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ADVANCES IN INHERITED RETINAL DISEASES: Improving Patient Outcomes with Genetic Testing, Treatment Options, Genetic Counseling, and Advocacy

Prevalence of Inherited Retinal Diseases and Rate of Genetic Testing

Inherited retinal diseases constitute a large group of rare monogenic diseases that primarily affect the retina, resulting in vision impairment and ultimately blindness in a lot of patients.[1] Although prevalence may vary by region or population, approximately 1 in 2,000 people have inherited retinal diseases based on global estimates.[2-5] These diseases are caused by gene error and are sometimes referred to as genetic eye diseases, hereditary retinal diseases, inherited retinal dystrophies, or inherited retinal degenerations.[6] Over the next few decades, severe vision impairment and blindness in the global population is predicted to increase with the increasing aging population worldwide.[7] Around one-third of cases of blindness or severe visual impairment have a genetic basis, either as part of a multifactorial etiology or as the direct result of genetic mutations, as is the case in patients with inherited retinal diseases.[8,9] Genetic testing is particularly relevant as gene-targeted therapies for inherited retinal disease subtypes have become available.[1] Early genetic testing is crucial to determine if patients are eligible for the approved gene therapy and clinical trials.[1] However, despite the advantages associated with early genetic testing, a recent analysis found that genetic testing was infrequently used, with only 1.5% of individuals with inherited retinal diseases undergoing genetic testing ordered from an ophthalmologist's office.[10]

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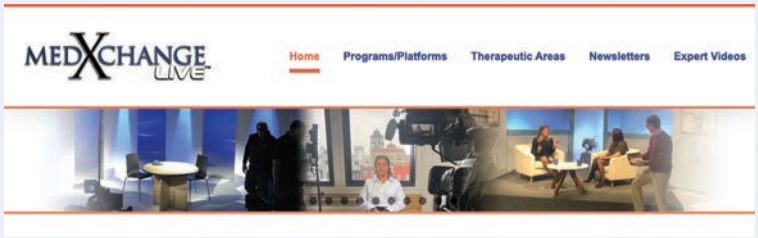


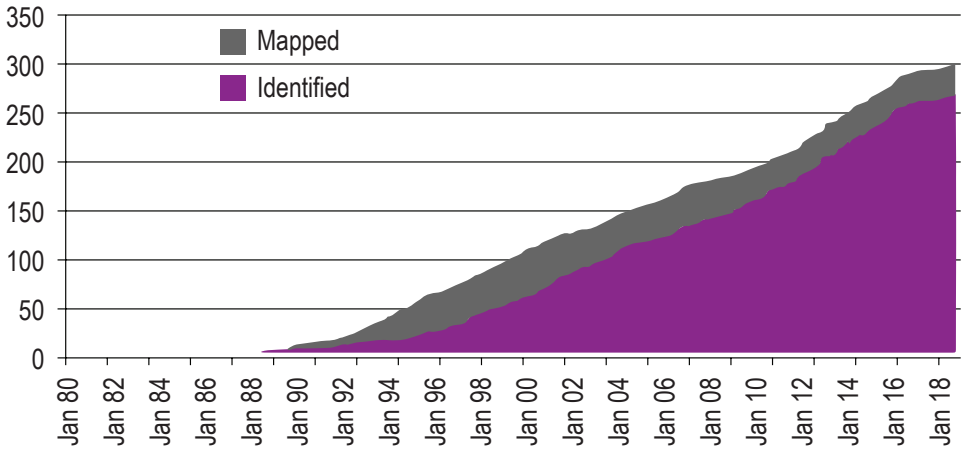
Genes Related to Inherited Retinal Diseases Continue to Be Discovered

Inherited retinal diseases can occur when one or more genes are not working properly.[2,11,12] Inherited retinal diseases are enormously heterogeneous from a genetic standpoint, with over 280 genes cloned to date and over 300 mapped.[13-15] Of the genes related to inherited retinal diseases that have been discovered, 100 have been discovered over the past 10 years.[13-15] Each of the over 300 causal genes discovered so far has numerous variants associated with diseases that range from point mutations to large changes, such as deletions and duplications.[13-15] As science continues to advance uncovering more genes related to inherited retinal diseases, genetic testing technology also continues to evolve.[16]

