An Educational Webinar on Batten Disease

Wednesday, December 12, 2018
Our Purpose, Our Mission:

The purpose of Global Genes is to *Connect, Empower and Inspire* the rare disease community.
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Clinical Overview of Batten disease

Presented by: Emily de los Reyes, MD
Batten’s disease

Objectives
To discuss diagnosis and current classification
To review current treatment and future modes of therapy for Batten’s disease
Batten’s disease

Grants and Consultancy

Biomarin
Charlotte and Gwennyth Gray foundation
Amicus
Batten’s disease

- Most common clinical cause of dementia in children
  - Previously normal children may lose their ability to talk or walk, cognitive or learning disability
- Lysosomal disease
- Autosomal recessive (two family members have the genetic abnormality)
- 100 molecular mutations with 14 presumed genes
- Accumulation of lipopigments called lipofuscin
Batten’s disease

- Language delay or language regression
- Blindness, typically the presenting symptom in CLN3
- Epilepsy
- Motor regression, balance issues
- Cognitive decline
- Sleep disorders
Lysosome

If the mitochondria is the powerhouse

Lysosome

- Degrades and takes up protein
- Roles in macroautophagy and microautophagy
- Lysosome associated membrane proteins
  - Proteins facilitate the uptake and digestion.
Lysosomal disease

- Loss of normal function of specific lysosomal acid hydrolases, which act to degrade large complex substrates that have been targeted for degradation after endocytosis or autophagy.
- Accumulation of partially degraded substrate affects the architecture and function of cells, tissues, and organs. In some cases the accumulate substrate may be cytotoxic.
There Are 14 Types Of NCL And 13 Known Genes-classification And Characteristics Of NCLs

<table>
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<th>Disease</th>
<th>Onset &amp; clinical phenotype</th>
<th>Gene</th>
<th>Protein</th>
</tr>
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<tr>
<td>CLN1</td>
<td>Infantile classic, late-infantile, juvenile, adult</td>
<td>CLN1 (PPT1)</td>
<td>Palmitoyl protein thioesterase 1</td>
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<tr>
<td>CLN2</td>
<td>Classic late-infantile phenotype; atypical phenotypes: infantile, juvenile, protracted; SCAR7</td>
<td>CLN2 (TPP1)</td>
<td>Tripeptidyl peptidase 1</td>
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<tr>
<td>CLN3</td>
<td>Juvenile classic</td>
<td>CLN3</td>
<td>Transmembrane protein</td>
</tr>
<tr>
<td>CLN4</td>
<td>Adult (autosomal dominant)</td>
<td>CLN4 (DNAJC5)</td>
<td>Soluble cysteine string protein</td>
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<tr>
<td>CLN5</td>
<td>Late-infantile variant, juvenile, adult</td>
<td>CLN5</td>
<td>Soluble lysosomal protein</td>
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<tr>
<td>CLN6</td>
<td>Late-infantile variant, adult (Kufs type A)</td>
<td>CLN6</td>
<td>Transmembrane protein</td>
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<tr>
<td>CLN7</td>
<td>Late-infantile variant</td>
<td>CLN7</td>
<td>Transmembrane protein</td>
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<td>CLN8</td>
<td>Late-infantile variant</td>
<td>CLN8</td>
<td>Transmembrane protein</td>
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<tr>
<td>CLN9</td>
<td>Juvenile variant</td>
<td>-</td>
<td>-</td>
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<tr>
<td>CLN10</td>
<td>Congenital class, late-infantile, juvenile, adult</td>
<td>CLN10 (CTSD)</td>
<td>Cathepsin D</td>
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<tr>
<td>CLN11</td>
<td>Adult</td>
<td>CLN11 (GRN)</td>
<td>Progranulin</td>
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<td>CLN12</td>
<td>Juvenile</td>
<td>CLN12 (ATP13A2)</td>
<td>ATPase</td>
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<tr>
<td>CLN13</td>
<td>Adult (Kufs type B)</td>
<td>CLN13 (CTSF)</td>
<td>Cathepsin F</td>
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<tr>
<td>CLN14</td>
<td>Infantile</td>
<td>CLN14 (KCTD7)</td>
<td>Potassium channel protein</td>
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Neuronal Ceroid Lipofuscinosis

