MEDXCHANGE LIVE™ SPECIAL EDITION

ADVANCES IN
Inherited Retinal Diseases

Unmet Needs and Future Perspectives in Inherited Retinal Diseases

Vision loss ranks highest in the general population's fear of all possible disabilities, with inherited retinal diseases (IRDs) being significantly disabling conditions mostly affecting working-age people.[1,2] As a clinically and genetically heterogeneous group of neurodegenerative disorders, IRDs lead to severe visual impairment and, in some individuals, complete blindness.[2] With an estimated global prevalence of approximately 1 in 2,000, IRDs effect more than 2 million people worldwide and remain a leading cause of blindness.[2,3] Pathophysiological understanding and treatment options are limited in IRDs, with research and medical progress needed to solve the unmet needs.[4] The irreversible progress of vision loss, the underuse of genetic testing, and lack of effective treatments highlight the unmet needs for patients with IRDs (**Figure**).[5]

Genetic testing and counseling are important components of IRD assessment and are valuable tools for diagnosis, in providing information to patients and family, and to assess potential eligibility for clinical trial participation.[6] Although it is understood that genetic testing is an important tool for patients with IRD, additional research is necessary in the field of genetic testing and counseling.[4] Currently, genetic sequencing via panel diagnostics can identify the genetic cause of the disorder in approximately two thirds of patients.[7] Although over 270 disease genes have been identified,[8] there is an unmet medical need to improve the sensitivity of genetic testing in order to increase the identification of pathogenic mutations in more cases, to incorporate genetic testing into research and diagnostic testing, to increase the ability to determine which rare variants are disease-causing, to identify genetic modifiers of disease severity, and to improve access to molecular genetic diagnostic testing, test result evaluation, and genetic counseling, including increased coverage of testing costs.[4,6] In addition, disease models are needed to increase the limited understanding of pathophysiology and mechanisms of IRDs, to identify novel treatment targets, and to provide proof of concept for potential therapeutic strategies.[6]

Unmet Needs in Inherited Retinal Disease[4]

- Improved understanding of the mechanisms of IRDs
- Improved sensitivity of testing
- Identification of novel therapy targets
- New therapeutic approaches
- Cell-specific targeting
- Improved patient access to genetic testing and counseling

IRDs are largely incurable and, left untreated, can result in severe visual impairment or blindness, but the treatment landscape is rapidly changing.[9] Currently, therapeutic options are limited, but new treatments, including gene therapy, stem cell therapy, retinal prostheses, and direct brain stimulation are being tested.[2,4] The recent FDA approval of gene augmentation therapy for RPE65-associated IRD using adeno-associated viruses is an important advancement that suggests that similar treatment approaches may be used for additional IRDs.[10] Despite such advancements, cell-specific targeting and additional treatments for IRDs remain an unmet need.[4] Although there are unmet medical needs regarding IRD treatments and the disease burden associated with IRDs, the recent advancements in retinal genetics, imaging, and molecular biology, have led to improved diagnosis and the development of and clinical trials for novel therapeutics.[11-16]

