Barth Syndrome Foundation: Strategic Initiatives Program

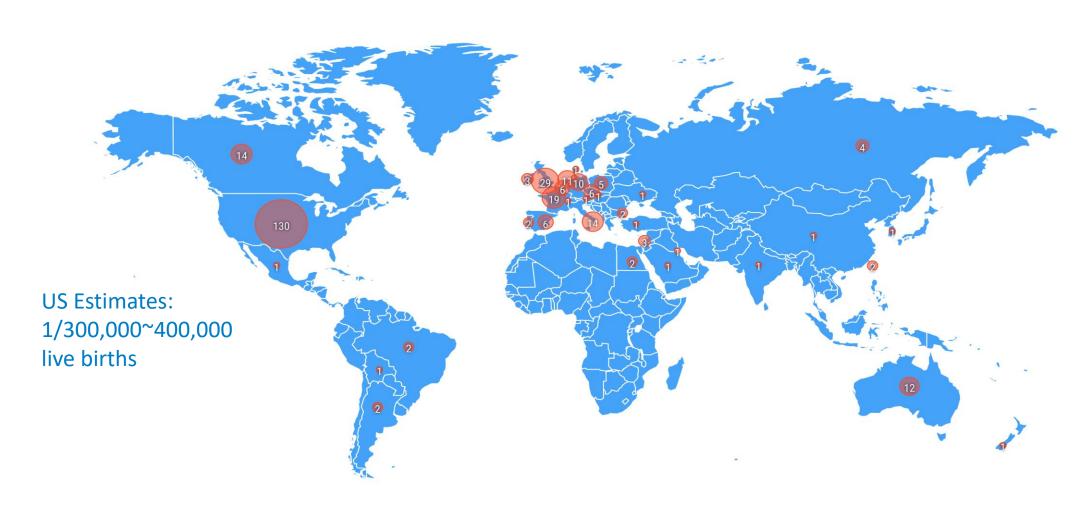


Barth Syndrome Foundation

- 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
 - Affected individual & family engagement and education
 - Research Presentations
 - On-site research



And made possible, and on behalf of 308 individuals across 37 countries

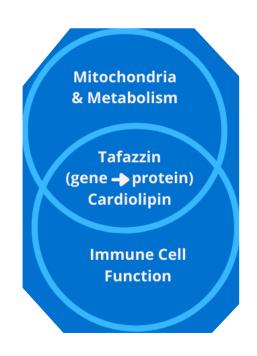




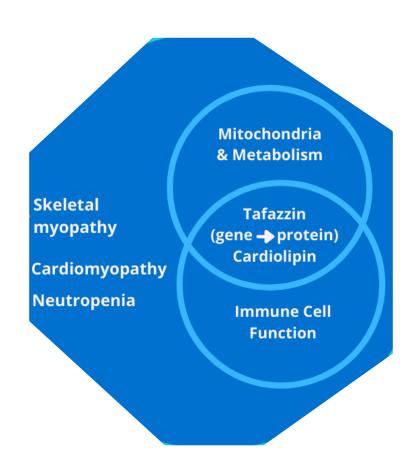
Barth syndrome is a complex, multi-system disorder

