RARE science™

Accelerating Cures for RARE Kids

OUR MISSION

Accelerating the identification of more immediate medicines for kids with rare disease potentially through repurposing currently approved drugs and other therapeutic approaches.

2017 ACCOMPLISHMENTS

2,500

Children with rare diseases identified through the RARE Bear Program

>350

Rare Diseases represented

38

Countries representing Patient Families

Kicked-Off

Development of Health Tracker App

1

Global iPStem Cell Research Program to Find an Effective Treatment for a Rare Disease (ADCY5)

OUR STRATEGY

1) GLOBAL OUTREACH TO CHILDREN

Through our RARE Bear program, Rare Science has found children with 200 different rare diseases spanning across 35 countries and let them know that they are not alone

2) BUILD PATIENT COMMUNITIES

Rare Science connects families whose children suffer from the same rare diseases and offers tools to help them share valuable information with each other in coping with their diseases

3) Drive Collaborative Research Alliances

Rare Science is developing collaborations with university labs and industry to facilitate the research needed to find viable treatments for rare diseases that affect our most vulnerable patient population, children.

2018 GOALS

5,000

Children with rare diseases identified through the RARE Bear Program

700

Rare Diseases represented

70

Countries representing Patient Families

1

Patient Community Using Health Tracker App

2

Additional Global iPSC Research Programs to Find Effective Treatments for a Rare Disease

info@rarescience.org

www.rarescience.org

501(c)(3) organization
47-1744221

Rare Patient Journey
and Rare Research and Development

Patient Journey

- Need for Urgency
- Finding a Diagnosis
- Access to Information
- Available Therapies
- Support and Guidance
- Access to Therapeutics
- Reimbursement

Research and Development

- Access to Patients and Biosamples
- Scattered islands of data
- Small data sets
- Research and Clinical Strategy Coordination
- Clinical Trials/Regulatory Process
- Access to Funding

The path and voice of the patient and rare disease research and development paths need to be integrated to accelerate diagnosis and treatment options

RARE science™
Accelerating Cures for RARE Kids

Example: ADCY5 Patient Community Building

Community Support

- Patient outreach – identify patient families world-wide
- Build Patient Community
- Obtaining critical number for statistical significance of observable traits/clinical phenotype
- Scientific Education
- Participation

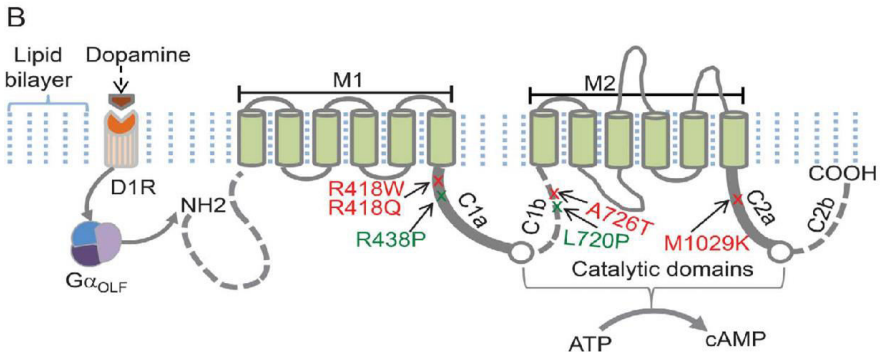
ADCY5
.ORG

RARE science™
Accelerating Cures for RARE Kids

ADCY5 iPSC Program

Genotype - Phenotype

ADCY5 known patient mutations	n
c.1253G>A (p.Arg418Gln)	2
c.2176G>A (p.Ala726Thr)	1
c.1252C>G, p.Arg418Gly	1
c.2088+1G>A	1
c.1252C>T (p.Arg418Trp)	3
c.1252C>T, p.Arg418Trp, c.5645A>G, p.Asp1882Ser DOCK c.2557C>T, p.R853C FAT4 c.524G>T, p.R175L, c4000G>4, p.V1334M	1
Total	9



RARE science™
Accelerating Cures for RARE Kids

Key Take Aways

- One challenge of solving rare and common disease will be the interpretation and validation of new causal genes, variants and gene-gene interactions
- Rare Disease patient communities and new Technologies can reveal molecular pathways that are masked by the biological complexities of common disease
- Empowering RARE Disease Patient Families with tools to build global communities ad drive research to clinic will impact global health
- Do not underestimate the power of the “sharing economy of good”
- Scaling will require all members of the rare disease community eco-system

RARE science™
Accelerating Cures for RARE Kids