Mapped and Identified Retinal Disease Genes 1980 – January 2023[14]

317 Disease-causing genes mapped as of January 2023

281 Disease-causing genes cloned as of January 2023



Retesting after a Negative Genetic Testing Result

If genetic testing results are negative for pathogenic variants, this could either indicate that the patient does not have a genetic cause of disease or that the molecular cause of the disease has not been identified, which is more often the case.[22,23] It is possible that patients with suspected inherited retinal disease receiving a negative genetic test result may harbor variants in currently unknown inherited retinal disease genes or in regions of known genes not identified using the genetic testing method.[23] In these patients, genetic reevaluation would be valuable and, because new pathogenic genetic variants are continually being identified, should be considered every 2 to 5 years.[23] Retesting may be dependent on the mode of genetic analysis originally used.[23] For example, reanalysis of existing genetic testing data, for example, in patients having previous whole exome or whole genome analysis, may be simpler and cost less than a complete reevaluation.[23] In those patients having a genetic retest conducted, it is important to assess whether the testing used will be substantially different than the previous analyses, as diagnostic tests may not be updated regularly.[23]

> Retesting should be considered in patients with suspected inherited retinal disease with negative genetic testing results.

Genetic Testing: Innovations and Impact

The genetic heterogeneity of inherited retinal diseases can make it challenging to reach a specific diagnosis.[13-16] A single gene may be associated with multiple phenotypes.[17,18] Genetic testing has become the standard for reaching a more precise diagnosis.[1] A more accurate diagnosis helps inform the best course of action for future medical management.[1] The American Academy of Ophthalmology recommends genetic testing for most patients suspected to have an inherited retinal disease.[18] Genetic testing can help identify the genetic variant in up to 80% of people with inherited retinal diseases.[19] Genetic testing allows physicians to confirm a genetic diagnosis more precisely in most patients by identifying and confirming the genetic cause of vision loss or impairment.[18] Clinical assessment and genetic testing of patients with inherited retinal diseases go hand in hand, and one should not be interpreted without the other to ensure accuracy.[18] Knowing the precise genetic cause of vision loss or impairment is important because such information can help patients and physicians to have a better understanding of how vision may change over time and to understand additional potential health impacts in other areas of the body in addition to the eye.[11,18]

Genetic testing can help identify the genetic variant in up to 80% of people with inherited retinal diseases.

Genetic testing can also help physicians to assess if patients are eligible for clinical trials and emerging treatments.[1] Importantly, genetic testing can weigh the potential risks of other family members and help physicians and genetic counselors to educate patients and family members about available support groups and low-vision resources.[17] It is well established that early genetic testing allows physicians to detect inherited retinal diseases sooner, and uncover more possibilities for patients, such as potential treatments or clinical trials.[19,20] However, despite the known advantages of early genetic testing, it is still underused in the ophthalmologist offices, with only 1.5% of individuals with inherited retinal diseases having genetic testing ordered from their ophthalmologist.[10]

Family History Alone Is Not Enough

Obtaining a thorough family history can help assess a patient's risk or identify a likely pattern of inheritance, but family history alone does not tell the full story. Although most patients who have no known family history have autosomal recessive disease, some may have dominant disease due to a *de novo* pathogenic variant. Others may have a dominant disease exhibiting incomplete penetrance. Still there are those who may have no known family history of X-linked disease.[8,21] In fact, nearly 50% of patients with retinitis pigmentosa have no known family history of retinal disease.[21] Therefore, family history along with genetic testing and clinical examination are needed to establish the diagnosis of inherited retinal diseases.

