundus camera' at question '31 [Z]' ( Which device for ocular image recording do you use in IRD patients?)

🗨 Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Clarus - Device:

Optos - Device:

RetCam, Phoenix - Device:

Other:



## Which Electrophysiological tests do you use in IRD patients?

\*

Only answer this question if the following conditions are met:

Answer was 'Electrophysiology (ERG, EOG, VEP, FST)' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

🗨 Check all that apply

Please choose **all** that apply:

Ganzfeld-ERG (ffERG)

Multifocal ERG (mfERG)

Electrooculogram (EOG)

Visually Evoked Potential (VEP)

Dark adaptometry (DA)

Full-Field Light Sensitivity Threshold Testing (FST)

## Which Ganzfeld-ERG (ffERG) device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Ganzfeld-ERG (ffERG)' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

🗳️ Choose one of the following answers

🗳️ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Espion, Diagnosis LLC

RETIport/scan21, Roland consult

Nicolet Spirit

Other

## Which multifocal ERG (mfERG) device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Multifocal ERG (mfERG) ' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

🗳️ Choose one of the following answers

🗳️ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

VERIS, EDI inc.

RETIport/scan21, Roland consult

Other

## Which Electrooculogram (EOG) device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Electrooculogram (EOG) ' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Espion , Diagnosis LLC

RETIport/scan21, Roland consult

Nicolet Spirit

Other

## Which Visually Evoked Potential (VEP) device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Visually Evoked Potential (VEP) ' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Espion , Diagnosis LLC

RETIport/scan21, Roland consult

Nicolet Spirit

Other

## Which Dark adaptometry (DA) device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Dark adaptometry (DA)' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

🗳️ Choose one of the following answers

🗳️ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Espion , Diagnosis LLC

Roland Consult

Other

## Please specify the Full-Field Light Sensitivity Threshold Testing (FST). \*

Only answer this question if the following conditions are met:

Answer was 'Full-Field Light Sensitivity Threshold Testing (FST)' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

🗳️ Choose one of the following answers

🗳️ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

red, blue, white testing

white testing only

## Which Full-Field Light Sensitivity Threshold Testing device do you use in IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Full-Field Light Sensitivity Threshold Testing (FST)' at question '34 [AA]' ( Which Electrophysiological tests do you use in IRD patients?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

Espion , Diagnosis LLC

Roland Consult

Other

## Which methods of visual field testing do you use in IRD patients?

\*

Only answer this question if the following conditions are met:

Answer was 'Perimetry' at question '24 [S]' (1. Which parameters are applied for setting a clinical diagnosis of IRD at your centre?)

❗ Check all that apply

Please choose **all** that apply:

Kinetic perimetry

Static perimetry

Fundus-controlled perimetry

## Please specify the kinetic perimetry device used in IRD patients. \*

Only answer this question if the following conditions are met:

Answer was 'Kinetic perimetry' at question '42 [AB]' ( Which methods of visual field testing do you use in IRD patients? )

**!** Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Goldmann (manual)

Octopus - Device:

Other:



## Please specify the static perimetry device used in IRD patients. \*

Only answer this question if the following conditions are met:

Answer was 'Static perimetry' at question '42 [AB]' ( Which methods of visual field testing do you use in IRD patients? )

**!** Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Humphrey

Octopus - Device:

Twinfield

Other:

## Please specify the Fundus-controlled perimetry device used in IRD patients. \*

Only answer this question if the following conditions are met:

Answer was 'Fundus-controlled perimetry' at question '42 [AB]' ( Which methods of visual field testing do you use in IRD patients? )

📌 Check all that apply

Please choose **all** that apply:

MP1, Nidek

MP3, Nidek

Compass, Centrevue

Other:

## IRD Genetic testing and counselling

### Section 3

### 1. Are IRD patients from your centre tested genetically? \*

Only answer this question if the following conditions are met:

Answer was 'Manage IRD patients at our centre' at question '7 [G]' (2. How do you manage IRD patients?)

