2025 CSNK2A1 Foundation Impact Report

Fueling Discovery, Built for Impact

FOUNDATION OVERVIEW

OCNDS Discovered: **2016**

Foundation Established: **2018**

Mission:

To find a cure for Okur-Chung Neurodevelopmental Syndrome (OCNDS) and ensure affected individuals live happy and fulfilling lives.

Vision:

A world where OCNDS is recognized, understood & treatable.

Main Symptoms:

- Speech Delay or Inability to Speak
- Cognitive Impairment
- Sleep Issues
- Autism Spectrum Disorder Traits
- Global Developmental Delay
- Brain Abnormalities & Hypotonia

Range of Impact:

OCNDS affected every patient differently.
The symptoms range from mild to severe.

Global Reach:

377 +86 individuals from
48 +6 countries speaking
30 +5 languages

All (+) are increases since January 2024

Our Team:

Governance
Board of Directors

Science
Scientific Advisory Board
Chief Science Officer

Lived Experience
Parent Advisory Board
Regional Ambassadors
Families Voices

6 Key Focus Areas:

- Research
- Advocacy
- Awareness
- Fundraising
- Family Support
- Capacity Building

How it All Connects:

OCNDS is complex, and so is the path to answers. Our work is intentionally connected across six focus areas, from listening to families and collecting real-world data to driving research, awareness, advocacy, fundraising, and capacity building. Together, these efforts reinforce one another, moving us closer to treatments while improving quality of life today.

RESEARCH

We advanced OCNDS research by integrating natural history data, translational models, and community-informed priorities to build readiness for disease-modifying treatments.

Research Output & Translation

- **Awarded 6 new research grants**
- **Co-funded a Young Investigator Award** recognizing OCNDS researcher Dr. Sunanjay Bajaj, **supporting early-career talent** and advancing OCNDS research.
- **Testing 7 FDA-approved drugs already on the pharmacy shelves** in our animal models to treat some of the most debilitating OCNDS symptoms
- **Generated 7 variant-specific fly models** to study sleep and circadian disruption
- **Advanced 5 case reports** addressing gaps in the medical literature
- OCNDS research literature grew to **48 total publications (+10)**, signaling continued momentum across the field
- **Applied for 9 highly competitive grants; received 5 grants** totaling **$290,194** (some as in-kind support)
- **59 researchers and clinicians attended** our 2025 Scientific and Family Conference

Real-World Data & Collaboration

- **Hosted** our **1st in-person OCNDS crowdsourcing session** of future research priorities between researchers and families at our largest conference to date.
- **Expanded** our **research network** to **122 researchers** and **clinicians (+38)**
- **Collected 50 biological samples** at our conference through COMBINEDBrain
- **Convened 4 scientific roundtables** to accelerate cross-disciplinary collaboration
- **Enrolled 9 new families** in Citizen Health, totaling **71** for centralized medical record storage and research participation.
- **Launched** our first **Gene Therapy Community Poll** to understand how families feel about gene-based approaches as a critical step toward targeted, potentially disease-modifying treatments for OCNDS

Simons Searchlight Natural History & Data

- **Expanded** the Simons Searchlight OCNDS **cohort by 40, totaling 187 participants**
- **Verified genetic reports** for **35** new participants, totaling **161 participants**
- **Collected survey data** from **38** participants, totaling **147 participants**
- **Published the Foundation's 1st study using Simons Searchlight data**, advancing clinical understanding of OCNDS, **viewed over 3,000 times** and **downloaded over 350 times**

ADVOCACY & POLICY

We supported policies that enable research, improve access to care, and accelerate innovation.

- **Signed on to 27 federal** and **state advocacy efforts**
- **Elevated OCNDS in the Irish Dáil** (main legislative body in Ireland) for the 1st time through parent advocacy
- **Supported parent testimony in Massachusetts** to advance official recognition of OCNDS Awareness Day
- **Advanced the ICD-10 application**, which was formally reviewed by the ICD-10-CM Committee, with encouraging feedback and next steps underway

AWARENESS

We expanded visibility for OCNDS to help families find community sooner and drive earlier recognition by clinicians and researchers.