- **Most common types**
  - CLN1 (Infantile NCL), Soluble enzyme,
    - PPT, palmitoyl protein thioesterase
  - CLN2 (Late Infantile NCL), Soluble enzyme,
    - TPP, tripeptidyl peptidase
  - CLN3 (Juvenile NCL)
    - Transmembrane protein
In The Old Days……

- Age of Onset
- Clinical Phenotype
- Ultrastructural Characteristics
  - Granular Osmiophilic Deposits (GROD)
  - Curvilinear bodies
  - Fingerprint bodies
The Multidisciplinary Team Is Essential For The Management.

CLN2 Patient & Family

- Seizures
- Vision
- Mobility
- Communication
- Feeding
- Speech/feeding therapist
- Palliative care team
- Physical therapist
- Pediatric neurologist
- Pediatrician
- Ophthalmologist
- Genetic counsellor
- Irritability & Pain
- Bladder & bowels
- Sleep
- Community nursing team
- Gastroenterologist
- Metabolic specialist
- Occupational therapist
- Pediatrician
- Community nursing team
- Gastroenterologist
- Metabolic specialist
- Occupational therapist

Treatment

Symptomatic treatment

• Seizures
  • Myoclonus, Partial seizures, Generalized tonic clonic
  • Zonisamide, Valproic acid, Clobazam

• Spasticity

• Therapies: Physical, Occupational and Speech
Medical issues

- Swallowing problems
  - Speech/Swallow therapies
  - Thickening feeds
  - G tube, dependent of families.

- Respiratory
  - Potentially treatable, Flu
  - Tendency to worsen disease
Symptomatic treatment

Immunizations
No reason to hold immunizations, if families wish, we can space it out

Nutrition
Weight gain, good nutrition very important
Adequate hydration
Nutritional reserves
Goals Of Care Evolve As The Disease Progresses

**Quality of Life**
- **Maintenance of function** to support QoL as disease progresses
- **Maintenance of QoL** as disease progresses

**Medical management**
- **Symptom management**
- **Ongoing symptom management**
  - Prevention and management of complications (e.g., respiratory, immobility)

**Family support**
- **Psychosocial support**
  - Transition support
- **These activities become more important**

**End-of-life care**
- **Early discussions of what the future looks like**
- **Implement palliative care concept** with multidisciplinary support for the patient, the parents and the family

Challenges of Delivering Therapies to the brain and spinal cord

CNS presents certain challenges
- mere size and distribution
- diversity of cell types
- blood brain barrier

Most drugs/therapies do not target the Brain
Batten’s treatment

Enzyme replacement therapy, approved in 2017 for the treatment of CLN2
Intracerebral gene therapy for CLN2
Clinical trials for CLN3 and CLN6 disease
BMT
Stem cell
Neuroprotective agents, Seizure control
BBDF’s Strategy

Other Neurological Disease Groups

BBDF

Other Batten Families & Foundations
Hugs for Hudson
AT BEYOND BATTEN DISEASE FOUNDATION

www.willherndon.org

Directed Funds

BIND
BATTEN

CHASE "JASPER" PETERSON FUND AT BEYOND BATTEN DISEASE FOUNDATION

Tyler’s Mission
Go Tyler! Beat Batten!

RARE Sisters
Bonding through Batten
Partner and Leverage
BBDF has matched every donation dollar for dollar by leveraging partnerships and co-funding resulting in $26 million in research over the last 10 years.
Funded Research

2008 - 2016
$20 Million
40 Research Projects

2016
Drug Discovery

2017 - 2018
$6 Million

FUND the treatment
PREPARE Support
research for a cure
Research Program Highlights

• Built a rare disease genetic test
• Funded research initiatives for:
  - Discovery of drug targets and medicines
  - Screened thousands of drugs for potential to treat juvenile Batten disease
  - Created Batten specific tools: biomarkers, animal models, antibodies
  - Developed and commercialized Batten iPS stem cell line
  - Advancing multiple approaches to treating CLN3 – exon skipping, gene therapy
• Support for international patient registries and natural history studies
• Funding initiative for biomarkers and clinical endpoints
Medical Breakthrough:

Treatment now in sight!

