### Selected Observational Studies and Clinical Treatment Trials Recruiting Children with Genetic Eye Disorders

<table>
<thead>
<tr>
<th>Condition(s) / Clinical Trials Identifier</th>
<th>Gene (if applicable)</th>
<th>Intervention (if applicable)</th>
<th>Phase</th>
<th>Status as of date accessed (7/17/20)</th>
<th>Age Eligibility</th>
<th>Location(s)</th>
<th>Contact Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Achromatopsia/ NCT03758404*</td>
<td>CNGA3</td>
<td>Gene replacement therapy (AAV-CNGA3)</td>
<td>I/II</td>
<td>Recruiting <em>(Moorfields)</em></td>
<td>3-15 years</td>
<td>London, UK</td>
<td>Moorfields/London, UK James Bainbridge, Prof (PI) <a href="mailto:james.bainbridge1@nhs.net">james.bainbridge1@nhs.net</a> Michel Michaelides, Prof (sub-PI) <a href="mailto:michel.michaelides@nhs.net">michel.michaelides@nhs.net</a> 2. U of M/Ann Arbor, US Contact: Adrienne Chen 734-232-9167 (PI: Cagri Besirli, MD) <a href="mailto:KelloggResearch@med.umich.edu">KelloggResearch@med.umich.edu</a></td>
</tr>
<tr>
<td>Cerebrotendinous</td>
<td>CYP27A1</td>
<td>Prevalence</td>
<td>N/A</td>
<td>Recruiting:</td>
<td>Initial</td>
<td>45 locations in the US -</td>
<td>Retrophin Medical Information</td>
</tr>
<tr>
<td>Study Title</td>
<td>Study Details</td>
<td>Start Date</td>
<td>Last Updated</td>
<td>Eligibility</td>
<td>Contact Information</td>
<td></td>
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<tr>
<td><strong>Xanthomatosis (CTX)</strong>&lt;br&gt;NCT02638220</td>
<td>study in early-onset cataracts</td>
<td>Start date 12/2015&lt;br&gt;Last updated: 11/9/18</td>
<td>diagnosis of bilateral idiopathic cataracts between 2 and 21 years</td>
<td>see clinicaltrials.gov (<a href="https://clinicaltrials.gov/ct2/show/NCT02638220?term=NCT02638220&amp;draw=2&amp;rank=1">https://clinicaltrials.gov/ct2/show/NCT02638220?term=NCT02638220&amp;draw=2&amp;rank=1</a>)</td>
<td>1-877-659-5518 <a href="mailto:medinfo@retrophin.com">medinfo@retrophin.com</a></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Disorders with obesity: Pro-opiomelanocortin (POMC) Deficiency Obesity (Heterozygous or Epigenetic), Leptin Receptor Deficiency Obesity, Bardet-Biedl Syndrome, Alstrom Syndrome, Smith-Magenis Syndrome</strong>&lt;br&gt;NCT03013543</td>
<td>Obesity associated with rare disorders as listed&lt;br&gt;Setmelanotide SC injection</td>
<td>2/3</td>
<td>Recruiting: Start date: 1/6/17&lt;br&gt;Last updated: 5/27/20</td>
<td>≥ 12 years</td>
<td>27 sites (see complete list by searching: NCT03013543)</td>
<td>Olga Ohayon&lt;br&gt;<a href="mailto:oohayon@rhythmtx.com">oohayon@rhythmtx.com</a></td>
<td></td>
</tr>
<tr>
<td><strong>Leber Congenital Amaurosis 10 (ILLUMINATE)/NCT03913143</strong></td>
<td>CEP290 c.2991+1655A&gt;G Mutation (IVS26)</td>
<td>1/2</td>
<td>Recruiting: Start date: 4/12/19&lt;br&gt;Last updated: 3/11/20</td>
<td>≥ 8 years</td>
<td>US: Miami, FL&lt;br&gt;Iowa City, IA&lt;br&gt;NY, NY&lt;br&gt;Portland, OR&lt;br&gt;Philadelphia, PA&lt;br&gt;Houston, TX&lt;br&gt;Several international sites</td>
<td>+31 (0)88 1667000 ProQR Clinical Trials Manager <a href="mailto:info@proqr.com">info@proqr.com</a></td>
<td></td>
</tr>
<tr>
<td><strong>Leber Congenital Amaurosis 10/NCT03872479</strong></td>
<td>CEP290 c.2991+1655A&gt;G Mutation (IVS26)</td>
<td>1/2</td>
<td>Recruiting Start Date: Last updated:</td>
<td>≥ 3 years</td>
<td>Miami, FL&lt;br&gt;Boston, MS&lt;br&gt;Ann Arbor, MI&lt;br&gt;Portland, OR</td>
<td>Clinical Trials Registry Team 1-877-277-8566&lt;br&gt;<a href="mailto:IR-CTRegistration@allergan.com">IR-CTRegistration@allergan.com</a></td>
<td></td>
</tr>
<tr>
<td><strong>Leber Hereditary Optic Neuropathy (LHON)/NCT02161380</strong></td>
<td>G11778A (mutation-specific&lt;sup&gt;4&lt;/sup&gt;)&lt;br&gt;AAV2-P1ND4v2</td>
