

**Selected Observational Studies and Clinical Treatment Trials Recruiting Children with Genetic Eye Disorders<sup>1</sup>**

Condition(s) / Clinical Trials Identifier <sup>2</sup>	Gene (if applicable)	Intervention (if applicable)	Phase <sup>3</sup>	Status as of date accessed (2/8/2021)	Age	Location(s)	Contact Information
Achromatopsia NCT02935517 <sup>a</sup>	<i>CNGA3</i>	Interventional: Gene replacement therapy (AAV-CNGA3)	1/2	Recruiting start date: 5/1/17 Last updated: 4/6/2021 Estimated last enrollment: 09/2021	≥ 6 years	Recruiting: 1 Boston, MA 2. NY, NY 3. Cincinnati, OH 4. Portland, OR 5. Miami, FL 6. Gainesville, FL Active/Not recruiting: 7. Philadelphia, 9. SF, CA 10. Madison, WI	Jill Dolgin, PharmD 833-770-2862 advocacy@agtc.com
Achromatopsia NCT02599922	<i>CNGB3</i>	Interventional: Gene replacement therapy (rAAV2tYF-PR1.7-hCNGB3)	1/2	Recruiting start date: 2/2016 Last updated: 4/6/2021 Estimated last enrollment: Sept 2021	≥ 6 years	Recruiting: 1. Boston, MA 2. NY, NY 3. Cincinnati, OH 4. Portland, OR 5. Miami, FL Gainesville, FL 6. Houston, TX Active/Not recruiting: 1. Durham, NC 2. San Fran, CA 3. Cleveland, OH 4. Chicago, IL 5. Madison, WI	Jill Dolgin, PharmD 833-770-2862 advocacy@agtc.com

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Cerebrotendinous Xanthomatosis (CTX) <sup>b</sup> NCT02638220	<i>CYP27A1</i>	Observational: Prevalence of <i>CTX</i> mutations in early-onset bilateral cataracts	NA	Recruiting start date 12/2015 Last updated: 11/9/18	Cataract Dx 2-21 years	Recruiting: 45 US locations (see <a href="http://clinicaltrials.gov">clinicaltrials.gov</a> for complete list)	Travere medical information: <a href="mailto:medinfo@travere.com">medinfo@travere.com</a> 1-877-659-5518
Cerebrotendinous Xanthomatosis (CTX) NCT04270682	<i>CYP27A1</i>	Interventional: Chenodeoxycholic Acid (CDCA)	3	Recruiting start date 01/31/2020 Last updated: 02/5/2021	1 mo to 16 years old	Recruiting: 1. New Orleans, Louisiana 2. Seattle, Washington 3. Austin, TX	Travere medical information: <a href="mailto:medinfo@travere.com">medinfo@travere.com</a> 1-877-659-5518
Galactosemia NCT04902781	Classic galactosemia	AT-007 oral (aldose reductase inhibitor)	2/3	Recruiting: Start date 05/26/2021 Last updated: 05/28/2021 Estimated last enrollment: 09/2021	2-17 years	Recruiting: 1. Aurora, CO 2. Atlanta, GA 3. Ann Arbor, Michigan	Riccardo Perfetti 212 220 9227 <a href="mailto:rperfetti@appliedtherapeutics.com">rperfetti@appliedtherapeutics.com</a> Francesca Lawson 212 220 9256 <a href="mailto:flawson@appliedtherapeutics.com">flawson@appliedtherapeutics.com</a>
Homocystinuria (HCU) NCT03406611	HCU caused by <i>CBS</i> mutations	OT-58 (PEG modified htCBS) subcutaneous	1/2	Recruiting: Start date 01/23/2018 Last updated: 01/05/2021	12-65 years	Recruiting: 1. Aurora, CO 2. Miami, FL 3. Indianapolis 4. Boston, MA 5. NY, NY 6. Philadelphia	Ferianas Grebikas Travere Therapeutics 877 659 5518 <a href="mailto:medinfo@travere.com">medinfo@travere.com</a>
Genetic disorders with obesity NCT03013543	POMC and Leptin Receptor Deficiency, Bardet Biedl, Alstrom, Smith-Magenis	Interventional: Setmelanotide subcutaneous injection	2/3	Recruiting start date: 1/6/17 Last updated: 7/16/21	≥ 6 years	Active, not recruiting: 57 sites (see <a href="http://clinicaltrials.gov">clinicaltrials.gov</a> for complete list)	Rhythm Pharmaceuticals Murray Stewart Olga Ohayon: <a href="mailto:oohayon@rhythmtx.com">oohayon@rhythmtx.com</a>

