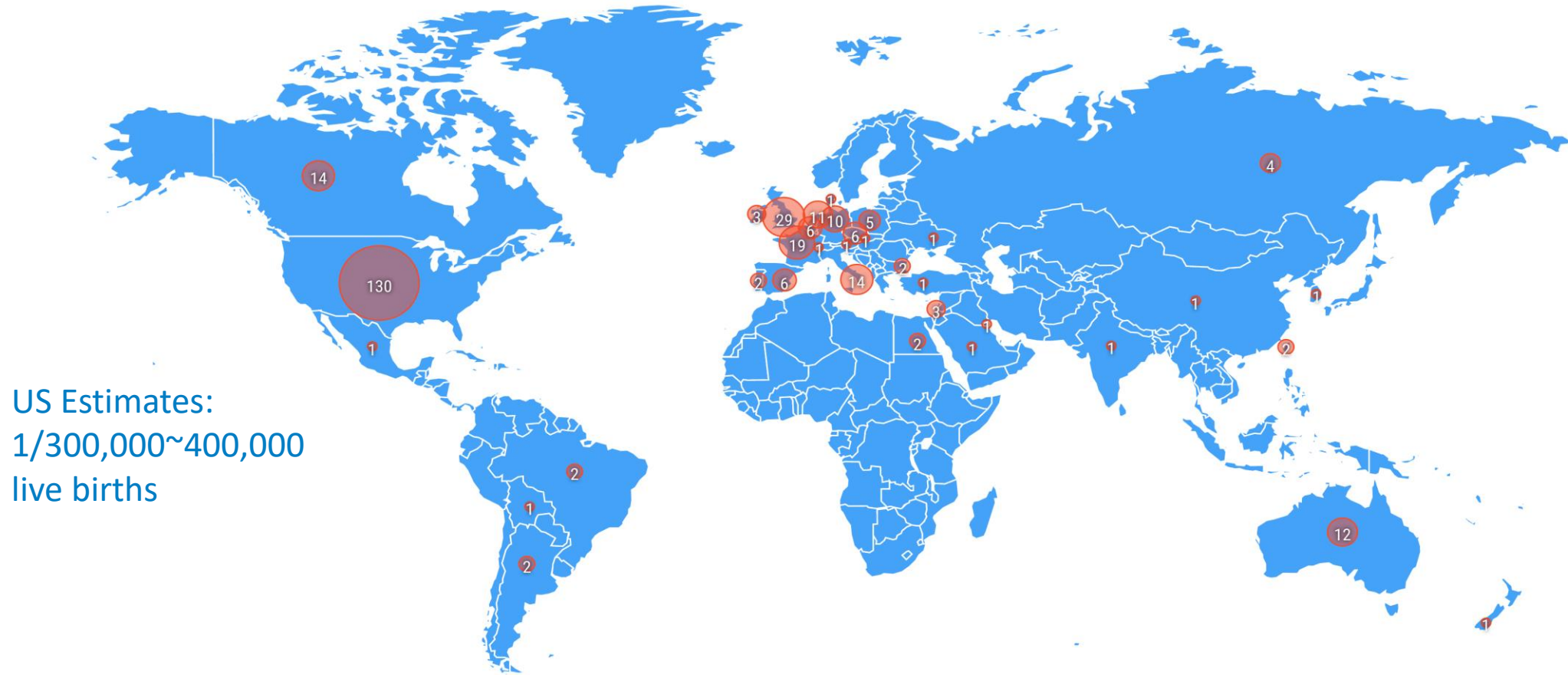


# Barth Syndrome Foundation: Strategic Initiatives Program

## 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

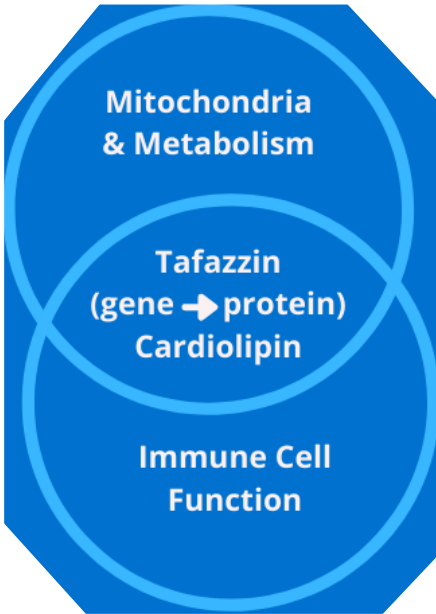
And made possible, and on behalf of 308 individuals across 37 countries