• These efforts have resulted in a treatment which slows the progression in Batten disease models.

• This treatment has undergone an expensive validation and development process to prepare for a clinical trial.

• Successful FDA PreIND meeting in May 2018. Clinical trial in 2019.
THE FDA Approval Process

PRECLINICAL

Lab Studies

192 Studies Pre IND

FDA Pathway for BBDF 101

PHASE I

Pre IND

• Is it safe?
• Is it toxic?

PHASE II

Juvenile Toxicity

PHASE III

Children under 18

PHASE III

What are the side effects in the larger population?
• Efficacy
• Optimal dosing

FDA NEW DRUG APPROVAL

• Application submitted
• Application reviewed
• Application approved
• Available to the public

FDA Approval

ODD and RDPRV

EXPANDED USE FOR ADULTS (18 AND OVER)
TODAY: Get a treatment to the kids who need it most to slow disease progression

TOMORROW: Buy time while we continue to support research for a cure

FOREVER: Leave a legacy that lives on for kids and families affected by Batten and many other diseases
Noreen Murphy
Patient and Family Education Coordinator
Our long-term vision is a world without Batten disease. Our mission is to support Batten families, fund and facilitate research for treatments and cures, and advocate for action.
How BDSRA Supports Families

Yearly 3-day family conference (~480 attendees)
Private Facebook group for parents and caregivers (~1,000 people)
Monthly Newsletter
Expert Facebook chats
Staff available for calls and connect families with experts in any topic
Connect families for 1 on 1 communication
Represent the patient voice to industry

www.bdsra.org
## Emerging Research in Batten Disease

<table>
<thead>
<tr>
<th>Type of Batten</th>
<th>CLN1</th>
<th>CLN2</th>
<th>CLN3</th>
<th>CLN5</th>
<th>CLN6</th>
<th>CLN7</th>
<th>CLN8</th>
<th>CLN10</th>
<th>ANCL</th>
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<td>BioMarin</td>
<td>Abeona Therapeutics, Inc.</td>
<td>UT Southwestern</td>
<td>Amicus Therapeutics</td>
<td>Foundation for Batten Hope</td>
<td>Amicus Therapeutics</td>
<td>Circumvent Pharmaceuticals</td>
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<td>Circumvent Pharmaceuticals</td>
<td>Polaryx Therapeutics, Inc.</td>
<td>Amicus Therapeutics</td>
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<td>Polaryx Therapeutics, Inc.</td>
<td>REGENXBIO</td>
<td>Beyond Batten Disease Foundation</td>
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<td>Spark Therapeutics</td>
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www.bdsra.org
Company Name: Abeona Therapeutics, Inc.
Type of Batten: CLN1 disease and CLN3 disease
Type of program: Gene Therapy
Program Information:
ABO-202(scAAV-CLN1) for CLN1 disease
One-time administration of the correct version of the CLN1 gene using Adeno-associated virus (AAV9)
Targeting initiation of a Phase 1/2 clinical trial (primary objective of evaluating safety) in 2019
Orphan Drug Designation (FDA)
Orphan Drug Designation (EMA)
Rare Pediatric Disease Designation (FDA)
ABO-201 (scAAV-CLN3) for CLN3 disease
One-time administration of the correct version of the CLN3 gene using Adeno-associated virus (AAV9)
Targeting initiation of a Phase 1/2 clinical trial (primary objective of evaluating safety) in 2019
Orphan Drug Designation (FDA)
Orphan Drug Designation (EMA)

Contact:
Judy Doyle, Patient Engagement Specialist
jdoyle@abeonatherapeutics.com
Michelle Berg, VP Patient Affairs and Community Engagement
mberg@abeonatherapeutics.com
Company Name: **Amicus Therapeutics**
Type of Batten: **CLN3, CLN6 and CLN8**
Type of program: Gene Therapy
Website: [https://www.amicusrx.com/programs-pipeline/](https://www.amicusrx.com/programs-pipeline/)
Contact: [patientadvocacy@amicusrx.com](mailto:patientadvocacy@amicusrx.com)
Company Name: **Foundation for Batten Hope**
Type of Batten: **CLN7**
Type of program: Gene Therapy