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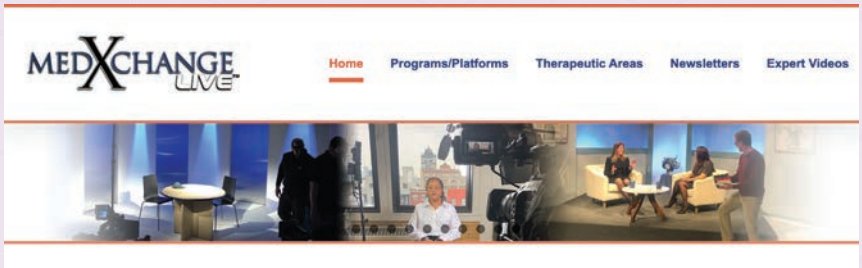

Inherited Retinal Diseases: A Heterogenous Group of Monogenic Diseases

Inherited retinal diseases comprise a large, heterogenous group of monogenic diseases exhibiting autosomal dominant or recessive, X-linked, and mitochondrial inheritance patterns.[1] Life threatening complications, such as cystoid macular edema, foveoschisis, macular hole, macular pucker, vitelliform lesions, foveal atrophy, and macular neovascularization, occur across a spectrum of inherited retinal diseases. Possible signs and symptoms, age of onset, and estimated prevalence varies within the group of inherited retinal diseases.[1] Most inherited retinal diseases primarily effect the retina and may result in vision impairment and ultimately blindness, but possible signs and symptoms vary for each disorder.[1] It has been estimated that 1 in 2,000 people have inherited retinal diseases,[1-5] but estimated prevalence rate for each of the inherited retinal diseases varies widely.

Inherited Retinal Disease	Possible Signs and Symptoms	Age of Onset	Estimated Prevalence
Retinitis pigmentosa[24,25]	Nyctalopia; blind spots; tunnel vision; loss of central vision	Presents in childhood or adulthood	Up to 1 in 3,000
X-linked retinitis pigmentosa[24-27]	Rapid progression of vision loss; legal blindness; early nyctalopia	Early onset	5% to 15% of patients with retinitis pigmentosa
Usher syndrome[28,29]	Three different clinical types (I, II, and III) with variable symptom severity and presentation; partial or total hearing loss and vision loss; loss of night vision occurs first; blind spots; tunnel vision	Presents at birth (type I, type II), or by adolescence or adulthood (type III)	Up to 1 in 6,000
Stargardt disease[30,31]	Slow, progressive loss of central vision; nyctalopia; color blindness	Presents in late childhood to early adulthood	Up to 1 in 8,000
Cone-rod dystrophy[32]	Decreased visual acuity; photophobia; loss of color vision; scotomas; loss of peripheral vision; legal blindness by mid-adulthood	Presents in childhood	Up to 1 in 30,000
Achromatopsia[33]	Partial or total absence of color vision (can only see black, white, and shades of gray); photophobia; nystagmus; reduced visual acuity; hyperopia	Presents at birth or early infancy	Up to 1 in 30,000
Leber congenital amaurosis[34]	Vision loss (at birth to early infancy); photophobia; nystagmus; hyperopia; keratoconus; Franceschetti's oculo-digital sign (eye poking, pressing, and rubbing)	Usually presents at infancy	Up to 1 in 33,000
Choroideremia[35]	Progressive atrophy of the outer retina and inner choroid; nyctalopia in early childhood; progressive loss of peripheral visual field and visual acuity; all individuals will develop blindness, most commonly in late adulthood	Usually presents in early childhood	Up to 1 in 50,000
Bardet-Biedl syndrome[36,37]	Nyctalopia; blind spots merge to produce tunnel vision over time; blurred central vision; legally blind by adolescence or early adulthood; renal malformations; obesity; postaxial polydactyly; hypogonadism	Ocular symptoms present in first decade of life	Up to 1 in 160,000 newborns

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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Various Treatment Options Being Studied for Inherited Retinal Disorders

Inherited retinal diseases are a group of rare eye disorders that can lead to serious vision impairment or loss.[1] Despite research being conducted, millions of people worldwide with inherited retinal diseases are still without available treatment options.[38] Genetic testing further supports clinical findings and has become the standard for reaching a more precise diagnosis.[17,18] A more accurate diagnosis helps inform the best course of action for future medical management.[17] In addition to the approved gene therapy, there are several other therapies in development for the treatment of inherited retinal disorders, including cell therapy, visual prosthetics, optogenetics, and RNA-based therapies.[15,39]