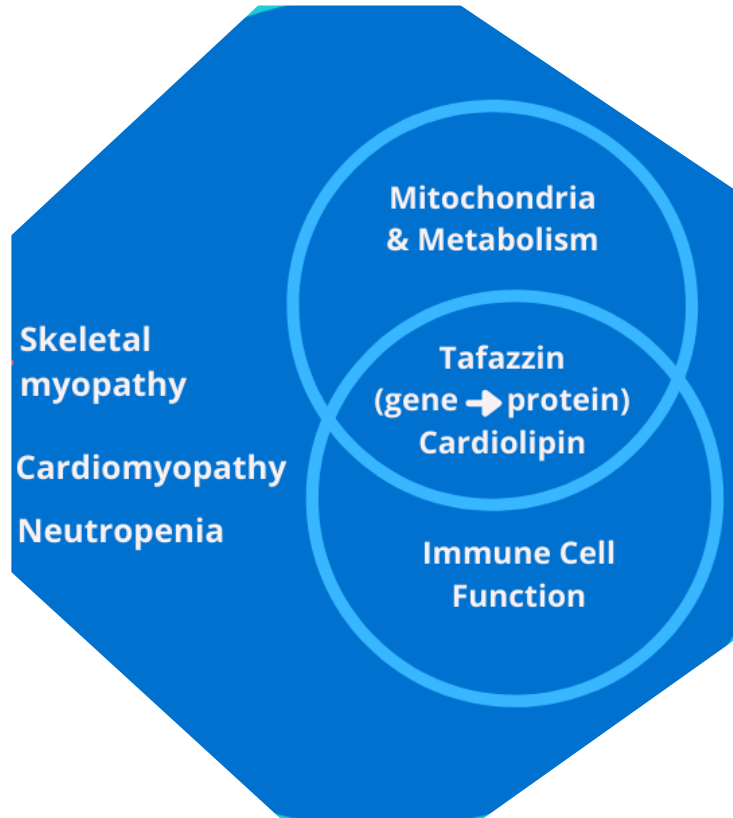


US Estimates:  
1/300,000~400,000  
live births

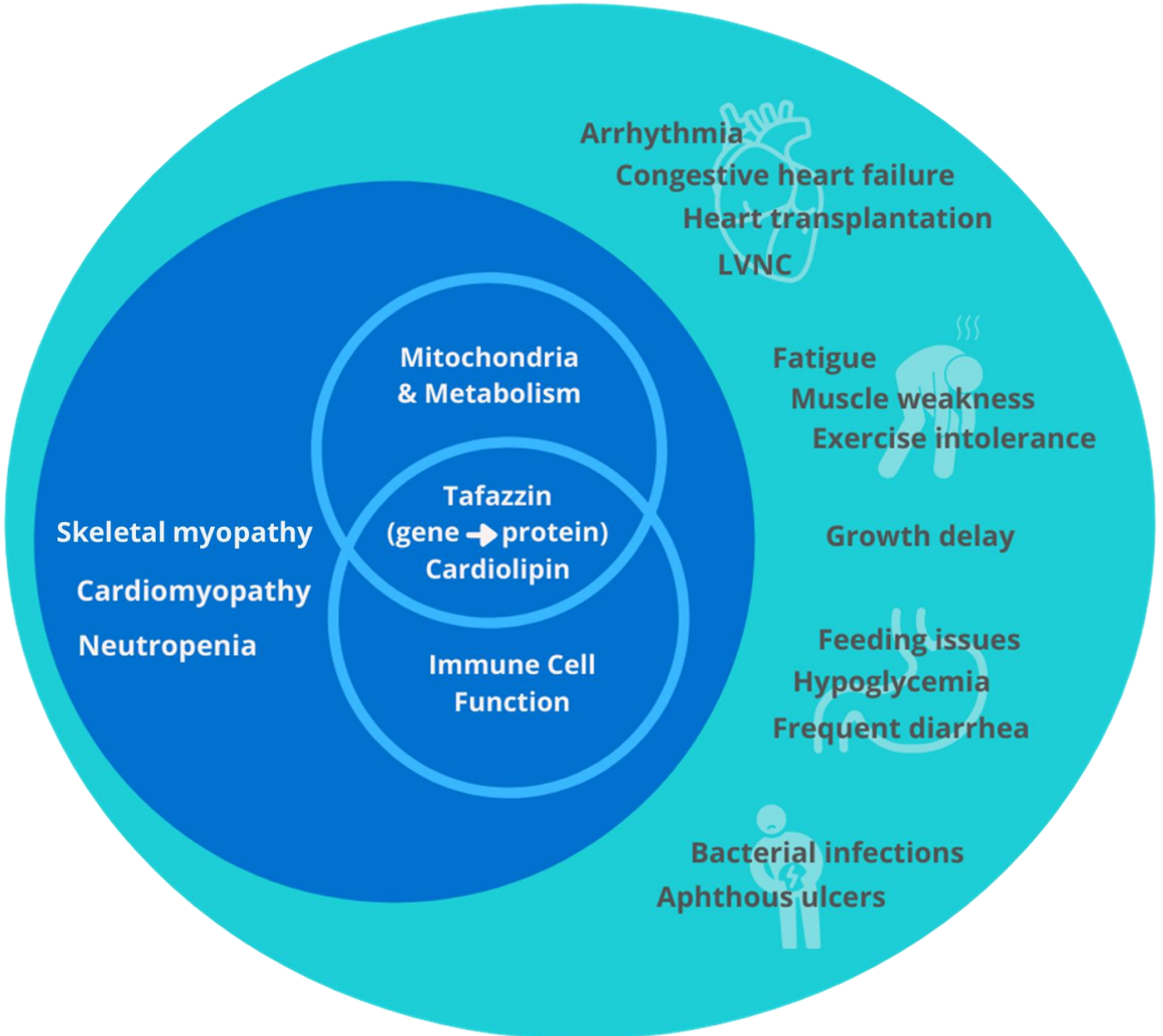
Barth syndrome is a complex, multi-system disorder