Tafazzin (gene → protein) Cardiolipin



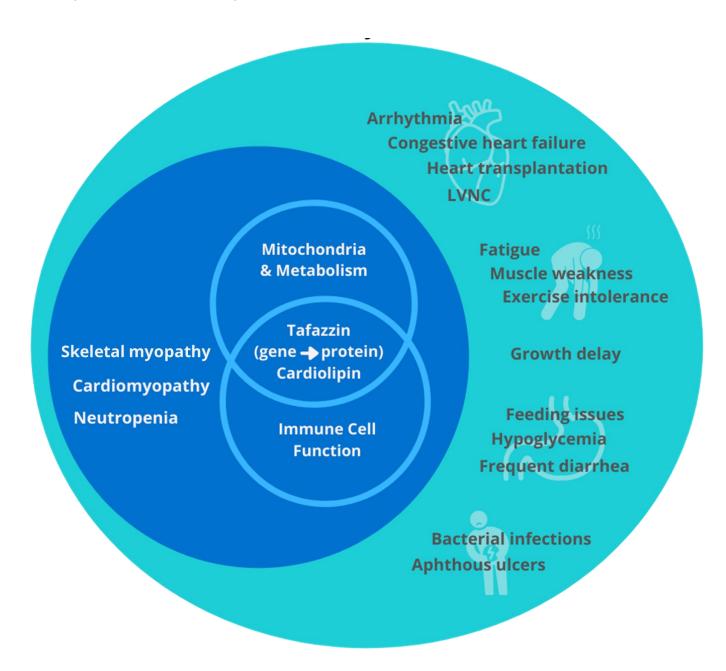








Barth syndrome is a complex, multi-system disorder





Barth Syndrome Foundation Research Grant Program

Project Plan

Application

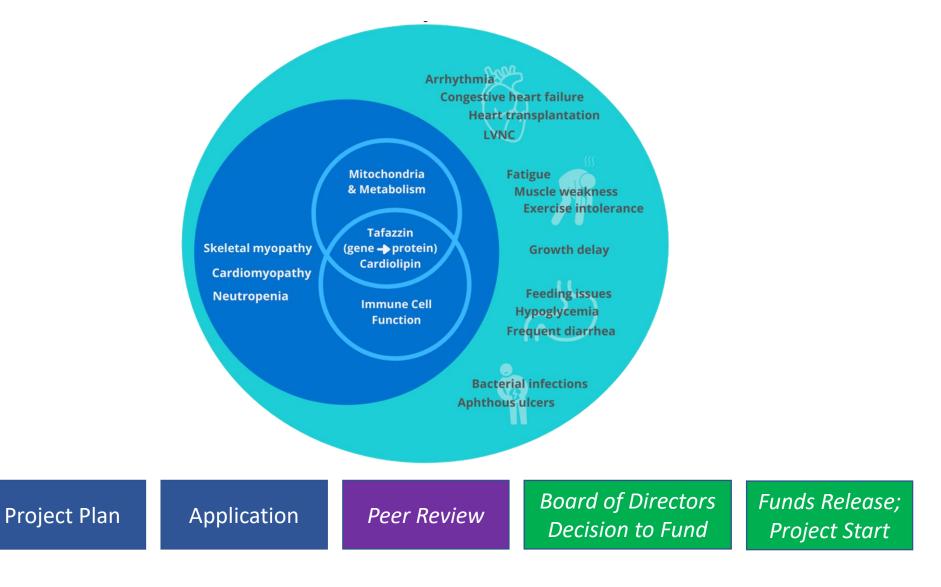
Peer Review

Board of Directors
Decision to Fund

Funds Release; Project Start



Barth Syndrome Foundation 2021 Strategic Planning Initiative





Project Plan

Application

Peer Review

Board of Directors
Decision to Fund

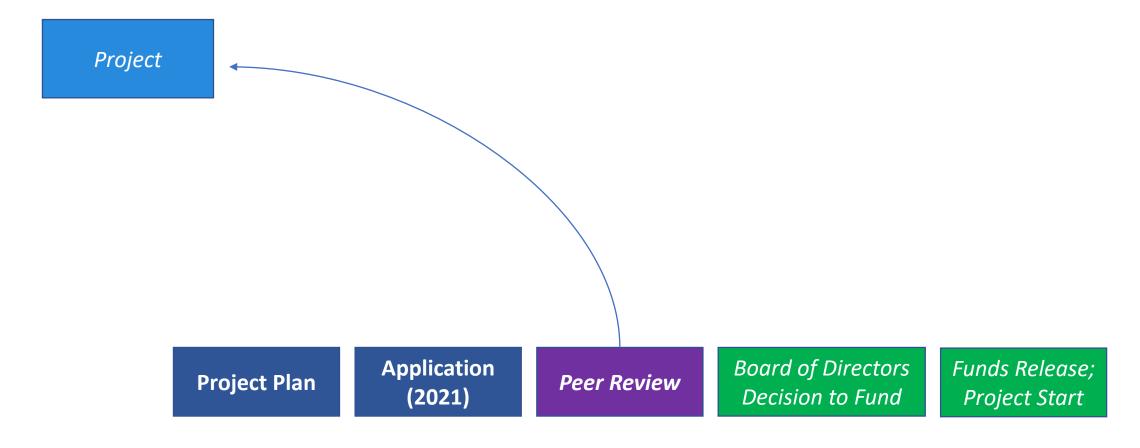
Funds Release; Project Start



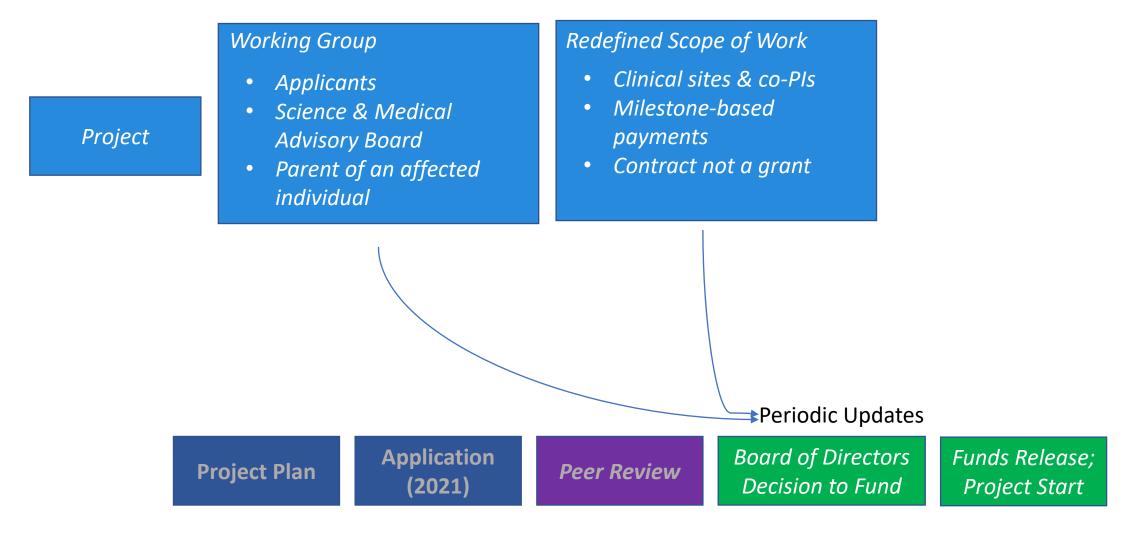
Project PlanApplication
(2021)Peer ReviewBoard of Directors
Decision to FundFunds Release;
Project Start

- Research & clinical impact
- Voiced unmet need











(2021)