Recruiting for Natural History Study:
- Clinical Research Program Manager:
  - Sam Hughes, (Samuel.Hughes@utsouthwestern.edu)
- Clinical Trial Research Medical Director:
  - Dr. Saima Kayani, USTW (Saima.Kayani@utsouthwestern.edu)

Current Status: Toxicology study, viral vector development and initial animal study in process.

Next Milestone: Recruiting for natural history study for CLN7 and CLN5 beginning in 2019. Please contact Dr. Kayani if interested.

For patient advocacy questions, please contact Gina Hann (ginahann@gmail.com)

[https://battenhope.org/batten-hope-video/](https://battenhope.org/batten-hope-video/)
Circumvent Pharmaceuticals Overview

- Company name: Circumvent Pharmaceuticals
- We are working in: CLN1, CLN3, CLN10, ANCL
- We are developing: Small molecule thioesterase mimetics
- www.circumventpharmaceuticals.com
- Update: We completed key non-GLP pharmacokinetic and toxicology preclinical studies validating our Development Candidate (compound for advancing towards clinical development) and are developing a clinical formulation for IND-enabling studies
- Contact info: misty@circumventpharmaceuticals.com
Indications: CLN2 and CLN3

Type of therapy: Small molecule molecule therapy which is safe, oral, and easily portable

Characteristics of lead compound (PLX-200)
- Gemfibrozil, an FDA-approved lipid lowering agent
- Increases lysosome biogenesis, anti-inflammatory mediators, remyelination, and neurotrophins
- Reduces inflammation, glial activation, lipofuscin in the brain, and apoptosis
- Increases survival in CLN2/Cln3 murine disease models

IND filing: Expected in 2Q 2019

Clinical trials: Expected in 3Q 2019 in the US

Key progress:
- Completed rat juvenile toxicity study and formulation development,
- Orphan drug designation for all subtypes of neuronal ceroid lipofuscinosis granted by FDA and EMA

Contact: Hahn-Jun Lee, M.Sc., Ph.D., President/CEO, Polaryx Therapeutics, Inc
140 E. Ridgewood Avenue, Suite# 415, South Tower, Paramus, NJ 07652 US
O: +1-201-940-7236, F: +1-201-940-7218, hahnjun7@polaryx.com
**REGENXBIO** is a leading clinical-stage biotechnology company seeking to improve lives through the curative potential of gene therapy based on its proprietary NAV® Technology Platform, which consists of exclusive rights to AAV7, AAV8, AAV9, AAVrh10 and over 100 other novel AAV vectors.

**REGENXBIO** is developing a gene therapy, **RGX-181**, for the treatment of CLN2 disease, a form of Batten disease.

- **RGX-181** is designed to use **REGENXBIO**’s AAV9 vector to deliver the TPP1 gene directly to the central nervous system (CNS) to induce sustained levels of TPP1, the enzyme deficient in children with CLN2 disease.

- The program is in the preclinical stage of development and the company continues to make progress to submit an Investigational New Drug Application (IND) to the US Food and Drug Administration (FDA) in 2019 to enable initiation of a global clinical trial for CLN2.

- RGX-181 was granted Orphan Drug Designation by the FDA.

For more information on **REGENXBIO** and the CLN2 program, visit [www.regenxbio.com](http://www.regenxbio.com), or contact [patientadvocacy@regenxbio.com](mailto:patientadvocacy@regenxbio.com).
Spark Therapeutics is developing SPK-TPP1, an investigational gene therapy that has demonstrated compelling preclinical proof-of-concept in one naturally occurring preclinical model of TPP1 deficiency, a form of Batten disease. Batten disease is a fatal neurological disorder involving mutations of the TPP1 gene that begins in early childhood. We have received orphan product designation from the U.S. FDA for SPK-TPP1 for the treatment of CLN2 disease (neuronal ceroid lipofuscinosis [NCL]) caused by TPP1 deficiency.