Types of Therapies Being Studied for the Treatment of Inherited Retinal Disorders

Additional gene replacement therapies[40]	• The only approved treatment available for inherited retinal diseases is a gene replacement therapy using adeno-associated virus (AAV) and nonviral delivery vectors, voretigene neparvovec • Additional gene-based therapies that are different from voretigene neparvovec but also have the potential to treat inherited retinal diseases are being studied • These include genome editing via the CRISPR/Cas9 system
Cell therapy[41]	• Cell therapy includes the introduction of stem cells to replace degenerated cells through delivery to target tissues (i.e., photoreceptors and retinal pigment epithelium) • Cell therapies are expected to slow disease progression and restore some visual functions, but there are several limitations to such therapies
RNA-based therapies[9]	• RNA-based therapies are a novel approach within precision medicine that have demonstrated success, particularly in rare diseases • Three antisense oligonucleotides (ASOs) are currently in development for the treatment of specific inherited retinal diseases • RNA-based therapies have the potential to bring meaningful vision benefit to people living with inherited retinal diseases that can lead to blindness
Retinal prostheses[42]	• These implantable devices aim to restore the vision of blind patients suffering from retinal degeneration, mainly by artificially stimulating the remaining retinal neurons • Some retinal prostheses have successfully reached the stage of clinical trials; however, these devices can only restore vision partially
Optogenetics[43]	• Primarily aimed at rendering secondary and tertiary neurons of the retina light-sensitive in order to replace degenerate or dysfunctional photoreceptors • Optogenetic approaches provide a causative gene-independent strategy, which may prove suitable for a variety of patients with inherited retinal diseases • Optogenetic approaches to vision restoration yielded promising results in preclinical trials

Clinical Trials for Inherited Retinal Disease Treatments

Over 30 clinical trials are either completed or underway for different types of inherited retinal diseases, such as retinitis pigmentosa, X-linked retinitis pigmentosa, Leber congenital amaurosis, achromatopsia, Usher syndrome, and Stargardt disease.[44] However, the number of clinical trials for inherited retinal diseases has increased rapidly and is expected to advance as therapies emerge and additional qualified patients are identified.[45]

Impact of Vision Loss on Quality-of-Life

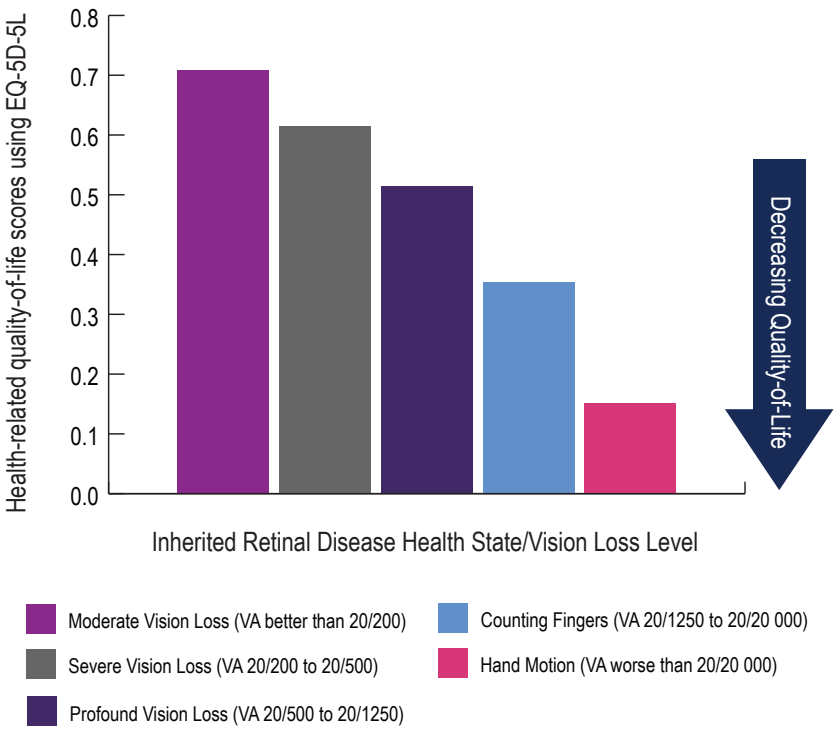
In the field of clinical ophthalmology, many of the commonly used visual function assessments do not effectively reflect the significant morbidity of inherited retinal diseases and the effect of these disorders on patient quality-of-life.[46] In the last decade, emphasis has been placed on the development and implementation of patient-performance or task-focused assessments, that may have greater ability to demonstrate the improvement or preservation of the patient's quality-of-life provided by treatment interventions.[46] Inherited retinal diseases can cause severe and progressive loss of peripheral visual field, nyctalopia, and ultimately loss of central vision, all of which are associated with severe impairment in functional vision and quality-of-life as well an impact on patients' psychological health, relationships, and family life.[47] Many areas of quality-of-life are affected by this level of vision loss including independence, mental health, and the ability to engage socially with others.[47] Patients' declining vision limits their opportunities during their formative teenage and young adult years, the same period in which their peers' opportunities are expanding.[47] Over the next few decades, by the year 2050, mild vision impairment is predicted to affect 360 million people altogether, moderate-to-severe vision impairment is predicted to affect 474 million people, and the number of people with blindness in the global population is predicted to increase to 61 million.[7] As a patient loses their vision, it has a major impact on the patient's quality-of-life.[47]