Most Common Inherited Retinal Diseases

RETINITIS PIGMENTOSA[20–22]	• Most frequent IRD • Progressive hereditary retinal dystrophy in which degeneration of retinal photoreceptors causes nyctalopia and progressive visual field defects • Caused by variations in 60 genes that affect the retina; depending on gene affected • Can appear during childhood or adulthood • X-linked retinitis pigmentosa represent some of the most severe forms of retinitis pigmentosa, resulting in early onset in childhood and rapid progression to blindness by the time patients reach 20 to 30 years old
CHORIORETINAL DSYTROPHY[23–28]	• Characterized by a diffuse, progressive atrophy of the choroid, retinal pigment epithelium, and retina • Structural alterations of the central retina in the early stage of the disease • Exact pathogenesis not completely understood • Remarkable for chronic progressive visual loss, including early onset night blindness and peripheral visual field reduction, with patients usually retaining good central visual acuity into their 50s
STARGARDT MACULAR DYSTROPHY[3,29–31]	• Associated with progressive central vision loss • Genetically and clinically heterogeneous disease • Leading cause of adolescent/young adult vision impairment
ACHROMATOPSIA[32,33]	• Rare inherited autosomal recessive disease • Caused by mutations in either CNGB3 or CNGA3 • Prevents cone photoreceptors from functioning • Patients legally blind from birth and usually suffer from severely reduced visual acuity of 20/200 or worse; a disabling sensitivity to light, or photophobia; total color blindness; and involuntary back and forth eye movements, or nystagmus
AGE-RELATED MACULAR DEGENERATION (AMD); NEOVASCULAR AMD, AND GEOGRAPHIC ATROPHY[5,13,34]	• Most common cause of blindness in developed countries, accounting for 8·7% of all blindness worldwide • Patients often have low levels of CD59, a protein that protects the retina from damage caused by an essential part of the body's natural immune response called complement • Sight loss from advanced AMD occurs either as neovascular (or “wet”) AMD or geographic atrophy • Geographic atrophy, which affects around 5 million people globally, and is one of the leading causes of blindness in people over age 50; 1 in 4 people over 90 are affected
LEBER CONGENITAL AMAUROSIS[35–38]	• Primarily affects the retina • People typically have severe visual impairment beginning in infancy • Considered one of the most severe Inherited Retinal Diseases • One of the most common IRDs (>5% of all IRDs) • A monogenic autosomal recessive disease that affects ~1:30,000 newborns
PROGRESSIVE CONE AND CONE-ROD DYSTROPHIES[7,39,40]	• Clinically and genetically heterogeneous group of IRDs, with at least 30 genes implicated • Characterized by cone photoreceptor degeneration, which may be followed by subsequent rod photoreceptor loss • People typically present with progressive loss of central vision, color vision disturbance and photophobia • Estimated incidence ranges from 1:20,000–100,000

THEIR GENES GAVE THEM BUTTON NOSES, BROWN HAIR, AND VISION LOSS

To date, science has discovered more than 270 genes related to **inherited retinal diseases**.[1] With the evolution of genetic testing comes the ability to more precisely diagnose your patients.

More answers may uncover more possibilities for active clinical trials, emerging treatments, and even identifying underlying conditions beyond vision issues.

MOVE FORWARD WITH MORE ANSWERS

Scan to find more answers about genetic testing and retesting at **EyesOnGenes.com**.

Reference: 1. Branham K, Schlegel D, Fahim AT, Jayasundera KT. Genetic testing for inherited retinal degenerations: triumphs and tribulations. *Am J Med Genet C Semin Med Genet.* 2020;184(3):571-577.

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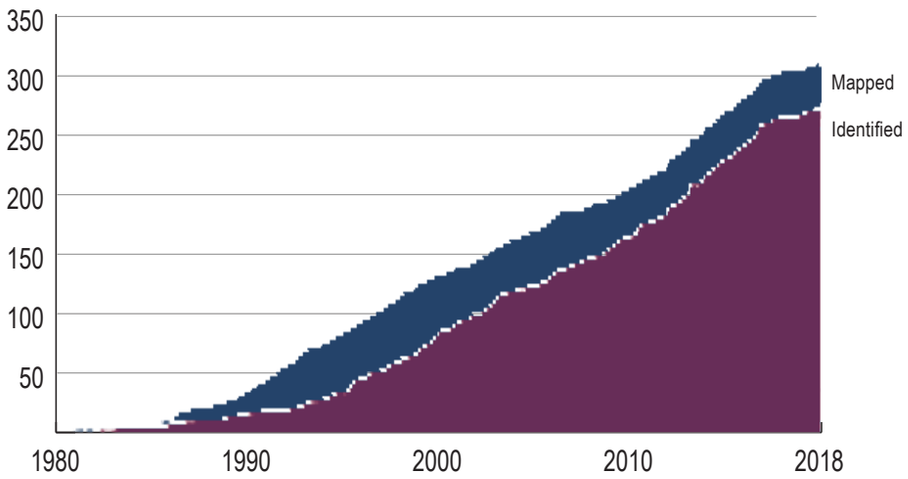
History of the Diagnosis of Inherited Retinal Diseases

In the late 1980s and early 1990s, when the first IRD genes were discovered, ophthalmic genetics was largely a descriptive subspecialty with the primary goals of the ophthalmologist being to name the patient's condition and discern the inheritance pattern.[17-20] At that time, the chance that a molecular diagnosis could be accomplished for the average patient with an IRD was less than 5%, as genetic tests were only performed by a few research laboratories.[20] The main limitations to molecular diagnosis were the overall lack of knowledge regarding the human genome and the relatively rudimentary and painstaking methods for testing.[20] Molecular tests in were focused by the clinical features of the family being studied.[20] Many things have changed in ophthalmic genetics in the past 25 years, including the more widespread availability of preimplantation genetic testing to reduce the recurrence of severe genetic diseases and the introduction of CRISPR-based genome editing.[21-28] Since the identification of the first gene responsible for an IRD back in 1988, huge progress has been made in the field of molecular testing, leading to the identification of over 270 disease-causing genes (**Figure**).[8,29]

Number of Mapped and Identified Retinal Disease Genes from 1980–2018[8]



Adapted from: https://sph.uth.edu/retnet/sum-dis.htm.