<td>1</td>
<td>Recruiting: Start date: 6/11/14&lt;br&gt;Last updated: 10/13/19</td>
<td>≥ 15 years</td>
<td>Miami, FL</td>
<td>John Guy, MD&lt;br&gt;<a href="mailto:jguy@med.miami.gov">jguy@med.miami.gov</a>&lt;br&gt;302-326-6036</td>
<td></td>
</tr>
<tr>
<td><strong>Leber Hereditary LHON but</strong></td>
<td>Observational</td>
<td>N/A</td>
<td>Recruiting: Start date: 9/27/17</td>
<td>All ages</td>
<td>Recruiting: 1. Doheny Eye Center,</td>
<td>Contact: Barrett Katz, MD</td>
<td></td>
</tr>
<tr>
<td>Study Title</td>
<td>Enrollment Criteria</td>
<td>Status</td>
<td>Duration</td>
<td>Location</td>
<td>Contact Information</td>
<td></td>
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</tbody>
</table>
| Optic Neuropathy (LHON)/NCT03295071                                       | aiming for 75% with 11778/ND4 mutation                                                | Last updated: 4/24/2019 | N/A                       | Pasadena, CA  
2. Emory University, Atlanta, GA  
3. MEEI, Boston, MA  
4. Wills Eye, Phil PA  
5. Alkek Eye, Houston, TX | +1 646 831 3799 bkatz@gensight-biologics.com  
Contact: Magali Taiel, MD +33 (0)7 62 89 12 52 mtaiel@gensight-biologics.com |
| Neuronal Ceroid Lipofuscinosis (Juvenile Batten Disease)/NCT03307304       | CLN3                                                                                 | Recruiting: Start date:  
10/11/2017  
Last updated:  
6/1/20   | 0-100 years                                                                 | Recruiting: Bethesda, MD, NIH | N Dang Do, M.D  
301-496-8849  
an.dangdo@nih.gov |
| Neuronal Ceroid Lipofuscinoses (CLN2) Spinocerebellar Ataxia, autosomal recessive 7/ NCT04098211 | CLN2 (atypical TTP1 deficiency)                                                      | Recruiting: Start Date:  
11/1/19  
Last updated:  
2/5/20   | ≥ 4 years                                                                | US, Orange, CA Children’s Hospital of Orange County | Raymond Wang, MD  
7145093344  
rawang@choc.org |
| Retinal Disease (any) Virtual Reality Mobility Assessment of Functional Vision in Retinal Disease | Any – retinal dystrophy affecting vision                                          | Recruiting: Start date:  
2/28/20  
Last updated:  
6/11/20   | ≥ 5 years with retinal dystrophy affecting vision                              | US, Bethesda, MD, NIH  
Australia, Sydney, Sydney Eye Hospital | US: Daniel W. Claus, R.N.; (301) 496-9058,  
daniel.claus@nih.gov  
Australia: John Grigg, M.D.  
61293827300  
john.grigg@sydney.edu.au |
| Retinitis Pigmentosa NCT03845218                                          | Any                                                                                  | Recruiting: Start date:  
2/19/19  
Last updated:  
2/5/20   | > 12 years                                                               | US, Bethesda, MD, NIH | Cathy Kangale-Whitney, R.N.  
(301) 402-4174  
cathy.kangale-whitney@nih.gov  
P: Catherine A Cukras, M.D |
| Smith-Lemli-Opitz-Syndrome/ NCT01773278                                   | DHC R7                                                                               | Recruiting: Start date:  
1/23/13  
Last updated:  
10/3/19   | Up to 65 years                                                             | Aurora, CO (Children’s Hospital Colorado) | Ellen Elias, MD  
720-777-5401  
Ellen.elias@childrenscolorado.org |
<table>
<thead>
<tr>
<th>Disease / Trial Identification / Gene</th>
<th>Gene</th>
<th>Medication / Procedure</th>
<th>Phase</th>
<th>Status</th>
<th>Start Date</th>
<th>End Date</th>
<th>Age</th>
<th>Sites</th>
<th>Contact Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Smith-Lemli-Opitz-Syndrome / NCT03720990</td>
