# DMLA 2004 - Hereditary Retinal Dystrophy 2004

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## General

### Identification

| | |
|---|---|
| Detailed name | Hereditary Retinal Dystrophy 2004 |
| Sign or acronym | DMLA 2004 |
| CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation | CNIL |

### General Aspects

| | |
|---|---|
| Medical area | Ophthalmology<br>Radiology and medical imaging |
| Health determinants | Genetic |
| Keywords | visual function, Health episodes, morphometric data, retina, impact, quality of life, assessment, disability |

### Scientific investigator(s) (Contact)

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| Organization | INSERM |

## Collaborations

## Funding

| | |
|---|---|
| Funding status | Mixed |
| Details | ANR, Inserm, CHNO 15-20, Communauté Européenne, Ministère de la Recherche et de l'Enseignement Supérieur, Ministère de la Santé, Fondation FIGHTING BLINDNESS (USA), FONDATION VOIRE ET ENTENDRE, FONDATION POUR LA RECHERCHE MEDICALE |

## Governance of the database

| | |
|---|---|
| Sponsor(s) or organisation(s) responsible | CHNO DES QUINZE-VINGTS |
| Organisation status | Public |

## Additional contact

# Main features

## Type of database

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|---|---|
| Type of database | Study databases |
| Study databases (details) | Case control study |
| Database recruitment is carried out by an intermediary | A selection of health institutions and services |
| Database recruitment is carried out as part of an interventional study | No |
| Additional information regarding sample selection. | Prospective Other bodies active in creating this cohort: CHU, CHG |

## Database objective

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|---|---|
| Main objective | General objective: to study genotype-phenotype |

| | |
|---|---|
| | correlations Secondary objectives: - to study morphofunctional correlations - to identify new functional or morphological markers - to research predictive signs of progression. |
| Inclusion criteria | Individuals with hereditary retinal dystrophy Related individuals |
| **Population type** | |
| Age | Adulthood (19 to 24 years)<br>Adulthood (25 to 44 years)<br>Adulthood (45 to 64 years)<br>Elderly (65 to 79 years)<br>Great age (80 years and more) |
| Population covered | Sick population |
| Gender | Male<br>Woman |
| Geography area | National |
| Detail of the geography area | Multicentric cohort throughout France (13 centres) |
| **Data collection** | |
| **Dates** | |
| Date of first collection (YYYY or MM/YYYY) | 10/2004 |
| **Size of the database** | |
| Size of the database (number of individuals) | [1000-10 000[ individuals |
| Details of the number of individuals | 1844 |
| **Data** | |
| Database activity | Data collection completed |
| Type of data collected | Clinical data<br>Declarative data<br>Paraclinical data<br>Biological data |
| Clinical data (detail) | Direct physical measures<br>Medical registration |

| | |
|---|---|
| Declarative data (detail) | Paper self-questionnaire<br>Face to face interview |
| Paraclinical data (detail) | Imaging, visual acuity, visual field assessment, colour vision examination, electroretinography |
| Biological data (detail) | Type of samples taken: Blood |
| Presence of a biobank | Yes |
| Contents of biobank | DNA |
| Details of biobank content | DNA bank |
| Health parameters studied | Quality of life/health perception |
| **Procedures** | |
| Data collection method | Self-administered questionnaire: manual input Interview: manual input Biological analysis: manual input |
| Participant monitoring | Yes |
| Details on monitoring of participants | (Indefinite duration) |
| Links to administrative sources | No |
| **Promotion and access** | |
| **Promotion** | |
| Link to the document | http://bjo.bmj.com/content/82/9/996.long |
| **Access** | |
| Terms of data access (charter for data provision, format of data, availability delay) | Data may be used by academic teams |
| Access to aggregated data | Access on specific project only |
| Access to individual data | Access on specific project only |