The Importance of Genetic Testing[30]

Identifying the genetic cause of disease is an important part of care for patients with IRDs. Results from genetic testing provide an accurate diagnosis that can assist in identifying optimal treatment for patients with IRDs. In addition, a diagnosis from genetic testing can inform patients about the potential disease risk to family members as well as to other organs in the patient's body that may be affected. In the case of infants and young children, genetic testing can assist in identifying those children at risk of other health problems as well as those who may benefit from therapy. Along with a physician, a genetic counselor can discuss genetic testing results with patients in order to guide the patient and the patient's family through the impact of an IRD diagnosis on them, their family, and future family planning decisions.

Advancement in Therapeutic Strategies for Inherited Retinal Diseases

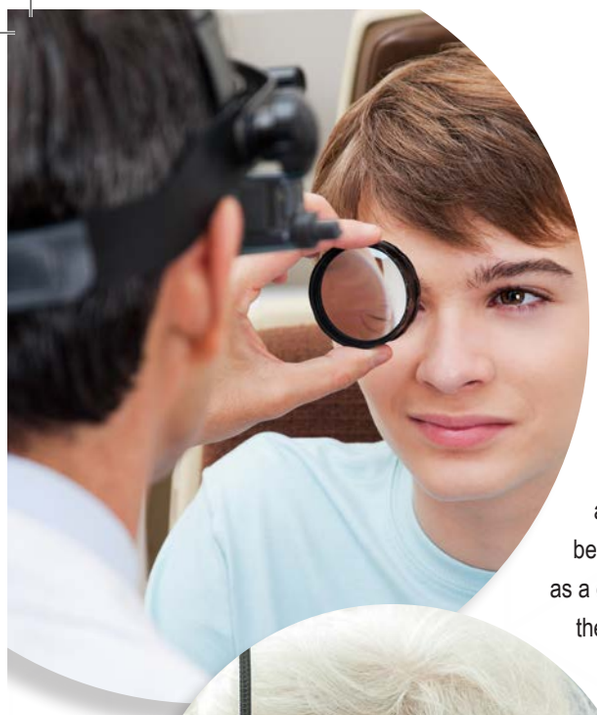
Despite the establishment of diagnostic advances, new treatment options, low-vision aids, and assistance from specialist services, the management IRDs remains largely suboptimal.[2] Since major diagnostic advances did not go along with the development of vision-sparing or -restoring treatments, IRDs have long been thought of as largely incurable diseases.[20] Despite some advancement and new treatment options, not all IRDs have proven treatments that halt progression or restore lost vision.[31] Therefore, current management for some IRDs still consists only of symptomatic alleviation, including refractive correction, use of tinted spectacles/contact lenses, and low vision aids.[31] Over the last decades, the view of IRDs being untreatable has changed, as novel therapeutic options started to be explored, and some treatments transitioning into the clinical setting.[2] In fact, major advances in the study of IRDs have started exploration to develop treatments as part of the emerging field of precision medicine.[32] As a result, the growth of clinical trials for IRDs has increased rapidly over the past decade and is expected to further accelerate as more therapeutic possibilities emerge and qualified participants are identified.[32] Some of the therapies currently under investigation, including gene therapy, stem cell therapy, retinal prosthetics, and even direct brain stimulation, aim to halt disease progression, to return some degree of sight, or to actively stimulate sight (**Table**).[2] Therefore, this is now an exciting time in regards to IRD patient care, with healthcare professionals finally being able to provide treatments that can potentially prevent blindness for patients with certain IRDs.[33] Significant advances have been made over the past 25 years in retinal prosthesis systems, with the development of several different novel engineering and surgical approaches leading to partial visual restoration, improvement in coarse objective function, and increased performance of everyday tasks.[34] Cell-based strategies, including autologously derived induced pluripotent stem cells, have advanced and overcome technical and ethical concerns, with initial data from clinical trials demonstrating promising results.[35] Among the innovative treatments being developed, gene therapies are currently the most promising, with the introduction of the first FDA- and EMA-approved gene therapy (voretigene neparvovec) delivering healthy copies of the defective gene to target tissue and paving the way for continued IRD gene-therapy research.[2,36]