Visibility & Recognition

- **Developed and launched the OCNDS Wikipedia page**
- **Illuminated 15+ landmarks worldwide** for OCNDS Awareness Day, including Niagara Falls & High Level Bridge in Canada
- **Secured 5 official proclamations** declaring April 5 as OCNDS Awareness Day

Storytelling & Reach

- **Published 13+ blog posts** featuring families and advocates
- **Developed 5+ intern-authored family stories**, each paired with a companion reel to extend reach
- **Produced 8+ short-form reels** amplifying written stories through visual storytelling, with our Instagram and Facebook accounts **averaging 13,500+ views a month.**
- **Amplified awareness through 9+ podcasts, videos, and media features**, expanding awareness beyond the OCNDS community
- **Launched 3+ national and international awareness campaigns** featuring OCNDS national and international campaigns featuring OCNDS families and advocates

FUNDRAISING

We mobilized community support to fuel research, family programs, and advocacy.

- **Raised $690,000+ in 2025**
- **Shattered records at the Annual Golf Tournament**, raising **$530,000+**
- **Raised $130,000+ with a $50,000 matching gift for Giving Tuesday** from Joan and Charlie Davis
- **Engaged 230 participants across 14 countries** through our virtual Run, Walk & Roll Event, raising **$36,000+**
- **Activated grassroots fundraising**, including **32 Facebook fundraisers** & community-led events

FAMILY SUPPORT

We strengthened connection, education, and access for families worldwide.

Community & Connection

- **Welcomed 86 new families across 48 (+6) countries** and **30 (+5) languages**
- **Hosted 3 regional ambassador meetups** in the Netherlands, England, and France
- **Facilitated 5 multilingual family Zoom calls**
- **Hosted an international family call** with **16 families in China**
- **Planned & hosted our largest in-person conference to date with almost 200 attendees**, including families, researchers & clinicians, **awarding $10,000 in travel scholarships** & **covering all meals and lodging** to help **reduce financial barriers for attendees**

Education & Resources

- **Created** and **launched a Family Resource Planning Guide** that helps families better understand family planning considerations related to OCNDS
- **Published 12 Science Snapshots** simplifying complex research topics
- **Developed** and **launched an educational advocacy tool and webinar** for school-based support
- **Highlighted 12 Milestone Mondays** on our social media channels, celebrating individual achievements
- **Translated 4 core resources into 30 languages**, including the family-friendly GeneReview, clinical one-pager, genetic variant guide, and family planning resource

CAPACITY BUILDING & PARTNERSHIPS

We invested in people, infrastructure, and partnerships to ensure long-term sustainability and outsized impact.

- **Maintained 27 active partnerships** across research, advocacy, and community engagement
- **Launched 2 new partnerships** in 2025: Fondation Maladies Rares (creating a pathway for French families and supporters to directly fund OCNDS research) and We the Action
- **Formed 1 formal alliance, the CK2 Butterfly Collective**, with the CSNK2B Foundation to align CK2-related communities

Leadership & Governance

- **Foundation leadership attended 11 conferences** spanning neuroscience, patient advocacy, and drug development
- **Strengthened scientific oversight** by welcoming **5 new Scientific Advisory Board** members, **Dr. Danielle Caefer, Dr. Michael Boland, Vanessa Vogel-Farley, Dr. Hilary Eaton, and Dr. Matt Eaton**
- **Held 6 Parent Advisory Board meetings with 11 active members**, ensuring that lived experience remains central to our programs, research priorities, and community engagement.
- **Launched 3rd Parent Advisory Board cohort**, welcoming **14 members**, expanding representation across North America, Europe, and the Middle East

Community in Action

- **Mobilized 70 volunteers worldwide**
- **Supported 15 interns** across research, education, storytelling, and operations
- **Recognized 4 Giving Impact Award honorees** for advancing research, community leadership, and Foundation capacity

The CSNK2A1 Foundation continues to accelerate discovery, strengthen community, and build the foundation needed to deliver future treatments for individuals living with OCNDS worldwide.

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