Patient Psychological Burden and Concerns Associated with Vision Loss

With changes to a patient's day-to-day life also comes a psychological burden. Patients may be anxious about the loss of their sight and the adaptations they have to make. Patients often become depressed as they lose some of their independence and are forced to rely more and more on family members.[48,49] This leaves patients with feelings of helplessness and frustration.[48-51] In addition to the impact of vision loss on quality-of-life, patients with inherited retinal diseases may have additional concerns causing psychological burden, including[50,51]:

- Passing down inherited retinal disease to children;
- Challenges in activities of daily living;
- Issues with mobility;
- Difficulty working or finding employment;
- Problems interacting with family and friends; and
- Overwhelming fear of blindness.

Decrease in Quality-of-Life with Worsening Vision in Patients with Inherited Retinal Diseases[47]



EQ-5D-5L= 5-level version of EuroQol-5 Dimension; VA=visual acuity.

The Importance of Genetic Counseling

Genetic testing not only allows for accurate identification of inheritance pattern, but also improves genetic counseling for affected individuals and their families.[52] Genetic counselors are critical to correctly support patients and caregivers in understanding the genetic diagnosis and the genetic testing results.[53] Therefore, genetic counseling should be provided before and after testing.[53] Before testing, patients and caregivers should be informed and prepared regarding the test limitations and the implications of the results.[18] Gene therapy, as a breakthrough chance to maintain or even improve current visual ability, has an emotional impact on patients, caregivers, and even clinicians.[53]

Therefore, the eligibility criteria and the possibilities to access gene therapy should be fully clarified for patients and caregivers through genetic counseling to ensure its applicability to as many patients as possible.[53,54]

> Genetic counseling should be provided before and after genetic testing.

The management of inherited retinal diseases should be promoted by optimizing a multidisciplinary approach, which is essential to meet the complex care needs of each patient.[55] Psychological support through genetic counselors remains critical, from the communication of the clinical diagnosis throughout the care pathway.[53] Genetic counseling allows community-based retina specialists to partner with genetic counselors, in person or via telephone or computer, to help disclose results to individuals and manage conversations regarding complex results and risks to family members.[23] It is important for patients to understand through genetic counseling that receiving genetic testing does not guarantee that they will receive a molecular diagnosis for their inherited retinal disorder, and that a positive genetic test result will not necessarily qualify them for a clinical trial or therapy.[56] In addition, it should be understood that not all of the genes and variants associated with inherited retinal diseases have been identified, and therefore testing may not detect the disease-causing variant for all patients.[56]