Innovative Research in IRDs[32]

For many years, IRDs have been thought of as a group of inherited disorders with no available treatments or cures. This old adage is being overturned by the efforts of vision scientists and an exciting cohort of clinical trials based on sound preclinical data. Although these efforts are still evolving, the possibilities for making a significant impact on the lives of individuals with IRDs has never been greater. These advances and clinical studies led to the first approved gene therapy treatment (voretigene neparvovec) paving the way for additional innovative research. Advances in DNA and RNA therapies, cell transplantation, and combinatorial therapeutics are expected to be major drivers in innovative therapies, with over 30 clinical trials being either completed or underway to explore the potential of gene therapy as well as other technologies for different types of IRDs, such as retinitis pigmentosa, X-linked retinitis pigmentosa, Leber congenital amaurosis, achromatopsia, Usher syndrome, and Stargardt disease.

Therapeutic Options Being Studies for Inherited Retinal Diseases

Gene Therapy[9]	Advances in DNA delivery to the retina have led to a new era of research into gene therapy for IRDs, with the retina being a favorable target for administering genetic vectors due to its immunoprivileged environment, direct visibility, and multiple methods to assess sensitivity and function
Retinal Prosthetics[34]	Retinal prosthesis have demonstrated partial visual restoration, with improvement in both coarse objective function and performance of everyday tasks
Cell Therapy[35]	Despite significant challenges, stem-cell therapies are reaching clinical applications in the retina with initial data from clinical studies confirming advantages
Direct Current Stimulation[37]	Ocular current stimulation with weak current intensities has shown positive effects on retinal nerve cells, which indicates that neurodegenerative ocular diseases could be treated with current stimulation of the eye



Gene Therapies for Patients with Inherited Retinal Diseases

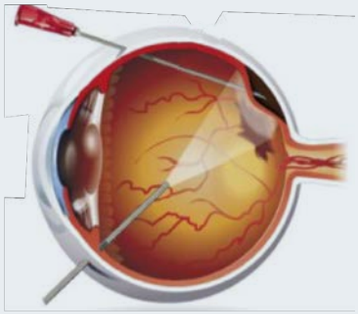
The immune-privileged characteristics of the eyes and the continuous identification of causative genes indicate that gene therapy holds great promise for the treatment of patients with IRDs.[5] Gene therapy has overcome many barriers to evolve from fundamental science to clinical development and has been demonstrated to be relatively safe in multiple clinical trials.[9,33] In addition to proven safety, gene therapy has been shown to be an effective treatment that is now becoming available for patients with previously untreatable IRDs.[32] Gene therapy has been thought of as a compelling treatment approach because of the monogenic nature associated with most IRDs, with the retina—due to its immunoprivileged environment, direct visibility, and multiple methods to assess sensitivity and function—being a favorable target for administering genetic vectors.[9] Generally,

> The development of gene augmentation therapy has created a highly promising avenue for treating various IRDs

gene therapy involves a subretinal or intravitreal injection of a viral vector, which infects target cells to deliver a therapeutic gene.[9] The approval of the first gene therapy (voretigene neparvovec) for patients with Leber's congenital amaurosis, due to mutations in the RPE65 gene, was just the beginning of gene therapy, with additional novel therapies and indications being tested through clinical trials.[16] The development of gene augmentation therapy has created a highly promising avenue for treating various IRDs in the future, which will certainly greatly increase patient's health-related quality-of-life.[36] Although gene therapy holds promise for treating a range of IRDs, careful selection of target diseases, choice of outcome measures, and the surgical challenges of vector delivery need to be considered.[2] In addition, the efficacy of retinal gene therapy depends upon accurate IRD diagnosis (i.e., phenotype and genotype).[2] Natural history studies, long term follow-up of patients receiving gene therapy, and advances in genetic testing and molecular diagnostics should be studied further thought clinical research to address these potential issues associated with genetic therapy and to expand the number of IRDs that can be treated with gene therapy.[2] Patient registries can also assist in gathering information to advance treatment outcomes.

