Envision a Brighter Future for Eye Diseases

The **EYE-RD Global Registry** is looking to collect more information about the inherited retinal diseases (IRDs) X-linked retinitis pigmentosa (XLRP) and achromatopsia (ACHM), to help advance research and treatment options.

Consider encouraging your patients to join the **EYE-RD Global Registry**, a community dedicated to advancing research and treatment options for IRDs. IRDs can be different for future generations, and it starts today.

About Inherited Retinal Diseases

IRDs are a group of diseases, including XLRP and ACHM, that can cause severe vision loss or blindness.[1,2]

XLRP, a severe and aggressive form of retinitis pigmentosa, is a degenerative retinal disease that affects rod and cone photoreceptors.[3] XLRP is characterized by early onset and rapid symptom progression, with night blindness and progressive visual field constriction leading to legal blindness by a median age of 45 years.[3,4] Approximately 70%-90% of XLRP cases involve *RPGR* mutations[5]; the majority of the clinical effects of *RPGR*-associated XLRP are due to variants in the ORF15 isoform, which is a challenging region to sequence.[6]

ACHM is a rare autosomal recessive cone disease characterized by severe visual impairment, including poor visual acuity as well as disabling sensitivity to light (photoaversion), which results in decreased visual function and avoidance of photopic conditions.[7-10] Occurring in approximately 1:30,000 live births worldwide, ACHM manifests at birth or early infancy.[7,8,11] Most cases of ACHM are complete, associated with a lack of cone function, and variants in the *CNGA3* and *CNGB3* genes represent approximately 80% of complete ACHM cases.[9,12,13]

Registry Objectives

The aim of the prospective, longitudinal, noninterventional **EYE-RD Global Registry** of patients genetically diagnosed with XLRP or ACHM is to describe the clinical course of disease through timely collection of relevant data from real-world clinical practice. These data will be used to address the research needs of the IRD community, providing insights into patient management and informing potential future treatments.[14]

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EYE-RD GLOBAL REGISTRY

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Registry Details

The **EYE-RD Global Registry** will last 8+ years. No visits, tests, or procedures beyond your patients' standard care are required. For this registry, patient health information from their regularly scheduled eyecare provider appointments will be collected for research purposes. Patients will also be asked questions to determine how IRD impacts their lives.[14]

Why
Ask Your Patients to Participate?

Data collected as part of this registry may help to progress medical research by better defining the natural history of disease progression in XLRP and ACHM, evaluating genotype-phenotype associations, and understanding the patient pathway prior to and after diagnosis. These data will be used to support the development of novel and potentially transformative treatments.

To learn more about whether your patients can take part in this registry, please contact **Info_EyeRDRegistry@its.jnj.com.**

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