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Leber Congenital Amaurosis 10 (ILLUMINATE) NCT03913143	<i>CEP290</i> c.2991+165 5A>G Mutation (IVS26) <sup>4</sup>	Interventional: Sepofarsen (antisense RNA oligo-nucleotide)	2/3	Recruiting start date: 4/01/2019 Last updated: 1/11/2021	≥ 8 years	Active, not recruiting: 21 locations in the US (see <a href="http://clinicaltrials.gov">clinicaltrials.gov</a> for complete list)	ProQR Clinical Trials Manager +31 (0)88 1667000 <a href="mailto:info@proqr.com">info@proqr.com</a>
Leber Congenital Amaurosis 10 NCT03872479	<i>CEP290</i> c.2991+165 5A>G Mutation (IVS26)	Interventional: AGN-151587 (EDIT-101)	1/2	Recruiting start date: 3/13/2019 Last updated: 12/22/2020 Estimated last enrollment: 03/2024	≥ 3 years	Recruiting: 1. Miami, FL 2. Boston, MA 3. Ann Arbor, MI 4. Portland, OR	Editas Medicine: 617-401-9007 <a href="mailto:patients@editasmed.com">patients@editasmed.com</a>
Leber Congenital Amaurosis NCT03920007	Biallelic <i>GUCY2D</i>	Interventional: SAR439483 unilateral subretinal	1/2	Recruiting start date: 4/18/2019 Last updated: 7/26/21 Estimated last enrollment: 02/2022	≥ 6 years	Recruiting: Philadelphia, PA	Michael DeSordi, BS 732 921 2093 <a href="mailto:Mike.desordi@atsenatx.com">Mike.desordi@atsenatx.com</a>
Neuronal Ceroid Lipofuscinosis (Juvenile Batten Disease) NCT03307304	<i>CLN3</i>	Observational	N/A	Recruiting start date: 10/11/2017 Last updated: 6/9/21	0-100 years	Recruiting: 1. Bethesda, MD, NIH	An N Dang Do, M.D 301-496-8849 <a href="mailto:an.dangdo@nih.gov">an.dangdo@nih.gov</a>
Neuronal Ceroid Lipofuscinosis (Juvenile Batten Disease) NCT04974307	<i>CLN3</i>	Interventional: optic-to-audio device in pediatric CLN3 patients with poor vision	N/A	Recruiting start date: 7/3/21 Last updated: 8/4/21 Estimated end of recruitment: 01/2025	6-18 years	Recruiting: 1. Bethesda, MD, NIH	An N Dang Do, M.D 301-496-8849 <a href="mailto:an.dangdo@nih.gov">an.dangdo@nih.gov</a>