Gene Therapy[2]

- Gene therapy is a safe and effective treatment that is now becoming available for patients with previously untreatable inherited retinal diseases
- Voretigene neparvovec became the first FDA- and EMA-approved direct gene therapy, and the Australian TGA followed suit in August 2020, representing a landmark in gene therapy to treat ocular conditions



- More approvals are projected to follow, with clinical trials underway for many other inherited retinal diseases
- Various strategies exist to target the many different genetic mutations that result in retinal pathology and have demonstrated efficacy in clinical trials
- The success of retinal gene therapy depends on the accurate diagnosis (phenotype and genotype) of patients suspected to have inherited retinal diseases

Inherited Retinal Disease Patient Registries

Prior to the 2018 approval of the gene augmentation therapy, voretigene neparvovec, by the FDA for retinal disease caused by biallelic pathogenic variants in the RPE65 gene, there were no approved therapies for any IRD.[38] While still the only FDA-approved therapy, there is now a vigorous pipeline of clinical trials with promising therapies, largely due in part to a 50-year history of investment and advocacy by people affected by IRDs.[38] Among patients with IRDs, great diversity is seen regarding age of onset, rate of disease progression, and mode of inheritance.[38,39] Patient registries collect information on a voluntary basis about individuals with a specific disease and can provide information for participants of varying health status who may be willing to participate in clinical trials.[39] Therefore, patient registries can provide health care professionals and researchers with first-hand information about people with IRDs, both individually and as a group, and over time, increase the understanding of IRDs.[39] Clinical characterization of patients, supported by a comprehensive genetic testing program, and natural history studies are critical to advance IRD diagnosis and management.[38] Through implementation of a patient registry, the patient perspective of disease as well as access to patients with IRDs can be facilitated through an integrated source of information about, and connection to, all people with an IRD; and registries to share those data, de-identified, with researchers and partners, in order to accelerate the development of treatments and cures.[38,39] Patient registries provide a secure database to collect data about people with IRDs.[38] Advances in molecular biology and therapeutics over the past three decades have transformed both our understanding of the pathways to blindness in IRDs and had paved the way for active interventions, with the first gene-replacement therapy – voretigene neparvovec – being approved; however, significant gaps remain in the knowledge of the natural history of IRDs. Natural history data can assist in providing accurate prognostic information to patients and their families and can assist in the assessment of emerging therapies in clinical trials. As new therapies are being developed, drugs are repurposed, and older treatments are reevaluated, IRD patient registries can assist in providing guidance to researchers regarding the appropriate timing of interventions, suitable outcome measures for clinical trials, patient history, and facilitate outcomes analysis. There is still a lot of knowledge missing in regards to IRD patients and treatment, and patient characterization is needed in order to improve this knowledge. For example, data in My Retina Tracker Registry helps increase understanding of how common each type of IRD is, how IRDs impact people's lives, how IRDs progresses, the genes that cause specific IRDs, and helps researchers and companies to efficiently find people who might be interested in participating in research studies and clinical trials.[40,41] Therefore, patients should be informed of registries and information about research in the field, including clinical trials (**Table**).

Examples of Patient Registries

FOUNDATION FIGHTING BLINDNESS My Retina Tracker® Registry	The **My Retina Tracker Registry** is a research database of people and families affected by IRDs (including retinitis pigmentosa, Leber congenital amaurosis, Stargardt disease, Usher syndrome, Best disease, choroideremia, and achromatopsia) designed to share de-identified information within research and clinical communities about people with IRD to help accelerate the discovery of treatments and cures.[40,41]
National Ophthalmic Disease Genotyping Network eyeGENE® A program of the National Institutes of Health	The National Ophthalmic Disease Genotyping and Phenotyping Network (**eyeGENE**) is a genomic medicine initiative created by the National Eye Institute (NEI), part of the National Institutes of Health (NIH), in partnership with clinics and laboratories, with the core mission of facilitating research into the causes and mechanisms of rare inherited eye diseases and accelerating pathways to treatments[42]
Save Sight Registries Seeing Outcomes	The Fight Inherited Retinal Blindness! (**FIRB!**) module draws upon the expertise of an international steering committee with expertise in IRDs, clinical ontology, clinical genetics, electroph ysiology, visual psychophysics, and vitreoretinal surgery to capture minimum data agreed by the steering committee via broad phenotypic groupings based upon the human phenotype ontology nomenclature, in an effort to track natural history and outcomes and to follow the administration of emerging treatments such as drug, gene, and cellular therapies in patients with IRDs[43]

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