Contact: patients@sparktx.com
Update on AAV9 gene therapy programs for Batten Disease

Kathrin Meyer
Principal Investigator at the Center for Gene Therapy
Assistant Professor at The Ohio State University
Nationwide Children's Hospital, Columbus Ohio
Disclosure Information

I have the following financial relationships to disclose:
  • Consultant for Amicus Therapeutics, Inc.

I will discuss the following off-label use and/or investigational use in my presentation:
  • Preclinical, proof of concept data from studies for the treatment of patients with Batten disease
Validated Gene Therapy Platform

Portfolio is Based on a Validated Gene Therapy Approach Across Multiple CNS Diseases

Clinically validated AAV gene therapy approach

- Nationwide Children’s Hospital Center for Gene Therapy (NCH)
- Intrathecal delivery with robust expression throughout CNS

Preclinical safety and efficacy studies replicated across multiple diseases at NCH

- SMA
- Rett Syndrome
- ALS
- CLN6
- CLN3

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
Single AAV9-CLN6 Administration Results in Reduction of Autofluorescent Substrate Material Throughout the Brain

Autofluorescent Storage Material Accumulation

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
CLN6: Preclinical Mouse Data – Somatosensory Glial Activation

Single AAV9-CLN6 Administration Results in Reduction of Glial Activation

Astrocyte Activation: Month 18

Microglial Activation: Month 18
CLN6: Preclinical Mouse Data
Motor Performance and Cognitive Behavior

Single AAV9-CLN6 Administration Improves Motor Performance & Cognitive Behavior Out to Month 24

Motor Performance

Water Maze Analysis

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy; Data on file
CLN6: Preclinical Mouse Data - Survival

Single AAV9-CLN6 Administration Significantly Extends Median Survival

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
CLN6 Expression in NHP Safety Study

Demonstrated Safety and Meaningful Transduction and CLN6 Expression Throughout the Brain in NHPs

Western Blot on various brain regions of AAV9-CLN6 injected juvenile NHPs

Source: Meyer 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, From mouse to human –Translating intrathecal gene therapy for NCLs; Data on file
Preclinical Proof of Concept Data in CLN3 Batten Disease
CLN3: Preclinical Mouse Data – Autofluorescent Substrate Material

Single AAV9-CLN3 Administration Results in Reduction of Autofluorescent Substrate Material Throughout the Brain

Autofluorescent Storage Material Accumulation

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
CLN3: Preclinical Mouse Data – Somatosensory Glial Activation

Single AAV9-CLN3 Administration Results in Reduction of Glial Activation

Astrocyte Activation: Month 8

Microglial Activation: Month 8

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
Single AAV9-CLN3 Administration Improves Motor Performance and Cognitive Behavior at Month 16

Motor Performance

Water Maze Analysis

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
Effect of AAV9-CLN3 On Survival Cannot Yet Be Determined Given Minimal Phenotype in Mouse Model

Source: Likhite 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, IND-enabling Preclinical Studies for Batten Disease Gene Therapy
CLN3 Expression in NHP Safety Study

Demonstrated Safety and Meaningful Transduction and Expression Throughout the Brain in NHPs

Reverse transcriptase qPCR on various brain regions of AAV9-CLN3 injected juvenile NHPs

Fold Change (normalized to NCL3 levels in Lumbar SC 4/7)

- Lumbar SC 4/7
- Temporal lobe
- Occipital lobe
- Cerebellum
- Thalamus
-Amygdala
- Corpus Callosum
- Pen-White Matter
- Cervical SC 4-7
- Thoracic SC 7-13
- Lumbar SC 1-4
- Retina
- Optic nerve

Note: CLN3 Western blot - data were not assessable

Source: Meyer 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, From mouse to human –Translating intrathecal gene therapy for NCLs
Preclinical Proof of Concept Data in CLN8 Batten Disease
CLN8: Preclinical Mouse Data – Autofluorescent Storage Material