📌 Choose one of the following answers

Please choose **only one** of the following:

Yes

No

Don't know

## 1a. Why your centre is not testing IRD patients genetically? \*

Only answer this question if the following conditions are met:

Answer was 'No' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🔔 Check all that apply

Please choose **all** that apply:

Lack of funding

Hospital administration constraints

No practical benefit for patient

Other:

## 1b. As your Centre is not testing IRD patients genetically are you referring patients to other institutions/labs? \*

Only answer this question if the following conditions are met:

Answer was 'No' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

Please choose **only one** of the following:

Yes

No

## 2. Where is the genetic testing performed? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🔔 Check all that apply

Please choose **all** that apply:

In-house

Externally

Don't know

### If in house, where is the genetic testing performed? \*

Only answer this question if the following conditions are met:

Answer was 'In-house' at question '49 [AE]' (2. Where is the genetic testing performed?)

📌 Check all that apply

Please choose **all** that apply:

Clinical Care

Research Lab

### If externally, where is the genetic testing performed? \*

Only answer this question if the following conditions are met:

Answer was 'Externally' at question '49 [AE]' (2. Where is the genetic testing performed?)

📌 Check all that apply

Please choose **all** that apply:

Research Lab

National lab

International lab

### 3. Is your genetic testing laboratory certified? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

📌 Choose one of the following answers

Please choose **only one** of the following:

Yes

No

Don't know

#### 4. Which local/national guidelines or protocols do you refer to?

\*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

📌 Check all that apply

Please choose **all** that apply:

- Clinical Laboratory Improvement Amendments (CLIA)
- National guidelines and protocols
- Don't know

#### 5. How long does it usually take to get the genetic testing result? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

📌 Choose one of the following answers

Please choose **only one** of the following:

- < 2 weeks
- 2 - 4 weeks
- Months
- Don't know

#### How many months? \*

Only answer this question if the following conditions are met:

Answer was 'Months' at question '54 [AH]' (5. How long does it usually take to get the genetic testing result?)

📌 Only numbers may be entered in this field.

Please write your answer here:

## 6. Do you offer genetic counselling? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Yes
- No
- Don't know

## 6a. Who provides genetic counselling to your patients? \*

Only answer this question if the following conditions are met:

Answer was 'No' at question '56 [AI]' (6. Do you offer genetic counselling?)

❗ Check all that apply

Please choose **all** that apply:

- Centre based genetic counsellor
- External genetic counsellor
- Centre based ophthalmogenetic counsellor
- External ophthalmogenetic counsellor

Other:

## 7. What estimated percentage of IRD patients has been genetically tested at your centre? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🗳️ Choose one of the following answers

Please choose **only one** of the following:

- 0 - 20%
- 21 - 40%
- 41 - 60%
- 61 - 80%
- 81 - 100%
- Don't know

## 8. What estimated percentage of your patients has been genetically solved? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🗳️ Choose one of the following answers

Please choose **only one** of the following:

- 0 - 20%
- 21 - 40%
- 41 - 60%
- 61 - 80%
- 81 - 100%
- Don't know

## 9. What is the percentage of patients tested on clinical grade and research grade (accredited / CLIA) tests?

\*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

❗ Only numbers may be entered in these fields.

❗ The sum must be between 0 and 100

Please write your answer(s) here:

Clinical grade only

Research grade only

Research grade + Clinical grade

Clinical grade negative + Research grade

## 10. Who covers the costs of genetic testing in your IRD patients? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

❗ Check all that apply

Please choose **all** that apply:

- Covered by public health service
- Covered by private health insurance
- Covered by centre budget
- Research funding only
- No coverage available

## 11. At which depth is genetic testing performed? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🗨 Check all that apply

Please choose **all** that apply:

- Whole Genome Sequencing (WGS)
- Whole Exom Sequencing (WES)
- Medical Exom Sequencing (MES)
- IRD-specific gene panel
- Diagnosis-directed Sanger sequencing
- Don't know

## Please specify the number of genes in each situation: \*

Only answer this question if the following conditions are met:

Answer was 'IRD-specific gene panel' at question '62 [AQ]' (11. At which depth is genetic testing performed?)