Access to Specialty Care and Genetic Counseling Increased through Telemedicine

Early and accurate diagnosis is necessary for individuals with inherited retinal disorders in order to enable patient decision-making, to identify suitable clinical studies or available treatment opportunities, and to improve patient outcomes.[23] Therefore, ensuring that patients with inherited retinal diseases have access to specialty care is very important, including access to genetic testing and to care from an inherited retinal disease specialist.[23] Access to inherited retinal disease specialists varies depending on geographic area, with some patients having to travel long distances to receive specialized care.[57] Therefore, telemedicine has become very useful in helping patients to obtain access to inherited retinal disease specialists, genetic testing, and genetic counseling.[57] Raising awareness among ophthalmologists, optometrists, and other health care providers of the need for and availability of specialized inherited retinal disease care through telemedicine for patients who may not have access to an inherited retinal disease specialist could also be beneficial for reducing the time for patients to obtain a diagnosis and necessary specialty care.[57,58] Telemedicine-based genetic counseling services are becoming more widely available to support geographically or economically challenged patients in accessing specialists in ocular genetics, especially in the United States.[23,57,58] Through telemedicine, electronic patient monitoring becomes available and patient consultations tan take place via telephone during which ocular genetic counselors can provide remote counseling to overcome geographical constraints and expand access to patient care.[58] Telemedicine allows community-based retina specialists or physicians to partner with telephone-based genetic counselors to help disclose results to individuals and manage conversations regarding complex results and risks to family members.[23] While adequate genetic testing resources and specialty providers may not be unavailable in many geographic regions, improved understanding of genetics and increased use of telemedicine may help meet the needs of patients to fully understand their inherited retinal disease diagnosis.[23]

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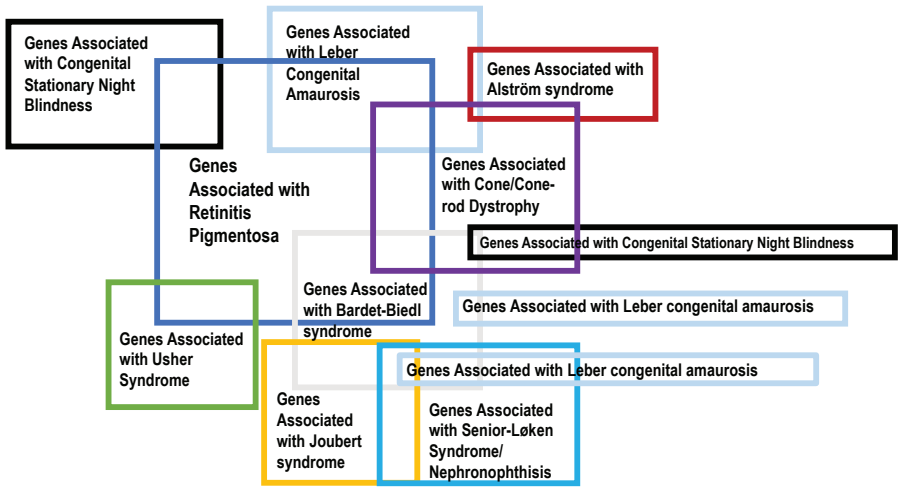
Genetic Overlaps of Inherited Retinal Diseases

Inherited retinal diseases present challenges in molecular diagnostics because of their high genetic heterogeneity, overlapping clinical presentations, and variability in inheritance patterns.[18,23] The genetic heterogeneity of inherited retinal diseases can make it challenging to reach a specific diagnosis.[23] A single gene may even be associated with multiple phenotypes, such as seen in multiple genes overlapping in retinitis pigmentosa, Bardet-Biedl syndrome, cone/cone-rod dystrophy, congenital stationary night blindness, Joubert syndrome, Leber congenital amaurosis; nephronophthisis, Senior-Løken syndrome, and Usher syndrome.[18,24,25] Genetic testing further supports clinical findings and has become the standard for reaching a more precise diagnosis for patients with inherited retinal diseases.[23] A more accurate diagnosis helps inform the best course of action for future medical management.[17,34]

Overlap in Genes Associated with Various Inherited Retinal Diseases[18]