Single AAV9-CLN8 Administration Results in Reduction of Autofluorescent Substrate Material Throughout the Brain

Source: Johnson 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, Testing the safety and efficacy of CLN8 gene therapy in the Cln8mnd mouse model
CLN8: Preclinical Mouse Data - Somatosensory Glial Activation

Single AAV9-CLN8 Administration Results in Reduction of Glial Activation

**Astrocyte Activation: Month 8**

**Microglial Activation: Month 8**

Source: Johnson 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, Testing the safety and efficacy of CLN8 gene therapy in the Cln8mnd mouse model
CLN8: Preclinical Mouse Data - Motor Performance and Cognitive Behavior

Single AAV9-CLN8 Administration Improves
Motor Performance & Cognitive Behavior Out to Month 10

Motor Performance

Water Maze Analysis

Source: Johnson 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, Testing the safety and efficacy of CLN8 gene therapy in the Cln8"mnd" mouse model
CLN8: Preclinical Mouse Data - Survival

Single AAV9-CLN8 Administration Significantly Extends Median Survival

Source: Johnson 2018, 16th International Conference on Neuronal Ceroid Lipofuscinoses, Testing the safety and efficacy of CLN8 gene therapy in the Cln8mnd mouse model
Efficacy Data: Matched Sibling Case Report

Encouraging Interim Efficacy Data in First Two Patients Treated with Gene Therapy with Two Years of Follow-up

- Two siblings (same genotype) treated with gene therapy at ages 2.8 and 5.3 years, respectively
- Two years post treatment, Hamburg motor and language scores indicate no disease progression in the younger sibling
- Disease progression in older sibling has shown evidence of stabilization

Source: Data on file
CLN6 Clinical Summary

Jay Barth, M.D.
CLN6: Clinical Data Summary

Encouraging Safety and Efficacy Data from an Ongoing Single-arm Phase 1/2 study

- Single-arm study with all patients receiving gene therapy
  - Single intrathecal administration
- Ten patients currently treated; additional patients in screening
- Generally well-tolerated
- Encouraging preliminary efficacy data
- Additional data to be presented in 2019
CLN6: Clinical Study Safety Summary Interim Data

**Data from an Ongoing Single-arm Phase 1/2 Study Indicate Single AAV9-CLN6 Administration is Generally Well Tolerated**

- Ten patients currently treated with single intrathecal administration
  - Average follow-up duration: 12 months (range 1-24 months)
- Adverse events (n=94 events reported)
  - Majority of adverse events (AEs) were mild and unrelated to treatment
  - Five Grade 3 (severe) AEs (defined as medically significant) reported in 4 patients
  - No Grade 4 (life-threatening) or Grade 5 (death) AEs reported to date
- T-cell response and antibody elevations not associated clinical manifestations
  - No changes in treatment required
- Data Safety Monitoring Board (DSMB) has permitted study to proceed and enroll additional patients
Acknowledgments
Introduction to Amicus and Patient & Professional Advocacy

Jayne C. Gershkowitz
Chief Patient Advocate
Global Genes Batten Disease Rare Webinar
December 12, 2018
Amicus Mission

Amicus Therapeutics is committed to improving the lives of patients and families affected by rare and orphan diseases.

-- Corporate Belief Statement
Amicus History

2002
Amicus founded on pharmacological chaperone technology from Mt. Sinai School of Medicine

2006
First Fabry patient treated in Amicus clinical trials

2007
Amicus initial public offering (NASDAQ: FOLD)

2009
First Fabry patient in Ph. 3 study

2012
Proof of concept for chaperone-ERT combination (CHART technology)

2013
Callidus acquisition (Pompe ERT)

2014
Migalastat positive data from two Ph. 3 studies

2015
International HQ
MAA Submission
Pompe ERT in clinic
Scioderm acquisition (EB)

2016-2018
Galafold™ EU + US approvals
Biologics pipeline expansion (CDKL5)
Entering Ph. 3 Pompe studies
Celenex acquisition and UPenn collaborations advance gene therapy at Amicus with multiple new rare disease pipeline programs