❗ Comment only when you choose an answer.

Please choose all that apply and provide a comment:

General panel

LCA panel

RP panel

CRD panel

Optic atrophy panel

Other:

## Involvement in Clinical Trials

### Section 4

## 1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases? \*

Only answer this question if the following conditions are met:

Answer was 'Manage IRD patients at our centre' at question '7 [G]' (2. How do you manage IRD patients?) *and* Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

🗳️ Choose one of the following answers

Please choose **only one** of the following:

- currently involved
- previously involved
- not involved
- interested in being involved

## Involvement in Clinical Trials

1a. Please answer the following parameters:

### Please fill in the following fields regarding a clinical study with gene therapies for IRD. \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' *or* 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

## RD addressed: \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' or 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

🚫 Check all that apply

Please choose **all** that apply:

LHON

LCA

EOSRD

Choroideremia

Retinitis pigmentosa

Cone Rod Dystrophy

Achromatopsia

Stargardt disease

Usher Syndrome

Other:

## Gene addressed: \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' or 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

🚫 Check all that apply

Please choose **all** that apply:

- ABCA4
- CHM
- CEP290
- CNGA3
- CNGB3
- MYO7A
- RHO
- RPGR
- RPE65
- gene independent

Other:

## Did your Centre have the leading PI? \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' or 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

Please choose **only one** of the following:

- Yes
- No

## From which centre was the leading PI? \*

Only answer this question if the following conditions are met:

Answer was 'No' at question '68 [A5]' (Did your Centre have the leading PI?)

Please write your answer here:

## Did your centre enroll patients? \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' or 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

Please choose **only one** of the following:

Yes

No

## Do you need to add an additional clinical trial? \*

Only answer this question if the following conditions are met:

Answer was 'currently involved ' or 'previously involved ' at question '64 [AR]' (1. Are you currently or have you previously been involved in a clinical study with gene therapies for inherited retinal diseases?)

Please choose **only one** of the following:

Yes

No

## Please fill in the following fields regarding a clinical study with gene therapies for IRD. \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '71 [A8]' (Do you need to add an additional clinical trial?)

## RD addressed: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '71 [A8]' (Do you need to add an additional clinical trial?)

🚫 Check all that apply

Please choose **all** that apply:

LHON

LCA

EOSRD

Choroideremia

Retinitis pigmentosa

Cone Rod Dystrophy

Achromatopsia

Stargardt disease

Usher Syndrome

Other:

## Gene addressed: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '71 [A8]' (Do you need to add an additional clinical trial?)

🚫 Check all that apply

Please choose **all** that apply:

ABCA4

CHM

CEP290

CNGA3

CNGB3

MYO7A

RHO

RPGR

RPE65

gene independent

Other:

### Did your Centre have the leading PI? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '71 [A8]' (Do you need to add an additional clinical trial?)

Please choose **only one** of the following:

- Yes
- No

### From which centre was the leading PI? \*

Only answer this question if the following conditions are met:

Answer was 'No' at question '75 [A12]' (Did your Centre have the leading PI?)

Please write your answer here:

### Did your centre enroll patients? \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '71 [A8]' (Do you need to add an additional clinical trial?)

Please choose **only one** of the following:

- Yes
- No

## RPE65 related IRDs

### Section 5

# 1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Manage IRD patients at our centre' at question '7 [G]' (2. How do you manage IRD patients?) *and* Answer was 'Yes' at question '46 [AC]' (1. Are IRD patients from your centre tested genetically?)