Tafazzin  
(gene → protein)  
Cardiolipin





# Barth syndrome is a complex, multi-system disorder



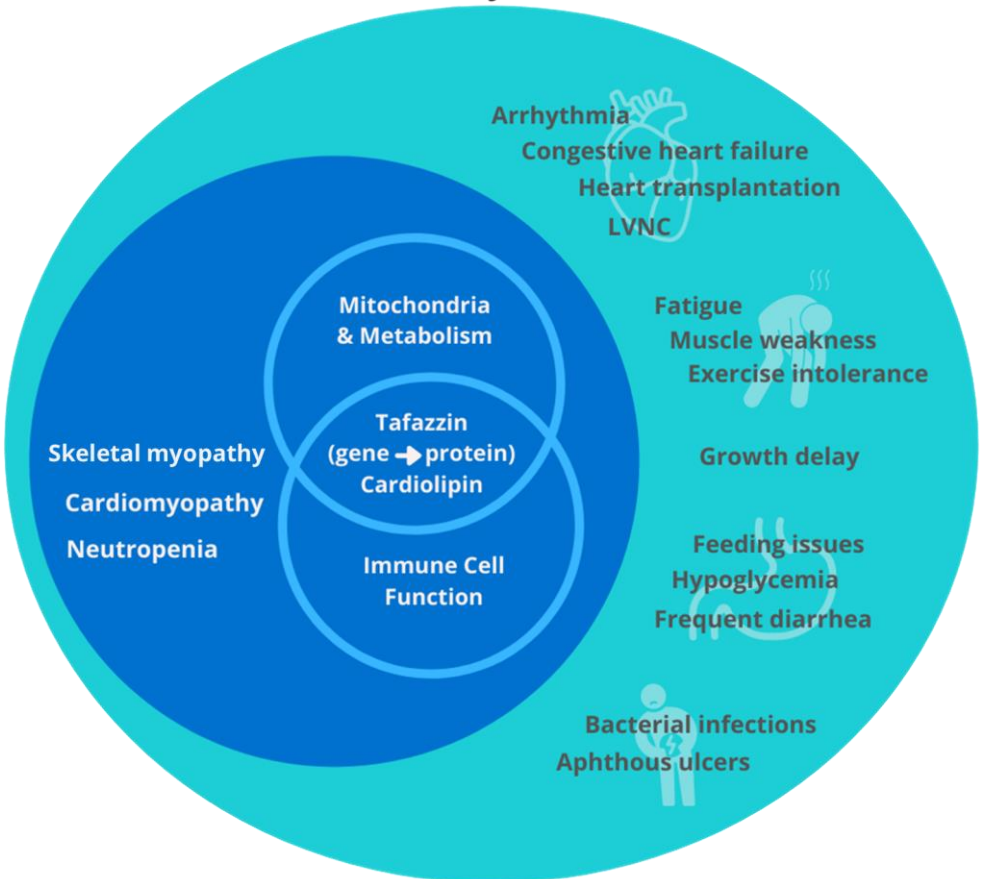
# Barth Syndrome Foundation **Research Grant** Program