Working Group Redefined Scope of Work Research & clinical impact Voiced unmet need Clinical sites & co-PIs **Applicants** Research Investment Milestone-based Science & Medical Preliminary data needed for Project **Advisory Board** payments an NIH-style natural history Contract not a grant Parent of an affected grant application individual Board of Directors Funds Release; **Application Project Plan** Peer Review

Decision to Fund



Project Start

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

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

Project Plan

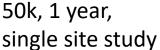
Application (2021)

Peer Review

Board of Directors Decision to Fund

Funds Release; Project Start

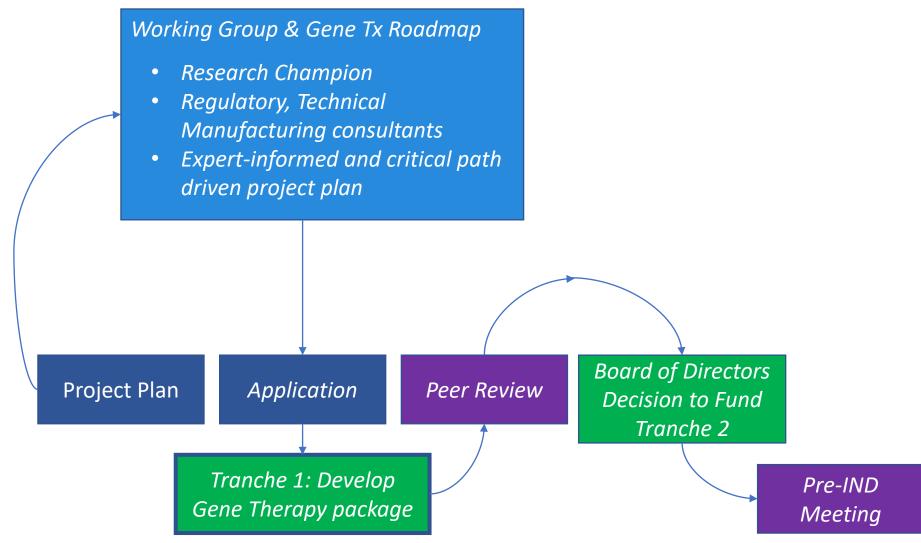
50k, 1 year, single site study



Awarded in Dec. 2022, 350k, 2-3 year, Central IRB & protocol; 2 participating sites; GUID-based data-sharing program