Please choose **only one** of the following:

- Yes
- No

## 1a. Please provide an estimate of the number of IRD patients with confirmed homozygous mutations in RPE65 that have been identified in your centre. \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

**i** Choose one of the following answers

Please choose **only one** of the following:

- 0
- 1 - 5
- 6 - 10
- 11 - 20
- > 20
- Don't know

**1b. Please provide an estimate of the number of IRD patients with confirmed compound heterozygous mutations in RPE65 that have been identified in your centre. \***

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

**!** Choose one of the following answers

Please choose **only one** of the following:

- 0
- 1 - 5
- 6 - 10
- 11 - 20
- > 20
- Don't know

**2. Please provide an estimate of the number of IRD patients with confirmed bi-allelic mutations according to ACMG class 1 or 2 that have been identified in your centre. \*<sup>1</sup>**

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

**!** Choose one of the following answers

Please choose **only one** of the following:

- 0
- 1-5
- 6-10
- 11-20
- > 20
- Don't know

<sup>1</sup> In fact, according to ACMG, class 1 and 2 should read class 4 and 5. The confusion comes from the text in Richards et al 2015 where the order of mutation by severity is (1) pathogenic, (2) likely pathogenic, (3) VUS, variants of unclear significance, (4) likely benign, (5) benign. In the context the question was asked it was clear that pathogenic and likely pathogenic mutations were meant.

### 3. Please provide an estimate of the number of IRD patients with confirmed bi-allelic mutations in RPE65 that are currently observed in your centre. \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- 0
- 1-5
- 6-10
- 11-20
- > 20
- Don't know

### 4. How many different RPE65 mutations have been identified in your IRD patients?

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

Please write your answer here:

## 5. How many of the identified RPE65 were already reported?

Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Only numbers may be entered in these fields.

❗ The sum must be between 0 and 100

Please write your answer(s) here:

Already reported

Novel

## 6. Please indicate the referral diagnosis of the RPE65-carrying patients your centre identified.

Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Only numbers may be entered in these fields.

❗ The sum must be between 0 and 100

Please write your answer(s) here:

Early-onset severe retinal dystrophy (EOSRD)

LCA

Retinitis pigmentosa, Rod-one Dystrophy

Unclassified visual impairment

## 7. In what percentage of IRD patients did your centre change the referral diagnosis?

Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Only numbers may be entered in this field.

Please write your answer here:

%

## 8. Does your centre follow a specific referral process for RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Yes
- No
- Don't know

## 9. How frequently does your centre recall the patients for follow up? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Every Six months
- Annually
- Biennially
- Longer
- Don't know
- Other

## 10. Does another institution follow these patients in addition? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Yes
- No
- Don't know

## 11. How frequently did you see your patients in the past? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Every six months
- Annually
- Biennially
- Longer
- Don't know

Other

## 12. Which visual field tests does your centre perform in RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

🚫 Check all that apply

Please choose **all** that apply:

- Static
- Kinetic perimetry
- Fundus-controlled perimetry

## Please specify the device: \*

Only answer this question if the following conditions are met:

Answer was 'Static' at question '91 [B12]' (12. Which visual field tests does your centre perform in RPE65 patients?)

🚫 Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Humphrey

Octopus - Device:

Twinfield

Other:

## Please specify the device used in kinetic perymetry. \*

Only answer this question if the following conditions are met:

Answer was 'Kinetic perimetry' at question '91 [B12]' (12. Which visual field tests does your centre perform in RPE65 patients?)

❗ Comment only when you choose an answer.

Please choose all that apply and provide a comment:

Goldmann (manual)

Octopus - Device:

Other:

## Please specify the device used for Fundus-controlled perimetry. \*

Only answer this question if the following conditions are met:

Answer was 'Fundus-controlled perimetry' at question '91 [B12]' (12. Which visual field tests does your centre perform in RPE65 patients?)

❗ Check all that apply

Please choose **all** that apply:

MP1, Nidek

MP3, Nidek

Compass, Centrevue MAIA

Other:

### 13. How many visual field tests (static, device) did your centre perform per each RPE65 patient?

Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

🗳️ Choose one of the following answers

Please choose **only one** of the following:

- < 5
- 5 - 10
- 11 - 20
- > 20
- Don't know

### 14. How many visual field tests (kinetic) did your centre perform for each RPE65 patient?

No. of tests / No. of patients. Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

🗳️ Choose one of the following answers

Please choose **only one** of the following:

- < 5
- 5 - 10
- 11 - 20
- > 20
- Don't know

## 15. Do you perform 2 ColorThreshold perimetry in your RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

Please choose **only one** of the following:

Yes

No

## Please specify the device: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '97 [B15]' (15. Do you perform 2 ColorThreshold perimetry in your RPE65 patients?)