*Italics are when BSF becomes involved*



# Barth Syndrome Foundation 2021 Strategic Planning Initiative



Project Plan

Application

Peer Review

Board of Directors  
Decision to Fund

Funds Release;  
Project Start

# Barth Syndrome Foundation **Strategic Initiatives** Program

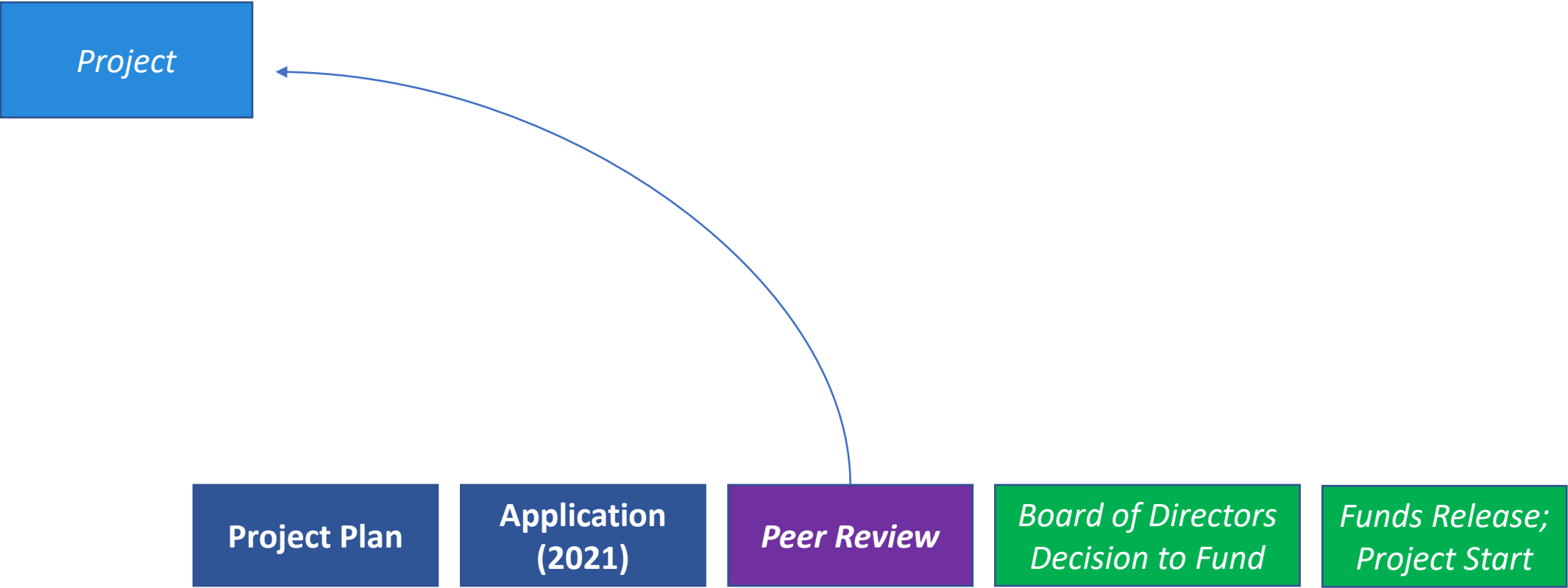


# Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia

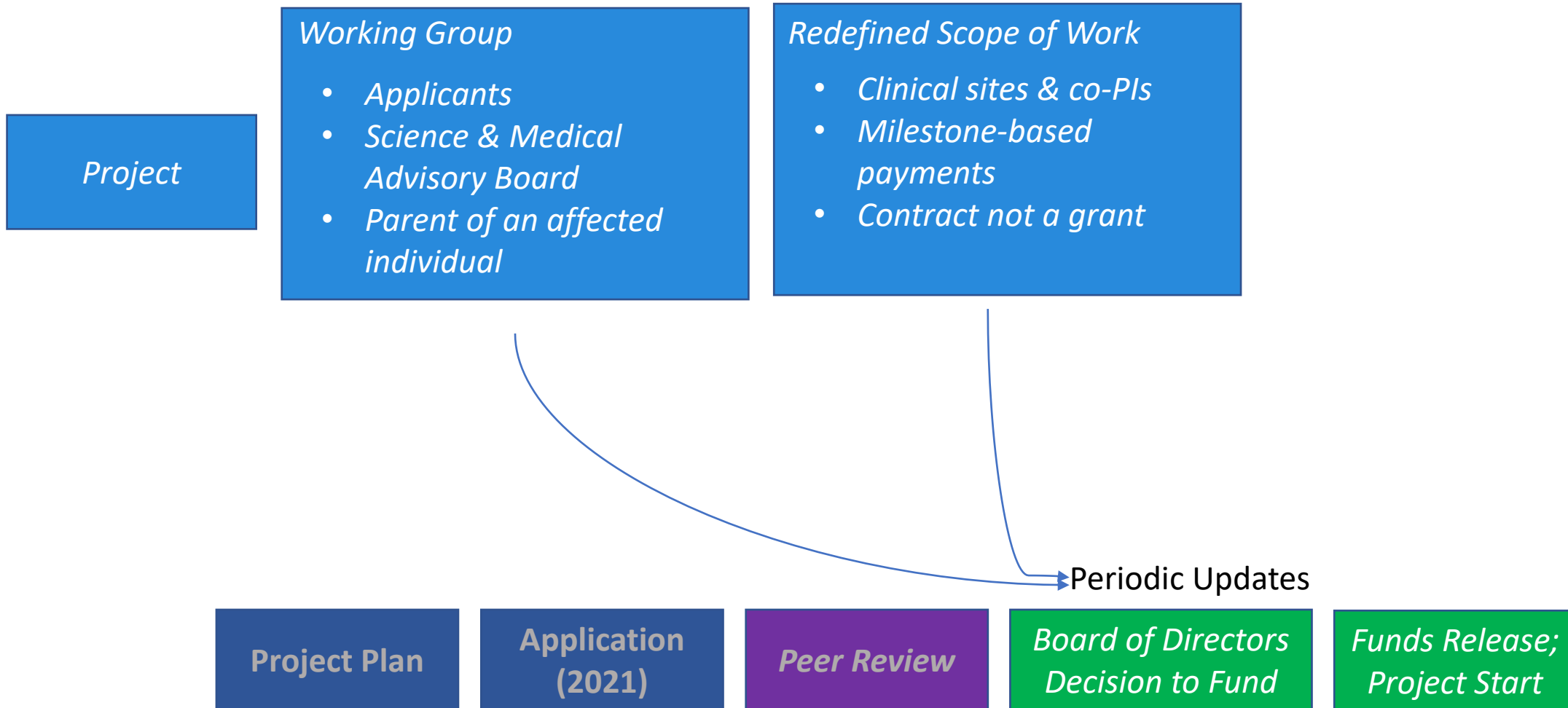


- Research & clinical impact
- Voiced unmet need

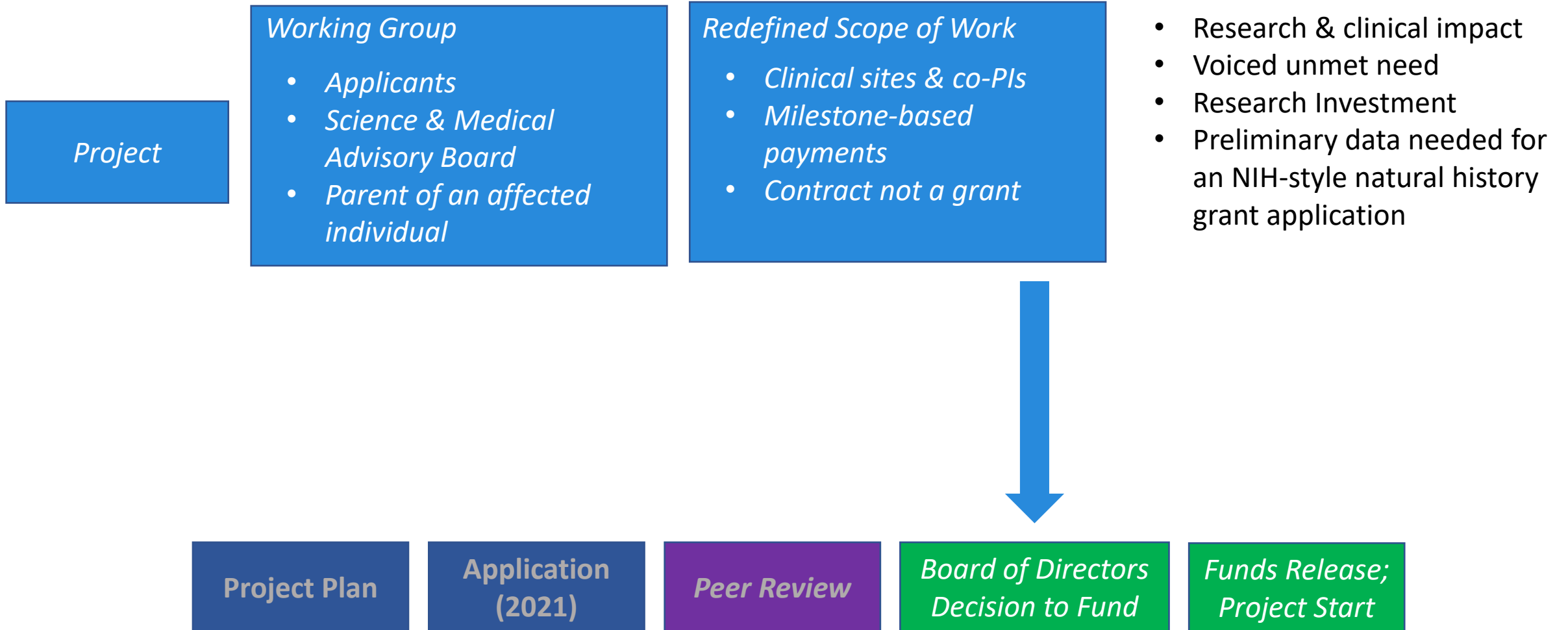
# Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia



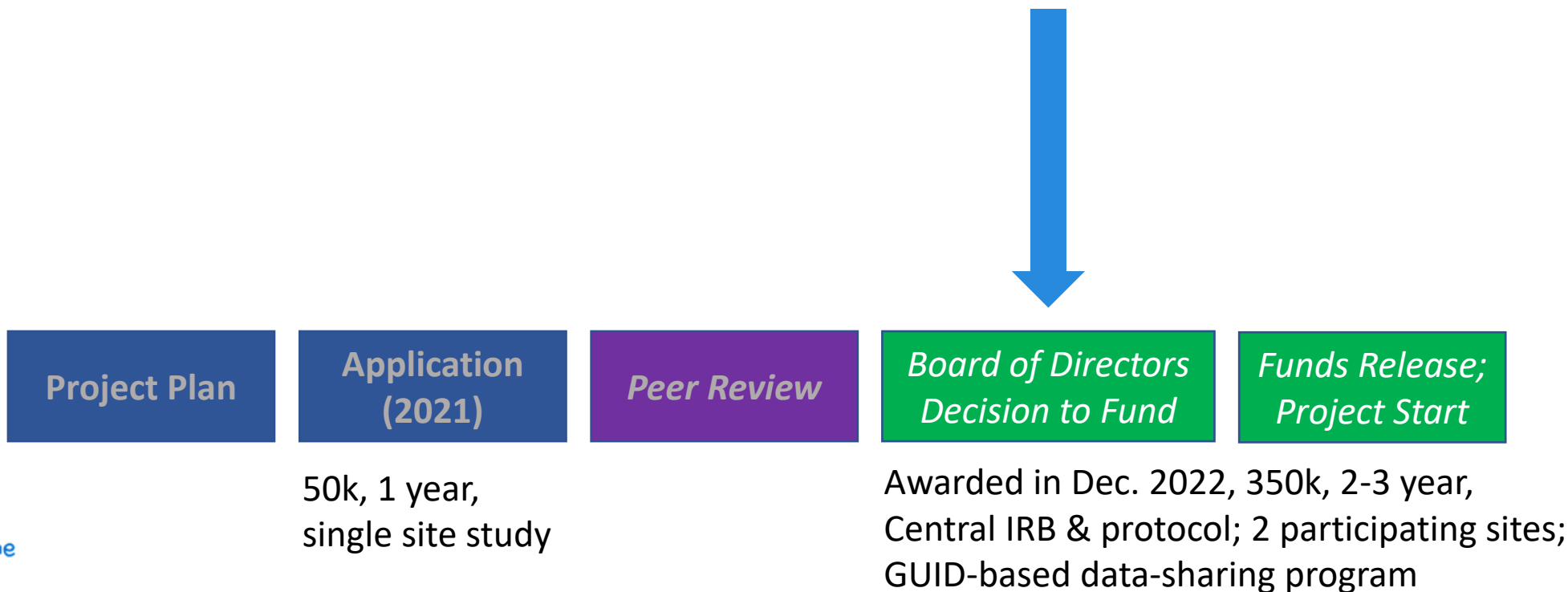
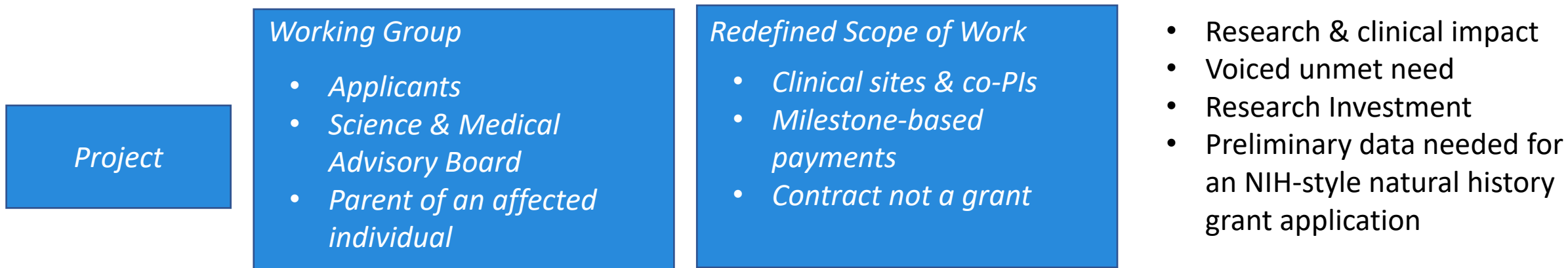
# Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia



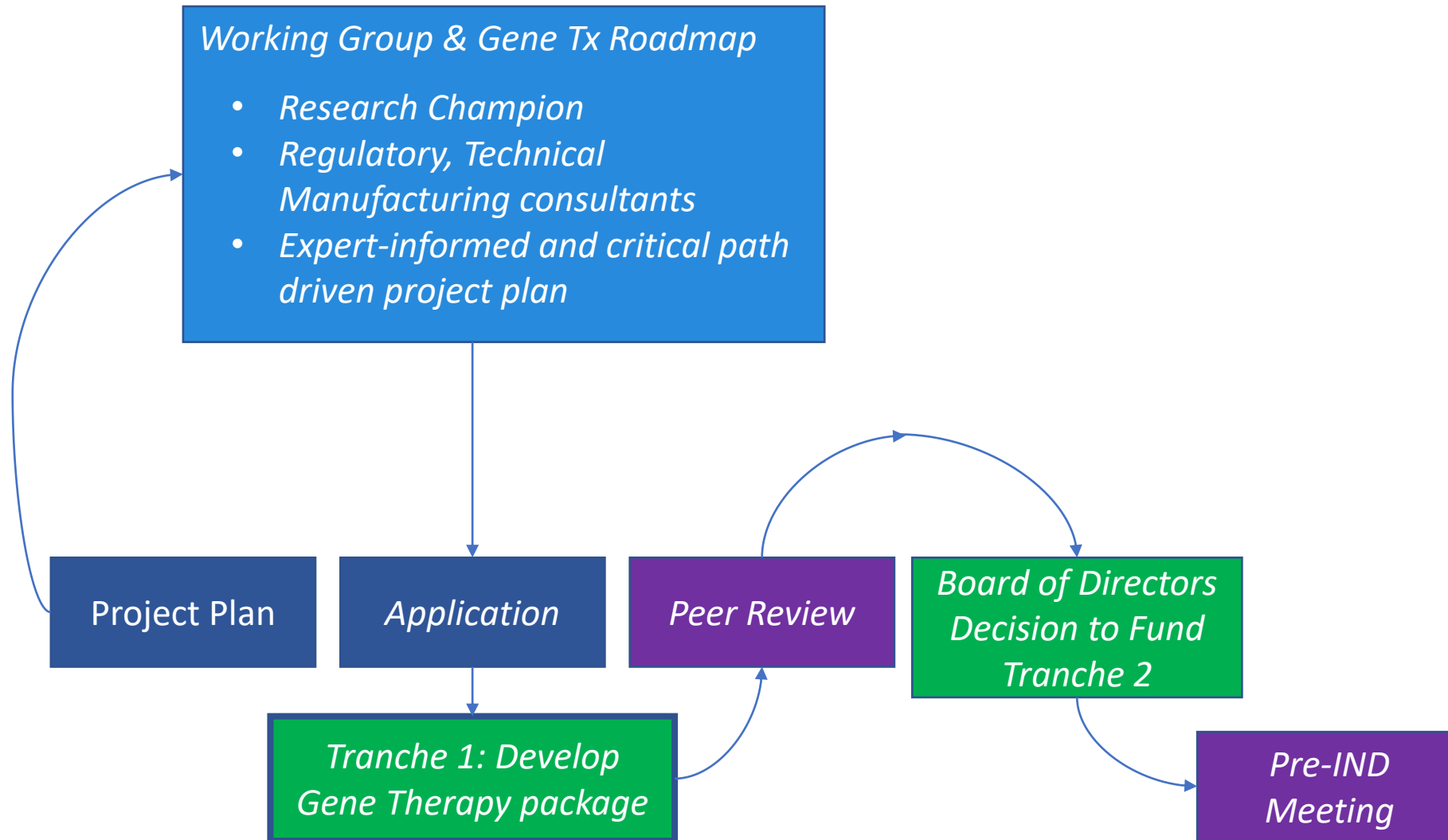
# Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia