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Neuronal Ceroid Lipofuscinosis (CLN2) NCT04098211	<i>CLN2</i> (atypical TTP1 deficiency)	Observational	N/A	Recruiting start date 9/23/19 Last updated 7/16/21	≥ 4 years	Active, not recruiting: Orange County, CA	Raymond Wang, MD 714-509-3344 <a href="mailto:rawang@choc.org">rawang@choc.org</a>
Neuronal Ceroid Lipofuscinoses NCT01873924	<i>CLN1-10</i>	Observational	N/A	Recruiting start date: 6/10/2013 Late updated: 4/8/2021	Any age	Recruiting: University of Rochester, NY	Amy Vierhile: 585-275-4782 <a href="mailto:amy_vierhile@urmc.rochester.edu">amy_vierhile@urmc.rochester.edu</a>
Virtual Reality Mobility Assessment of Functional Vision in Retinal Disease NCT04289571	Any retinal dystrophy affecting vision	Observational, interventional	1	Recruiting: Start date: 2/28/20 Last updated: 12/17/20	≥ 5 years	Recruiting: 1. Bethesda, MD 2. Sydney Eye Hospital, Australia	US: Daniel W. Claus, RN: (301) 496-9058, <a href="mailto:daniel.claus@nih.gov">daniel.claus@nih.gov</a> Australia: John Grigg, M.D. 61293827300 <a href="mailto:john.grigg@sydney.edu.au">john.grigg@sydney.edu.au</a>
Retinitis Pigmentosa NCT03845218	Any form of RP	Observational	NA	Recruiting started: 2/19/19 Last updated: 7/28/21	≥12 years	Recruiting: 1. Bethesda, MD	Angel Garced, RN 301 594 3141 <a href="mailto:garceda@nei.nih.gov">garceda@nei.nih.gov</a>
X linked Retinitis Pigmentosa NCT03316560	<i>RPGR</i>	Interventional: Gene replacement therapy (rAAV2tYF-GRK1-RPGR)	1/2	Recruiting start date: 4/18/18 Last updated: 7/15/2021 Expected last enrollment: 08/2022	≥6 years	Recruiting Cincinnati, OH Jacksonville, FL Boston, MA Durham, NC Cleveland, OH Dallas, TX Portland, OR Philadelphia, PA	Jill Dolgin, PharmD 833-770-2862 <a href="mailto:advocacy@agtc.com">advocacy@agtc.com</a>
X linked Retinitis Pigmentosa NCT04671433 & MCT04794101	<i>RPGR</i>	Interventional: AAV5-RPGR	3	Recruiting start date: 12/17/20 Last updated: 7/15/2021	≥5 years	Recruiting: Gainesville, FL Atlanta, Georgia Boston, MA Ann Arbor, MI Houston, TX	Julie Bakobaki +44 203725 2349 <a href="mailto:julie.bakobaki@meirgtx.com">julie.bakobaki@meirgtx.com</a>

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Retinoblastoma NCT01783535	<i>RBI</i> : Bilateral disease with one eye advanced	Interventional: Multiple arms including chemotherapy, plaque therapy, cryotherapy, laser and thermotherapy	2	Recruiting Start date: 2/5/2013 Last updated: 1/28/2021 Expected last enrollment: 11/2021	Any	Recruiting: St. Jude Children's Hospital, Memphis TN	Rachel Brennan, MD 866-278-5833 referralinfo@stjude.org
Retinoblastoma NCT04342572	<i>RBI</i>	Interventional: Intra-arterial melphalan chemotherapy	1	Recruiting Start date: 4/13/2020 Last updated: 10/14/2020	4 months and older	Recruiting: St. Louis Children's Hospital, Missouri	Margaret Reynolds 314 454 6026 Margaret.reynolds@wustl.edu
Smith-Lemli- Opitz-Syndrome NCT01773278	<i>DHCR7</i>	Interventional: Cholesterol & antioxidant supplements	2	Recruiting: Start date: 1/23/13 Last updated: 11/4/2020	<65 years	Recruiting: 1. Aurora, CO (Children's Hospital Colorado)	Ellen Elias, MD 720-777-5401 <a href="mailto:Ellen.elias@childrenscolorado.org">Ellen.elias@childrenscolorado.org</a>
Smith-Lemli- Opitz-Syndrome NCT03720990	<i>DHCR7</i>	Interventional: Cholic acid	1/2	Recruiting start date: 3/27/21 Last update: 5/3/2021	2-25 years	Recruiting: 1. Aurora, CO 2. Omaha, NE	Ellen Elias, MD 720-777-5401 <a href="mailto:Ellen.elias@childrenscolorado.org">Ellen.elias@childrenscolorado.org</a> Sara M Jones, RD 402-559-1747 <a href="mailto:SaraM.Jones@unmc.edu">SaraM.Jones@unmc.edu</a>
Stargardt Disease NCT02402660 <sup>c</sup> & NCT04239625 (extension trial)	<i>ABCA4</i>	Intervention: Oral ALK-001 <sup>5</sup> C20-D3-retinyl acetate	2	Recruiting start date: 3/30/15 Last updated: 7/23/21 Expected end of enrollment: 03/2022	8-70 years	Recruiting: Los Angeles, CA Gainesville, FL Miami, FL NY, NY Active but not recruiting Salt Lake, UT Milwaukee, WI Baltimore, MD	Leonide Saad, PhD 800-287-2755 <a href="mailto:trials@alkeus.com">trials@alkeus.com</a>