Please write your answer here:

## 16. Do you perform Full-Field Light Sensitivity Threshold Testing (FST) in your RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

Please choose **only one** of the following:

Yes

No

### Please specify: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '99 [B16]' (16. Do you perform Full-Field Light Sensitivity Threshold Testing (FST) in your RPE65 patients?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- red, blue, white testing
- white testing only

### Please specify the device: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '99 [B16]' (16. Do you perform Full-Field Light Sensitivity Threshold Testing (FST) in your RPE65 patients?)

❗ Check all that apply

Please choose **all** that apply:

- Espion , Diagnosis LLC
- Roland Consult

Other:

### 17. Do you perform Pupillometry in your RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

Please choose **only one** of the following:

- Yes
- No

### Please specify: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '102 [B17]' (17. Do you perform Pupillometry in your RPE65 patients?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Chromatic pupillometry
- White only pupillometry

### 18. Do you perform mobility parcours in your RPE65 patients?

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

Please choose **only one** of the following:

- Yes
- No

### Please specify: \*

Only answer this question if the following conditions are met:

Answer was 'Yes' at question '104 [B18]' (18. Do you perform mobility parcours in your RPE65 patients?)

❗ Choose one of the following answers

Please choose **only one** of the following:

- Spark parcours
- ORA Inc. parkour

Other

**19. How frequently do you perform fundus imaging in your RPE65 patients?  
Imaging / visit. Please provide an estimate.**

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

- Monthly
- Quarterly
- Twice a year
- Annually
- Biennially
- Longer
- Other

20. How frequently do you perform fundus autofluorescence recording in your RPE65 patients?  
Imaging / visit. Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

- Monthly
- Quarterly
- Twice a year
- Annually
- Biennially
- Longer
- Other

21. How frequently do you perform SD-OCT recording in your RPE65 patients?  
Imaging / visit. Please provide an estimate.

\*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

- Monthly
- Quarterly
- Twice a year
- Annually
- Biennially
- Longer
- Other

**22. How frequently do you perform visual acuity testing in your RPE65 patients?  
Imaging / visit. Please provide an estimate. \***

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

❗ Choose one of the following answers

❗ If you choose 'Other:' please also specify your choice in the accompanying text field.

Please choose **only one** of the following:

- Monthly
- Quarterly
- Twice a year
- Annually
- Biennially
- Longer
- Other

## 23. Which methods do you apply for visual acuity testing in your RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

🗨️ Check all that apply

Please choose **all** that apply:

- ETDRS charts
- Snellen charts
- Number charts
- Landolt rings
- Tumbling 'E' charts
- Berkeley Rudimentary Vision Test (BRVT)
- Teller acuity cards
- Lea Symbols®

Other:

## 24. Which methods do you apply for colour vision testing in your RPE65 patients? \*

Only answer this question if the following conditions are met:

Answer was NOT 'None' at question '6 [F]' (1. How many IRD patients are currently managed by your centre? ) *and* Answer was 'Yes' at question '78 [B1]' (1. Do you have IRD patients with confirmed bi-allelic mutations in RPE65 that have been identified in your centre?)

🗨️ Check all that apply

Please choose **all** that apply:

- Farnsworth Panel D15
- Lanthony Panel D15
- Ishihara cards
- Anomaloscope

Other:

## Please specify the Anomaloscope used: \*

Only answer this question if the following conditions are met:

Answer was 'Anomaloscope' at question '111 [B24]' (24. Which methods do you apply for colour vision testing in your RPE65 patients?)

🗨 Check all that apply

Please choose **all** that apply:

OCULUS

Nagel

Other:

**Thank you for taking the time to complete the IRD Survey Questionnaire.**

Submit your survey.

Thank you for completing this survey.