An Investigational Gene Therapy for X-linked Retinitis Pigmentosa

Treatment of X-linked retinitis pigmentosa remains challenging, and current treatments are not effective enough in restoring vision.[68] Gene therapy of X-linked retinitis pigmentosa, capable of restoring the functional RPGR gene, showed promising results in preclinical studies and clinical trials; however, there are currently no approved treatments.[69] Botaretigene sparoparvovec is being investigated for the treatment of patients with X-linked retinitis pigmentosa caused by disease-causing variants in the eye-specific form of the RPGR gene.[68-71] Through a one-time administration, botaretigene sparoparvovec is designed to deliver functional copies of the RPGR gene to counteract the loss of retinal cells with the goal of preserving and potentially restoring vision for patients with X-linked retinitis pigmentosa.[70] The Phase 3 LUMEOS clinical trial (NCT04671433) is actively dosing patients to study botaretigene sparoparvovec for the treatment of patients with X-linked retinitis pigmentosa with disease-causing variants in the RPGR gene.[71] Botaretigene sparoparvovec has been granted Fast Track and Orphan Drug designations by the US Food and Drug Administration (FDA) and PRIority MEdicines (PRIME), Advanced Therapy Medicinal Product (ATMP), and Orphan designations by the European Medicines Agency (EMA).[68,69]

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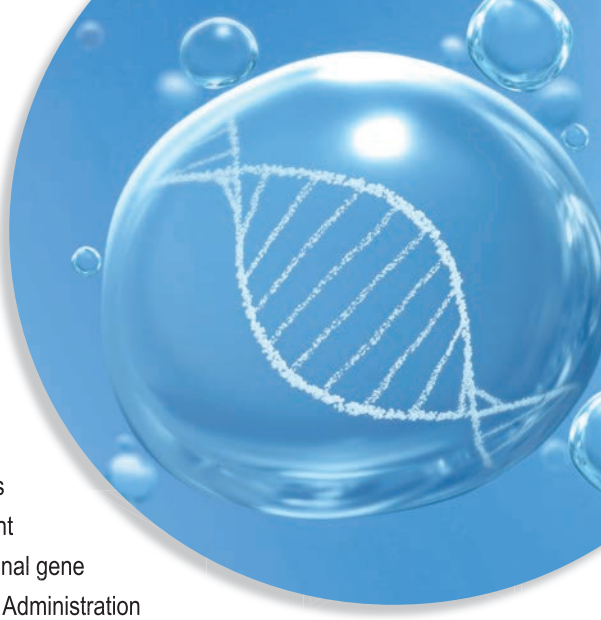

Gene Therapy for Inherited Retinal Diseases

As a one-time treatment with the potential to provide a curative clinical benefit, gene therapies offer an advantage over other treatments for a wide range of diseases.[59,60] As a heterogenous group of orphan eye diseases that typically result from monogenic mutations, inherited retinal diseases are good disorders to be treated with gene therapies.[40] Therefore, the retina has been thoroughly investigated over the past two decades for gene therapy interventions for inherited retinal diseases because it is immune-privileged, enclosed, and easily monitored.[61] Although historically limited by their previously incurable nature, advances made in the knowledge of genes and gene therapies have led to positive changes in the treatment paradigm for inherited retinal diseases.[9] Successful phase 1 through phase 3 studies with subretinal gene augmentation therapy resulted in the approval of voretigene neparvovec by the US Federal Drug Administration (FDA) in 2017 and the European Medicines Agency (EMA) in 2018 for the treatment of RPE65-associated retinal dystrophy.[62-64] The results of these studies were promising, with the majority of patients experiencing a significant increase in vision at reduced light levels and longer-term persistence of therapeutic effect up to 4 years reported.[62,65,66] Real-world data after approval showed the effectiveness of voretigene neparvovec therapy with stable median best-corrected visual acuity and mean retinal thickness, and improvements of low-luminance visual acuity, chromatic full-field-stimulus-threshold testing, and chromatic full-field-stimulus-threshold testing up to 32 months, with treatment effects being superior in pediatric patients.[67] Following the approval and successful clinical use of voretigene neparvovec, there has been a worldwide effort to find additional inherited retinal diseases that can be treated and potentially cured with retinal gene therapy.[59,62] The potential ability of gene therapies to provide durable and potentially curative health benefits for patients with inherited retinal diseases as well as demonstrated advantages of the eye as being a compartmentalized, small, immune-privileged structure has led to continued and increasing research efforts towards finding additional effective and safe gene therapy treatments.[59]

> Following the approval and successful clinical use of voretigene naparvovec, there has been an effort to find other inherited retinal diseases that can be treated with gene therapy.