<td><em>DHCR7</em></td>
<td>Cholic acid</td>
<td>1/2</td>
<td>Not yet recruiting</td>
<td>Last update: 3/5/20</td>
<td>2-25 years</td>
<td></td>
<td></td>
<td>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></td>
</tr>
<tr>
<td>Stargardt Disease / NCT02402660</td>
<td><em>ABCA4</em></td>
<td>Oral medication: ALK-001&lt;sup&gt;5&lt;/sup&gt;</td>
<td>2</td>
<td>Recruiting</td>
<td>Start date: 3/30/15 Last updated: 5/5/20</td>
<td>8-70 years</td>
<td>US: Los Angeles, CA Gainesville, FL Miami, FL Baltimore, MD NY, NY Salt Lake City, UT Milwaukee, WI</td>
<td>Leonide Saad, PhD 800-287-2755 <a href="mailto:trials@alkeus.com">trials@alkeus.com</a></td>
<td></td>
</tr>
<tr>
<td>Stargardt Disease / NCT03772665 [SEASTar]</td>
<td><em>ABCA4</em></td>
<td>Oral medication: Emixustat</td>
<td>3</td>
<td>Active, not recruiting</td>
<td>Start date: 12/11/18 Last updated: 5/7/20</td>
<td>≥ 16 years</td>
<td>US: San Francisco, CA Beverly Hills, CA Atlanta, GA Baltimore, MD Ann Arbor, MI Rochester, MN Durham, NC Portland, OR Dallas, TX Salt Lake City, UT Milwaukee, WI International sites (See clinical trials.gov)</td>
<td><a href="mailto:ClinicalTrials@acucela.com">ClinicalTrials@acucela.com</a> (206) 805 8310</td>
<td></td>
</tr>
<tr>
<td>X-Linked RP / NCT03316560</td>
<td><em>RPGR</em></td>
<td>Gene replacement: rAAV2tYF-GRK1-RPGR</td>
<td>1/2</td>
<td>Recruiting</td>
<td>Start date: 10/20/2017 Last updated: 12/11/19</td>
<td>≥ 6 years (male gender)</td>
<td>US: Recruiting: NY, NY Durham, NC Cincinnati, OH Portland, OR Dallas, TX Not yet recruiting: Golden, CO Boston, MA Cleveland, OH Philadelphia, PA</td>
<td>Jill Dolgin, PharmD 833-770-2862 <a href="mailto:advocacy@agtc.com">advocacy@agtc.com</a></td>
<td></td>
</tr>
<tr>
<td>X-Linked RP /</td>
<td><em>RPGR</em></td>
<td>Gene</td>
<td>2/3</td>
<td>Recruiting: Part I &gt; 18</td>
<td>UK (Several)</td>
<td></td>
<td></td>
<td>NightstaRx Ltd, a Biogen Company</td>
<td></td>
</tr>
</tbody>
</table>
### Abbreviation:
RP = retinitis pigmentosa; AAV = adeno-associated virus vector

### Footnotes:

1 Please refer to ClinicalTrials.gov for more information.

There are many more gene therapy trials for patients 18 years and older. (Examples include Stargardt (gene therapy, Zimura, Plaquenil). 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 1-800-683-5555


2 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.

Abbreviations: RP: retinitis pigmentosa

3 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

Patients must have a specific mutation in CEP290 to qualify (This is different than gene replacement therapy of RPE65, where any bi-allelic mutations would allow for potential treatment).

ALK-001: The investigational drug is a chemically-modified vitamin A designed as a replacement of vitamin A to prevent the formation of toxic vitamin A dimers in the eye. Trial participants will receive either ALK-001 or placebo.

References CNGA3 Gene Therapy (Hot off Press!):

Trials registration: ClinicalTrials.gov Identifier: NCT02610582.

PDF Text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7193523/?report=reader


Preliminary Results:

PDF Full Text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6753501/


References:


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 Intervventional Studies as defined by FDA (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
An inactive substance or treatment that looks the same as, and is given in the same way as, an active drug or intervention/treatment being studied.

Patient Questions to Ask:
Questions to Ask [https://clinicaltrials.gov/ct2/about-studies/learn#Questions](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 effective? 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?