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Stargardt Disease NCT04545736	<i>ABCA4</i>	Intervention: Oral metformin	1/2	Recruiting started: 9/11/20 Last updated: 5/4/21	≥12 years	Recruiting: 1. NEI, Bethesda, MD	Allison Bamji, RN 301-451-3437 bamjia@nei.nih.gov
Neurofibromatosis Type 1 NCT03871257 & NCT04166409	NF1 with low grade glioma	Interventional: Selumetinib versus Carboplatin/vincrist ine	3	Recruiting start date: 10/4/2019 Last updated: 07/2021	2-21 years	Recruiting: 96 locations (see clinicaltrials.gov for complete list of sites)	Jason R Fangusaro Pediatric Brain Tumor Consortium Peter M de Blank Children's Oncology Group
Neurofibromatosis Type 1 NCT04201457	NF1 with low grade glioma	Interventional: Addition of Plaquenil to dabrafenib/trametin ib in BRAF V600 mutation positive	1/2	Recruiting start date: 12/17/2019 Last updated: 03/2021	1-30 years	Recruiting: 12 locations (see clinicaltrials.gov for complete list of sites)	Vanetria Stokes, MS 901 595 5762 vanetria.stokes@stjude.org Stacye Richardson, MSHS 901 595 3783 stacye.richardson.stjude.org
Neurofibromatosis Type 1 NCT02343224	Optic gliomas or juvenile pilocytic astrocytoma	Interventional: Pegylated interferon alpha-2b	2	Recruiting start date: 01/21/15 Last updated: 01/26/2021	3-25 years	Recruiting: Children's healthcare of Atlanta, Georgia	Amanda Sanders 404-785-0305 <a href="mailto:Amanda.Sanders2@choa.org">Amanda.Sanders2@choa.org</a>
Neurofibromatosis Type 1 Associated Plexiform  Neurofibromas NCT03962543	NF 1 and peripheral neurofibrom as	Interventional: Mirdametinib PD-0325901 (MEK inhibitor)	2	Recruiting start date: 05/24/19 Last updated 2/4/21	≥2 years	Recruiting: 52 locations (see clinicaltrials.gov for complete list of sites)	Nicole H Leedon: 980-258-3225 clinical@springworkstx.com
Wolfram Syndrome NCT02829268	<i>WFS1</i> , <i>WFS2</i>	Interventional: Dantrolene sodium	1/2	Start date: 01/2017 Last updated: Jan 2021	5-60 years	Active, but not recruiting: St. Louis, Missouri	Stacy Hurst, RN, BSN +1-314-747-3294 <a href="mailto:shurst@wustl.edu">shurst@wustl.edu</a>

Abbreviation: RP = retinitis pigmentosa; AAV = adeno-associated virus vector

**Footnotes:**

<sup>1</sup>Please refer to ClinicalTrials.gov for more information. There are many more gene therapy trials for patients 18 years and older. There is also a registry and natural history observational study for many retinal degenerations. Foundation Fighting Blindness Retinal Dystrophy Registry (any retinal dystrophy, any age) contact: Joan Fisher [Coordinator@MyRetinaTracker.org](mailto:Coordinator@MyRetinaTracker.org) 1-800-683-555