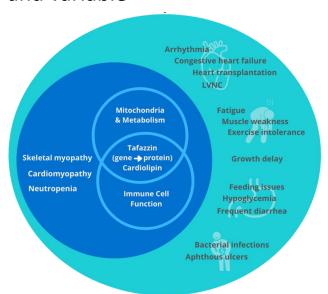


Barth Syndrome Foundation Strategic Initiatives Program: Novel Capsids Gene Therapy

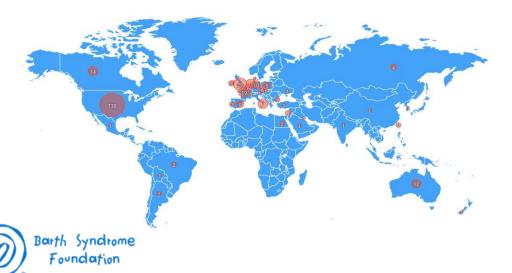




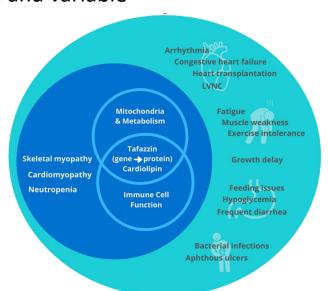
The phenotypic presentation of Barth syndrome is broad and variable



An ultra-rare disease



 The phenotypic presentation of Barth syndrome is broad and variable



An ultra-rare disease



Traditional Grant Program – Expanding Barth syndrome Biology

Project Plan

Application (2021)

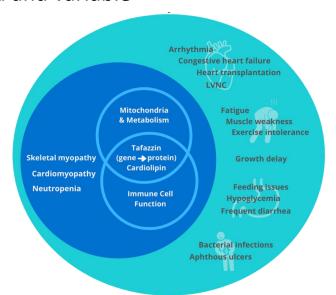
Peer Review

Board of Directors

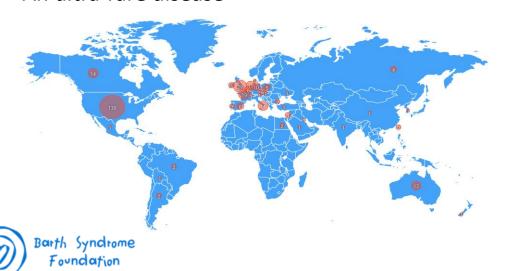
<u>Decision</u> to Fund

Funds Release; Project Start

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An ultra-rare disease



Traditional Grant Program – Expanding Barth syndrome Biology

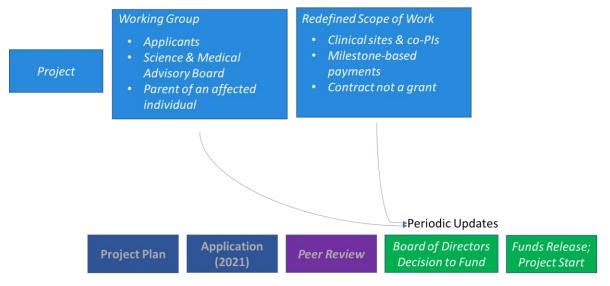
Project Plan

Application (2021)

Peer Review

Board of Directors Punds Release;
Project Start

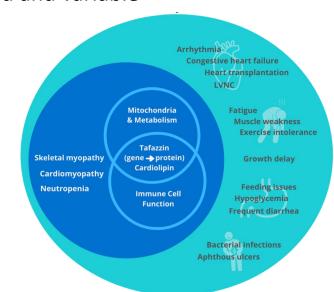
Strategic Initiatives Program – Translating Barth science into medicine



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

The phenotypic presentation of Barth syndrome is broad and variable



An ultra-rare disease





Melissa Huang





Emily Madalinski

Brett Smith



Emily Milligan



Shelley Bowen

