Barth Syndrome Foundation: Strategic Initiatives Program

Erik Lontok
Director of Research
Barth Syndrome Foundation

• Established in 2000
  • Mission – Saving lives through education, advances in treatment, and finding a cure for Barth syndrome
  • Vision – A world in which Barth syndrome no longer causes suffering or loss of life
• Facilitate and fund research ($6.4M) which has translated into follow-on funding ($32M) and two clinical trials in (US and UK)
• Support the Barth syndrome community of affected individuals and families with information and compassion
• Convene our entire community biennially since our first International Family, Science & Medicine Conference
  o Affected individual & family engagement and education
  o Research Presentations
  o On-site research
And made possible, and on behalf of 308 individuals across 37 countries

US Estimates:
1/300,000~400,000 live births
Barth syndrome is a complex, multi-system disorder
Barth syndrome is a complex, multi-system disorder
Barth Syndrome Foundation Research Grant Program

Project Plan  Application  Peer Review  Board of Directors Decision to Fund  Funds Release; Project Start

Italics are when BSF becomes involved
Barth Syndrome Foundation Strategic Initiatives Program

- Project Plan
- Application
- Peer Review
- Board of Directors Decision to Fund
- Funds Release; Project Start
Barth Syndrome Foundation Strategic Initiatives Program: Arrhythmia

- Research & clinical impact
- Voiced unmet need
Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia

**Project**

- Project Plan
- Application (2021)
- Peer Review
- Board of Directors Decision to Fund
- Funds Release; Project Start
Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia

**Project**
- Working Group
  - Applicants
  - Science & Medical Advisory Board
  - Parent of an affected individual
- Redefined Scope of Work
  - Clinical sites & co-PIs
  - Milestone-based payments
  - Contract not a grant

**Periodic Updates**

- Project Plan
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**Redefined Scope of Work**
- Clinical sites & co-PIs
- Milestone-based payments
- Contract not a grant

- Research & clinical impact
- Voiced unmet need
- Research Investment
- Preliminary data needed for an NIH-style natural history grant application

**Project Plan**
**Application (2021)**
**Peer Review**
**Board of Directors Decision to Fund**
**Funds Release; Project Start**
Barth Syndrome Foundation Strategic Initiatives Program: Arrhythmia

Project

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Project Plan
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Peer Review

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50k, 1 year, single site study

Awarded in Dec. 2022, 350k, 2-3 year, Central IRB & protocol; 2 participating sites; GUID-based data-sharing program
Barth Syndrome Foundation **Strategic Initiatives** Program: Novel Capsids Gene Therapy

**Working Group & Gene Tx Roadmap**
- Research Champion
- Regulatory, Technical Manufacturing consultants
- Expert-informed and critical path driven project plan

**Project Plan**

**Application**

**Peer Review**

**Board of Directors Decision to Fund Tranche 2**

**Tranche 1: Develop Gene Therapy package**

**Pre-IND Meeting**
Two research challenges of Barth syndrome

- The phenotypic presentation of Barth syndrome is broad and variable

- An ultra-rare disease
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Two research challenges of Barth syndrome

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Traditional Grant Program – Expanding Barth syndrome Biology

- Project Plan
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- Funds Release: Project Start

Strategic Initiatives Program – Translating Barth science into medicine

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  - Applicants
  - Science & Medical Advisory Board
  - Parent of an affected individual
- Redefined Scope of Work
  - Clinical sites & co-PIs
  - Milestone-based payments
  - Contract not a grant
- Periodic Updates

Key differences:
- Board-directed & milestone-driven
- Opportunistic and Methodical
- BSF support can take the form of:
  - Funds and/or Foundation research assets
  - Consultant support
  - Industry & non-profit partnerships
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