

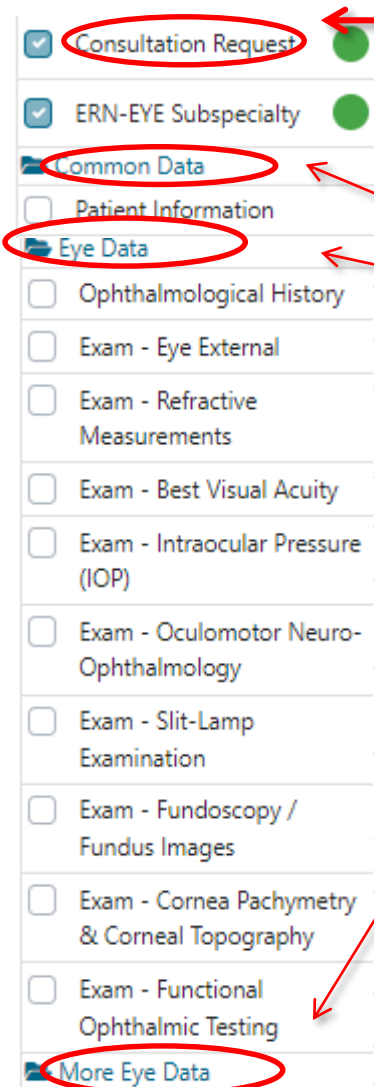


# Organisation of the data in the dataset

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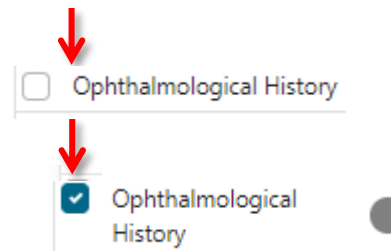
## Organisation of the data in the dataset



- The dataset is divided into 4 main sections:
  1. Consultation Request
  2. Common Data
  3. Eye Data
  4. More Eye Data

- These sections are divided into many subsections.

- To fill in the desired sections, click on the checkbox in front of the item.





# 1. Fill in the «Consultation request»: it is mandatory for all panels.

ERN-EYE CONSULTATION FORM  
✓ Consultation Request

It comprises the urgency of the panel, the reason for the consultation, the name of the leader and thematic area. All fields with an \* are mandatory.

- Consultation Request
- ERN-EYE Subspecialty
- Common Data
  - Patient Information
- Episode Description
  - Rare Disease Diagnosis
  - Comorbidities
  - Phenotype/Genetic Features and Biobanks
  - Family History
  - Health Behaviours
  - Allergies and Other Adverse Reactions
  - History of Past Illness and Disorders
  - Special Treatment Intervention
  - Surgical Procedures
  - Transplantation History
  - Medication Summary
  - Others

### CONSULTATION REQUEST

Do not use any nicknames liable to identify the patient, including his/her first name or last name.

\* Nickname:

Is urgency required for this panel?

\* Is this a repeat of a previous panel for the same patient but a different episode?  Yes  No  Don't know

\* Consultation request description:

\* Consultation request reason:  Diagnosis  Treatment  Other

\* Healthcare provider:

\* Point of care specialist:

\* ERN: ERN-EYE

\* Panel lead:

Panel Manager:

\* Primary thematic area:  Anterior Segment Rare Eye Diseases  Neuro Ophthalmology rare diseases  Pediatric Ophthalmology Rare Diseases  Retina Rare Eye Diseases

Secondary thematic areas:  Anterior Segment Rare Eye Diseases  Neuro Ophthalmology rare diseases  Retina Rare Eye Diseases





## 2. Fill in the sections and subsections you want.

E. g. Functional Ophthalmic Testing

Enter psycho-physical testing results (visual fields, microperimetry, colour vision tests) and also insert the results of objective tests (electrophysiology: VEP, ERG and EOG).

### Eye Data

- Ophthalmological History
- Exam - Eye External
- Exam - Refractive Measurements
- Exam - Best Visual Acuity
- Exam - Intraocular Pressure (IOP)
- Exam - Oculomotor Neuro-Ophthalmology
- Exam - Slit-Lamp Examination
- Exam - Fundoscopy / Fundus Images
- Exam - Cornea Pachymetry & Corneal Topography
- Exam - Functional Ophthalmic Testing

### Psycho-physical testing

Date:

- Visual fields performed:
- Kinetic visual fields, manual Goldmann (Classic) or automated Goldmann (Octopus)
  - Static visual fields, Humphrey or Octopus
  - Microperimetry, Centervue Maia or Nidek
  - Colour Vision
  - Other

### Objective tests

#### Electrophysiology

- Tests performed:
- Visual evoked potentials, Flash (VEP)
  - Pattern VEP
  - Ganzfeld-ERG (ffERG)
  - Multifocal ERG (mfERG)
  - Electro-oculogram (EOG)