# Barth Syndrome Foundation **Strategic Initiatives** Program: Arrhythmia



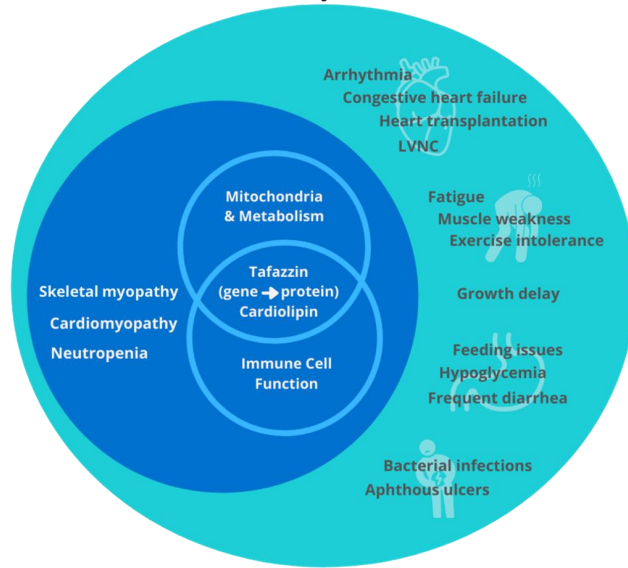
# Barth Syndrome Foundation **Strategic Initiatives** Program: Novel Capsids Gene Therapy



