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IRD. Inherited Retinal Dystrophy

NIHR BioResource – Rare Diseases study project

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V2 16/10/2019

Summary

Inherited retinal dystrophy (IRD) is the most common cause of blindness in the UK working population. Disability is life-long with a considerable burden to family and social support. The disorders are monogenic with considerable genetic and allelic heterogeneity. The accessibility of the retina to imaging, psychophysics and electrophysiology, allows an accurate clinical diagnosis in most affected persons, and has led to the discovery of a high proportion of causative genes compared to other systems.

The accessibility of the eye and retina, its immune-privilege status and ease of measurement, has led to IRD being a paradigm for precision medicine for gene-replacement and gene-directed pharmaceutical treatments, with an increasing number of active trials worldwide. Importantly therefore the BioResource aims to collect cohorts of patients with specific sub-types of IRD for recall to natural history studies, gene and drug trials, and cellular investigations.

Secondly, within each subtype of disorder there is often great variability of age-related severity, the cause of which is not known. Collecting cohorts of well phenotyped patients will allow the allelic and trans-acting genetic factors to be investigated, improving our understanding of the biological pathways underlying vision loss and providing opportunities for novel therapies.

Recruitment Criteria

At present at least, the BioResource does not aim to determine the molecular diagnosis in this group of patients (this is best served through NHS England Genetic services), and so recruitment is focused on patients and families who already have a genetic diagnosis.

Inclusion

- Patients with a clinical diagnosis of an IRD including those affecting rod, cone photoreceptors, RPE and inner retina, that are congenital or later onset.

We encourage the recruitment of multiple affected members of a single family.
We encourage the recruitment of syndromic disorders such as Usher and Bardet-Biedl syndromes.

Examples of sub-types of IRD include: retinitis pigmentosa, cone-rod dystrophy, macular dystrophy (including stargardt disease) and stationary disorders such as achromatopsia, CSNB and X-linked retinoschisis.

Exclusion

Those unlikely to have monogenic disease - Unilateral disorders, those with evidence of previous or present inflammation, retinal vascular disorders, retinal detachment, age-related macular disease.