Global Company
Building a World Class Organization

Global organization of 500+ employees dedicated to create, manufacture, test and deliver medicines for rare metabolic diseases

Global HQ
Cranbury, NJ

International HQ
United Kingdom

Canada

Netherlands

Italy

Germany

France

Spain

Japan

China (Mfg. Ops)

= headquarters
= offices
= presence
Amicus Vision: Delivering for Patients

To build a top-tier, fully integrated, global biotechnology company whose medicines treat 5,000+ patients by 2023

* all figures approximate

>350 Patients*

YE17

2023

5,000 Patients*
You are part of the Community of Us
At Amicus, we are driven by the journeys, experiences, involvement, relationships and outcomes of individuals and families living with rare disease.
At Amicus

- Knowing the PAOs and understanding the patient community
- Developing relationships externally and internally
- Working collaboratively with our partners and cross-functionally
- Mobilizing meaningful outcomes for patients, families, caregivers and healthcare professionals

Community of us
Educational Resources for Patient and Professional Communities

Disease awareness materials

- Fabry and Pompe infographics
- Signs and symptoms brochures
- CLN6 and CLN3 infographics *in progress*
- CDKL5 infographics *in progress*
Educational Resources for Patient and Professional Communities

General rare disease materials

- Adherence brochure
- How to read a clinical research publication
- Informed consent
- Drug development process
- Patient involvement in drug development
Tell Us

Rare Disease Day Events

Congresses & Meetings

Patient Advisory Boards, Focus Groups

HBD Lunch & Learn Events
Patient Advisory Boards

- Informed persons affected by a rare disease representing self and community at-large
- Nomination/application process
- Two-year commitment
- ~Two meetings/year; interim communications as necessary
- Broad viewpoints encouraged
- Backgrounds diverse by geography, age, disease experience
- Provide Amicus with insights into diagnostic odyssey, disease management; clinical research, meaningful endpoints, protocol design; broad community engagement, and more

- Inputs may assist in clinical trials, patient advocacy, business planning, patient services, educational programs
- Completes advisory triad: Medical Advisory Board, Scientific Advisory Board, Patient Advisory Board
P&PA Initiatives

Surveys capture patient, caregiver and HCP experience
Community Resources – Our Good Stuff Kit

One moment of appreciation each day is all it takes to develop a habit of positivity.

GoodStuff

Meet the CARTERS

Tucked away on the southern coast of England is the city of Portsmouth, where Margot Carter and her family live. Margot has a husband, 2 sons, and a daughter, and they know all been influenced by her optimistic outlook on life.

The Carters have more challenges than the average family. Margot lives with epilepsy, and 16-year-old Naomi lives with Fabry disease. Recognising the impact that chronic and rare disease can have, Margot looked for—and found—an opportunity to help herself and her family stay positive.

It began when Margot’s oldest son was very young, and Margot wanted to show him that there was “good stuff” in his life to be grateful for. Every night before he went to sleep, she would help him reflect on at least 10 positive things that had happened to him that day. Through this nightly practice, Margot witnessed the growing optimism in her son.

After Naomi was diagnosed with Fabry disease, Margot decided to expand the nightly practice with her son into a more active “visiting” experience that could be shared with her other children. This was the start of the “good stuff” jar.

Whenever something positive happens—big or small—the Carter family is encouraged to write notes about their achievements, thoughts, and activities to collect in a jar.

Initially, the Carters read the positive notes frequently, so that the family could see that even challenging days had good in them. Over time, reading aloud the “good stuff” became a less regular but still motivating event. Now they let the notes pile up throughout the whole year until the jar nearly overflows, and on every New Year’s Eve, the family gathers to read through each note. The occasion has become a special tradition for the Carters, helping them reflect and celebrate all the good in the year gone by.

“Naomi had to physically see something good happening.”
— Margot Carter
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Thank you
Global Genes Resources and Events

San Diego, California
September 20 – 22, 2019

Boston: March 30, 2019
Birmingham: May 4, 2019
Denver: May 18, 2019
Sioux Falls: July 13, 2019

Philadelphia
June 7, 2019
Thank You