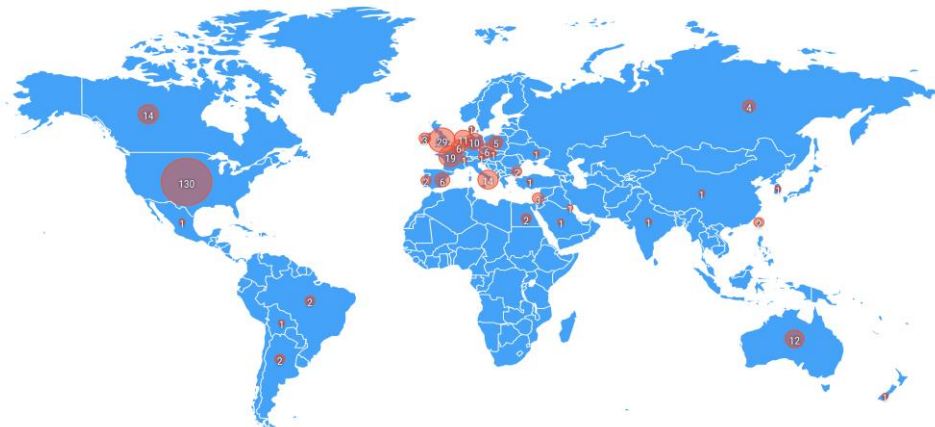


## Two research challenges of Barth syndrome

- The phenotypic presentation of Barth syndrome is broad and variable

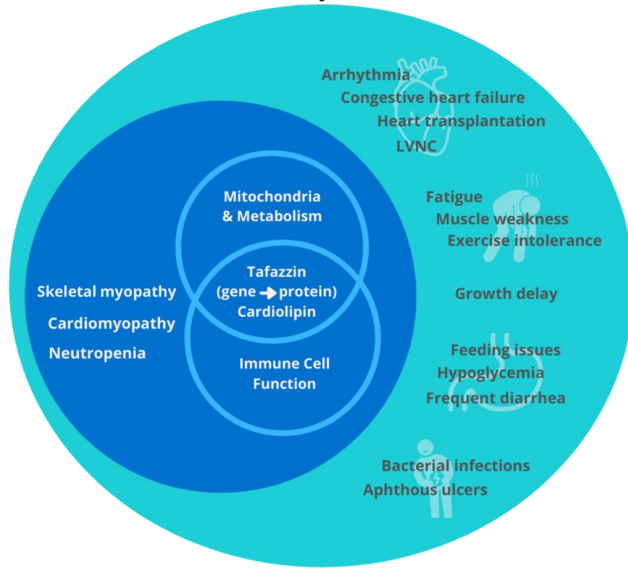


- An ultra-rare disease



## Two research challenges of Barth syndrome

- The phenotypic presentation of Barth syndrome is broad and variable



- An ultra-rare disease

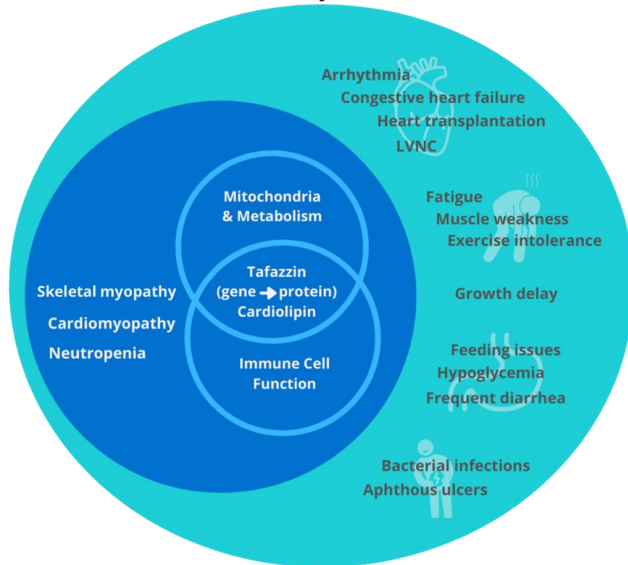


## Traditional Grant Program – Expanding Barth syndrome Biology



## Two research challenges of Barth syndrome

- The phenotypic presentation of Barth syndrome is broad and variable



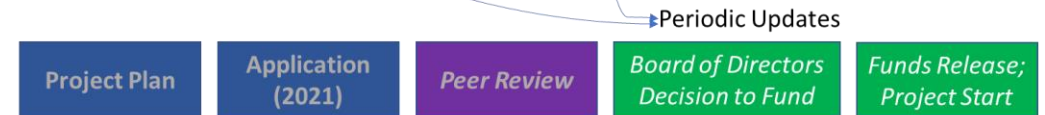
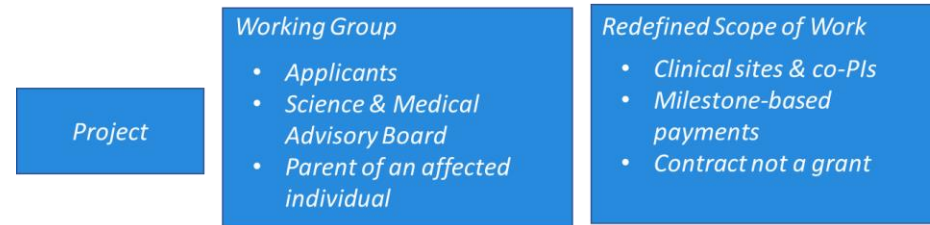
- An ultra-rare disease



## Traditional Grant Program – Expanding Barth syndrome Biology



## Strategic Initiatives Program – Translating Barth science into medicine

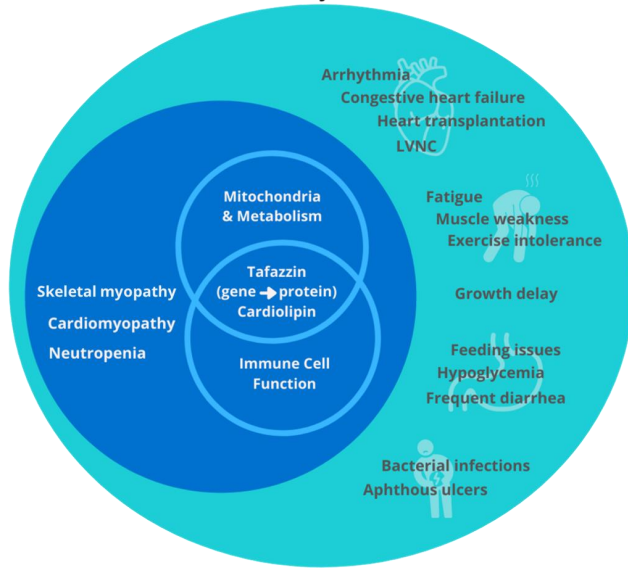


### Key differences:

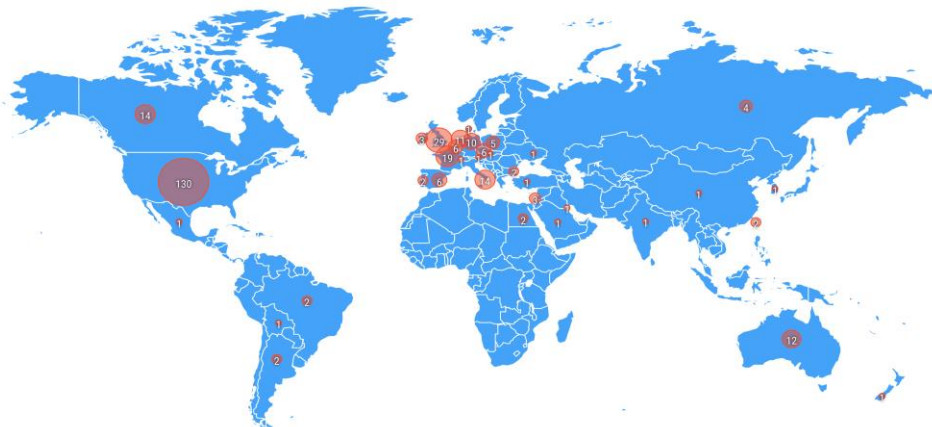
- Board-directed & milestone-driven
- Opportunistic and Methodical
- BSF support can take the form of:
  - Funds and/or Foundation research assets
  - Consultant support
  - Industry & non-profit partnerships

## Two research challenges of Barth syndrome

- The phenotypic presentation of Barth syndrome is broad and variable



- An ultra-rare disease



Melissa Huang



Emily Madalinski

Shelley Bowen



Emily Milligan



Brett Smith