



Year in Review

2021

Schaaf-Yang Syndrome
Research Program



FOUNDATION FOR
PRADER-WILLI
RESEARCH

When treatments for SYS have been found,
it will be because of **all of us.**

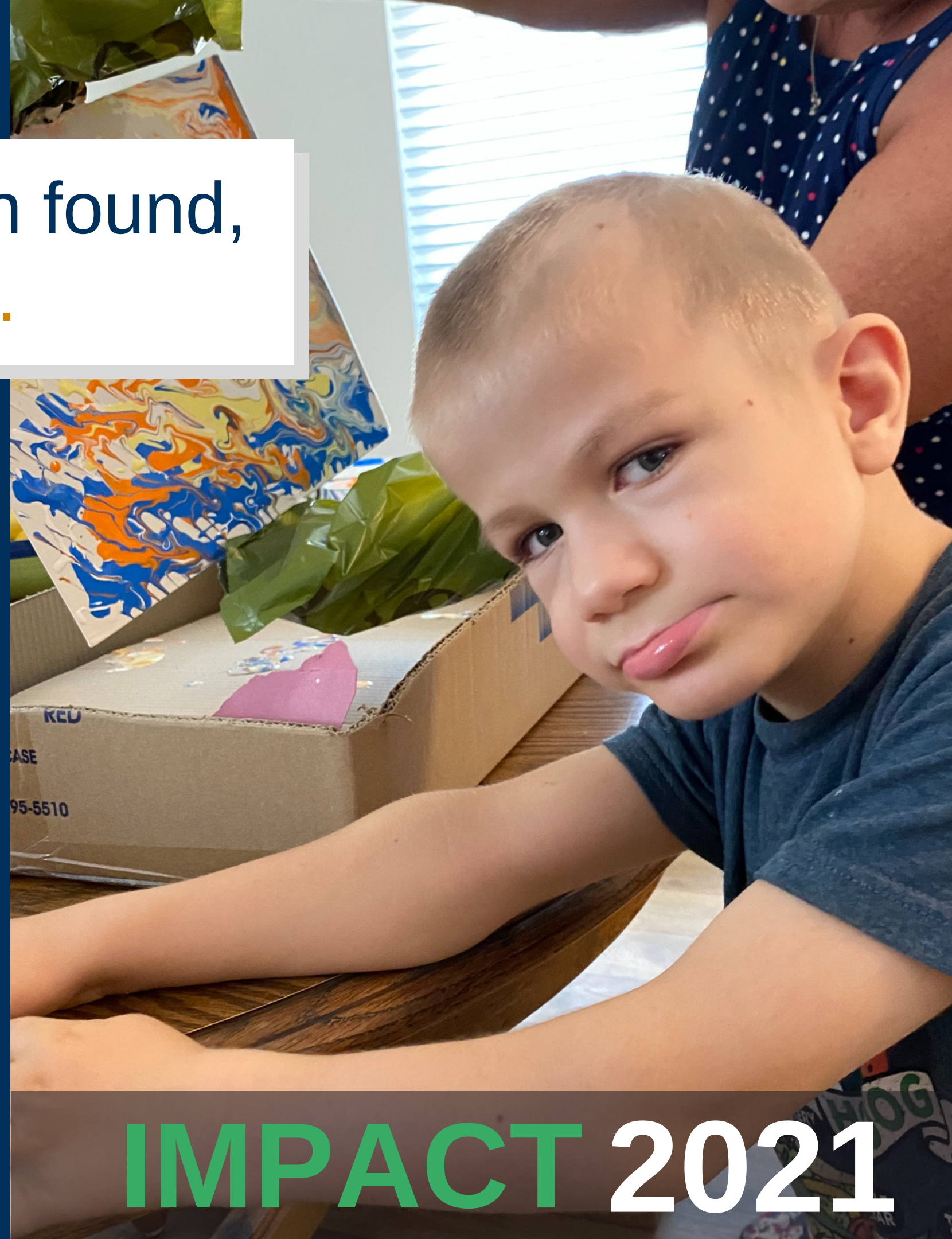
589

donations in 2021

\$219,148

raised for SYS research

IMPACT 2021



IMPACT
2021

SYS Community Publishes First Steps Guide for Newly Diagnosed Families

Information on caring for a loved one with Schaaf-Yang can be hard to find so we published a free download, ***First Steps: A Parent's Guide to Schaaf-Yang Syndrome***, that contains important insights and strategies for new parents caring for a loved one with SYS.



IMPACT
2021

SYS Patient Voices Questionnaire Documents Needs of SYS Community

Through the SYS Patient Voices Questionnaire, SYS caregivers from around the world provided **insight on the unmet medical needs of SYS, the impact SYS has on the family and caregiver, and treatment priorities for the SYS community.**

This questionnaire was an important first step to documenting the needs of the SYS community. Results will be published in 2022.



IMPACT

2021

Advancing the Discovery of New Therapies for Schaaf-Yang Syndrome


Research is critical to finding treatments, and an eventual cure for Schaaf-Yang syndrome. FPWR funded researchers continue to examine potential therapies that may lead to transformative treatments for SYS.

ADVANCING SYS THERAPEUTIC RESEARCH

Recent SYS Research Funded by the Foundation for Prader-Willi Research


2019 RAPAMYCIN AS A THERAPEUTIC OPTION
DR. CHRISTIAN SCHAAF

- mTor is overactive in SYS
- Rapamycin reduces mTor activity
- Improvements in strength and stamina were observed in a mouse model when given rapamycin.
- Additional research is needed before beginning a clinical trial in SYS.



2019 TARGETING SMCHD1
DR. MARNIE BLEWITT

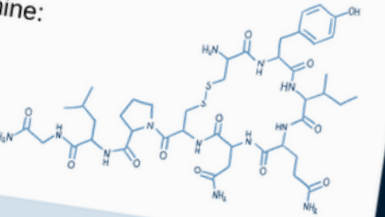
- A protein called SMCHD1 keeps many genes in their sleeping state
- Deleting SMCHD1 turns on silenced PWS genes (including MAGEL2)
- Early research suggests this could be a safe and effective therapeutic target
- Additional research is underway to determine if removing SMCHD1 may improve outcomes for people with PWS and SYS




NEWLY FUNDED!
2021 THE ROLE OF OXYTOCIN RECEPTOR- EXPRESSING ASTROCYTES IN SYS AND PWS
DR. CHRISTIAN SCHAAF

Specialized brain 'support' cells, called astrocytes, are responsive to oxytocin. In this project we will examine:

- How are astrocytes different in SYS and PWS from the typical brain?
- Could oxytocin treatment improve cognitive and social aspects of PWS and SYS?



YOU CAN HELP MAKE PROGRESS HAPPEN!
Would you like to be part of our team of movers and shakers who are making a difference in the lives of our loved ones with SYS? Send our team an email (info@fpwr.org) and we can help you get started!



2021

\$324,000



awarded to MAGEL2 & SYS research

3
Projects

2
Countries

3
Publications



MAGEL2 Research Awards 2021



The Role of Oxytocin Receptor-Expressing Astrocytes in Schaaf-Yang and Prader-Willi Syndromes. Christian Schaaf, MD PhD, University Hospital Heidelberg.

Comparative Behavioral and Proteomic Analysis Of Rat SNRPRN and MAGEL2 Models. Rodney Samaco, PhD, Baylor College of Medicine.

Orphan GPCRs and the Neurobiology Of Hyperphagia In Prader Willi Syndrome: Role Of GPR160. Gina Yosten, PhD, Saint Louis University.