ISCEV standards:  Yes  No



## Some remarkable sections



### ➤ Rare disease diagnosis

- ERN-EYE Subspecialty
- Common Data
- Patient Information
- Episode Description
- Rare Disease Diagnosis
- Comorbidities
- Phenotype/Genetic Features and Biobanks
- Family History
- Health Behaviours
- Allergies and Other Adverse Reactions
- History of Past Illness and Disorders
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- Medication Summary
- Others

**RARE DISEASE DIAGNOSIS**

Current Diagnosis

Search All Coding Systems:

Diagnosis Coding System:  ICD-10  ICC3-3  Orphanet  Other

Date of first symptoms onset:

Date of current Diagnosis:

Status of current Diagnosis:  Active  In Remission  Resolved  Relapse  Other

\* Diagnosis Provider:  ERN member  Affiliated ERN Member  Non-ERN member

\* Treatment Healthcare Provider:  ERN member  Affiliated ERN Member  Non-ERN member

Diagnostic Test:  LOINC  Text

Date of observation:

Result:

Working Diagnosis

Diagnosis coding systems available: ICD-10, ICC3-3, Orphanet or free text.  
Additionally, many items use **HPO terms** for description.



## Some remarkable sections



### ➤ Phenotypes/Genetic Features and Biobanks

Click on **+ Add genetic/phenotype/biobank** to add genetic information according to HGNC or free text.

The screenshot displays the ERN-EYE interface for adding genetic information. On the left, a sidebar lists various data categories, with 'Phenotype/Genetic Features and Biobanks' selected. The main content area is titled 'PHENOTYPE/GENETIC FEATURES AND BIOBANKS' and contains a form with the following fields:

- Genetic study performed:  LOINC  Text
- Gene Affected:  HGNC Symbols  Text
- DNA Variant: Please select one
- DNA Nucleotide reference sequence (RefSeq Id/LRG Id): Type value for auto-complete ...
- DNA Nucleotide variant description: e.g. g.123A>G
- RNA Variant: Please select one
- RNA Nucleotide reference sequence (LRG Id): Type value for auto-complete ...
- RNA Nucleotide variant description: e.g. g.123A>G
- Protein Variant: Please select one
- Protein sequence (RefSeq Id/LRG Id): Type value for auto-complete ...
- Protein variant description:
- Phenotypic Abnormality (HPO): Type value for auto-complete ...
- Other changes: e.g. DNA methylation
- Allelic State: Please select one
- Somatic Mutation as included in COSMIC:

A red arrow points to the '+ Add genetic/phenotype/biobank' button in the top right corner of the form.



## Some remarkable sections


### ➤ Family history

Draw or attach a pedigree.

- Consultation Request
- ERN-EYE Subspecialty
- Common Data**
- Patient Information
- Episode Description
- Rare Disease Diagnosis
- Comorbidities
- Phenotype/Genetic Features and Biobanks
- Family History

PEDIGREE

GrandMother  
 Father



proband  
Pheno: HP\_0040279

Edit Person
Relationships
Add Child
Add Parent
Add Partner
Add Sibling
Delete Person

Use this section to upload and display Pedigree images into CPMS. Please note that only permitted formats can be displayed (i.e. PNG, and JPEG)

### ➤ Imaging test and Medical documents

Drag and drop the files directly to the CPMS to upload files (.jpeg, .pdf or DICOM). Note that you can upload files in many sections in the CPMS.

- Systemic Testing
- More Eye Data**
  - Imaging Tests
  - Histology/Pathology
  - Ophthalmological Therapies
  - Rehabilitation Low Vision Aid
  - Participation in clinical trials
  - Additional Medical History
  - Medical Documents

Image:



Attach Image or File

Upload type:  File(s)  Dicom Study (.dcm)

Drop Documents, Images, Videos, CCDA documents here or click to upload.  
Upload one file at a time.



# Image viewer tool



- CPMS has an integrated image viewer tool (supported imaging techniques: CT scans MRIs, PET scans, Tomography, Echographs, RXs, Dicomized Static Images, Endoscopies).
- Edit the images (**but not during a meeting**), e.g.: change the viewport layout, invert colour, zoom,, rotate image to make use of this tool.

Attachment Uploaded by: eyehpuser1 testuser Date: 01/Mar/2022 Size: 4.7 MB

The screenshot shows the medDream WEB DICOM viewer interface. The top toolbar contains various tools: Window, Pan, Zoom, Measure, Rotate, Magnify, Scroll, Hangup, Fusion, Cine, Channel, Layout, Thumbnail, Forward, Export, Print, FullScreen, Reset, MPR, DICOM, Theme, Key, and Share. The main viewing area is divided into several panes. The left pane shows a patient list with 'Anon' selected and a grid of image thumbnails. The main area displays several MRI slices in different orientations (RPI, ASR, LAS, PIL) with technical details like '256 x 256', 'Z: 1.60', and 'W: 490 L:282'.