RNA Therapies: A Promising Treatment for Inherited Retinal Diseases

Due to the approval of an antisense oligonucleotide (ASO) over 25 years ago for the treatment of cytomegalovirus retinitis, the retina has continued to play an important role in RNA-based therapy development.[72] RNA therapies are highly specific and target the underlying genetic cause of a disease, but RNA therapies differ from gene therapy in several ways, including their mechanism of action, delivery, and permanency of effect.[45,73-75] Although it differs from gene therapy, which has been approved for the treatment of inherited retinal diseases (voretigene neparvovec), RNA therapy is a promising approach for treating these disorders and has demonstrated success in rare diseases.[45,75] In the setting of inherited retinal diseases, RNAs act at the RNA level, are administered via intravitreal injection, and have long-lasting but not permanent effects.[9] There are three investigational RNA-based therapies for inherited retinal diseases currently in clinical development for treatment of specific inherited retinal diseases, but none are yet approved.[9] The RNA-based therapies in development have demonstrated the potential to bring meaningful vision benefit to those with blinding inherited retinal diseases with early promising data in clinical trials.[9,76] These ASO therapies are targeting disease mutations which result in splicing defects.[9,77] One experimental drug has demonstrated promising results in a phase 1/2 clinical trial in patients with Leber congenital amaurosis, demonstrating vision improvement after 12 months of treatment.[62,77,78]

Evaluating Retinal Gene Therapy Treatment Success

After voretigene neparvovec delivers a functioning copy of a missing gene, protein transcription is enabled in retinal cells, a complex network of functional regeneration is begun, and visual functions are restored.[62,66,67] For voretigene neparvovec, the primary treatment target cell is the retinal pigment epithelial cell, and the photoreceptor is the secondary target in terms of visual function restoration or preservation.[79] The extent of functional regeneration is individualized in each patient.[79] Diagnostic and functional tests that have been used routinely by ophthalmologists cannot define small, subtle changes in visual functions that may be needed to assess the efficacy, safety, and durability of voretigene neparvovec treatment.[79] The strategy that has been recommended in the medical literature is the combination of multiple independent tests – the full-field light sensitivity threshold (FST), chromatic pupil campimetry (CPC), and dark-adapted chromatic perimetry (DACP) – that allows a multimodal insight into retinal function.[79] This test battery will best describe rod function change.[79] The recommendation for the use of FST, CPC and DACP is based on the clinical applicability, overlapping multidimensionality, short recoding duration, and patient orientation associated with the tests.[79]

Test Battery to Assess Voretigene Neparvovec Gene Therapy Efficacy and Durability[79]

Test Battery

FST

Using chromatic stimuli allows for the detection of the dark-adapted threshold for the entire retina

DACP

Can detect the retinotopically correct functional rescue of the rods related to the treated area

CPC

Provides pupillographic readouts, which are needed because the pupillary reflex depends on the photoreceptor cell number that drives the input into the reflex circuitry

CPC=chromatic pupil campimetry; DACP=dark-adapted chromatic perimetry; FST=full-field light sensitivity threshold.

Importance of Patient Advocacy Groups

Patients should be educated and provided with educational materials on disease background and treatment options so they can better understand inherited retinal diseases and available treatment options.[45] Educating patients regarding research in progress and new potential treatment options is not only informative but also very uplifting for patients.[45] Therefore, patients should be encouraged to be involved with organizations such as Foundation Fighting Blindness and/or social media groups as they can provide educational resources.[80] There are also several individual disease-focused foundations that provide great support to patients and families with inherited retinal diseases.[81,82] Overall, involvement with patient advocacy groups can be really empowering for patients and can be a great way for patients to learn about and get control of their condition, rather than being controlled by it.

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