Excellent Review Article on Gene Therapy: Garafalo AV, Cideciyan AV, Héon E, et al. Progress in treating inherited retinal diseases: Early subretinal gene therapy clinical trials and candidates for future initiatives [published online ahead of print, 2019 Dec 30]. *Prog Retin Eye Res.* 2019;100827. doi:10.1016/j.preteyeres.2019.100827

<sup>2</sup>What is a Clinical Trial Identifiers (NCT number)? The unique identification code given to each clinical study upon registration at ClinicalTrials.gov. The format is "NCT" followed by an 8-digit number (for example, NCT00000419). This can be used as search criteria for a specific clinical trial.

<sup>3</sup>**FDA Classification of Clinical Trial Phases: Source: <https://clinicaltrials.gov/ct2/about-studies/glossary>**

Phase: The stage of a clinical trial studying a drug or biological product, based on definitions developed by the U.S. Food and Drug Administration (FDA). The phase is based on the study's objective, the number of participants, and other characteristics. There are five phases: Early Phase 1 (formerly listed as Phase 0), Phase 1, Phase 2, Phase 3, and Phase 4. Not Applicable is used to describe trials without FDA-defined phases, including trials of devices or behavioral interventions.

Phase 1: A phase of research to describe clinical trials that focus on the safety of a drug. They are usually conducted with healthy volunteers, and the goal is to determine the drug's most frequent and serious adverse events and, often, how the drug is broken down and excreted by the body. These trials usually involve a small number of participants.

Phase 2: A phase of research to describe clinical trials that gather preliminary data on whether a drug works in people who have a certain condition/disease (that is, the drug's effectiveness). For example, participants receiving the drug may be compared to similar participants receiving a different treatment, usually an inactive substance (called a placebo) or a different drug. Safety continues to be evaluated, and short-term adverse events are studied.

Phase 3: A phase of research to describe clinical trials that gather more information about a drug's safety and effectiveness by studying different populations and different dosages and by using the drug in combination with other drugs. These studies typically involve more participants.

Phase 4: A phase of research to describe clinical trials occurring after FDA has approved a drug for marketing. They include postmarket requirement and commitment studies that are required of or agreed to by the study sponsor. These trials gather additional information about a drug's safety, efficacy, or optimal use.

Phase Not Applicable: Describes trials without FDA-defined phases, including trials of devices or behavioral interventions.  
Placebo

**Patient Questions to Ask: <https://clinicaltrials.gov/ct2/about-studies/learn#Questions>**

Anyone interested in participating in a clinical study should know as much as possible about the study and feel comfortable asking the research team questions about the study, the related procedures, and any expenses. The following questions may be helpful during such a discussion. Answers to some of these questions are provided in the informed consent document. Many of the questions are specific to clinical trials, but some also apply to observational studies.

- What is being studied?
- Why do researchers believe the intervention being tested might be effective? Why might it not be? Has it been tested before?
- What are the possible interventions that I might receive during the trial?
- How will it be determined which interventions I receive (for example, by chance)?
- Who will know which intervention I receive during the trial? Will I know? Will members of the research team know?
- How do the possible risks, side effects, and benefits of this trial compare with those of my current treatment?
- What will I have to do?
- What tests and procedures are involved?
- How often will I have to visit the hospital or clinic?
- Will hospitalization be required?
- How long will the study last?
- Who will pay for my participation?
- Will I be reimbursed for other expenses?
- What type of long-term follow-up care is part of this trial?
- If I benefit from the intervention, will I be allowed to continue receiving it after the trial ends?
- Will results of the study be provided to me?
- Who will oversee my medical care while I am participating in the trial?
- What are my options if I am injured during the study?

**Additional resources for patients:**

- Additional Information to Provide Patient:
- <https://clinicaltrials.gov/ct2/help/for-patient>
- <https://clinicaltrials.gov/ct2/about-studies/learn>
- Phases of Interventional Studies as defined by FDA (Source: <https://clinicaltrials.gov/ct2/about-studies/glossary>)