

Supporting Families to Increase Inclusion and Belonging Working with our regional RTS Support Group to Empower Families

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What is Rubinstein-Taybi Syndrome?

- Rubinstein -Taybi Syndrome (RTS)
- Identified by Dr. Jack Rubinstein and Dr. Hooshang Taybi in the 1960s as "broad-thumb hallux syndrome", later renamed RTS



Dr. Taybi (left) and Dr. Rubinstein at a conference

- Rare genetic syndrome that affects approximately 1 in 100,000 to 125,000 newborns each year
- 50-60% of all cases mutations in CREBBP gene on chromosome 16p13 (RTS Type 1)
- 3-8% of cases mutations on EP300 gene on chromosome 22 (RTS Type 2)



Sawyer (RTS Type 1) is all dressed up and ready to go!



Myliege, "Myles" (RTS Type 2) is ready for school!

- RTS Type 1 Physical Characteristics
 - Large, angular thumbs and broad first toes
 - Narrow palate & angular nose
 - Small stature
 - Downward slanting eyes; thick hair and eyelashes
 - Intellectual disability & behavioral challenges are common
 - Various medical complications throughout lifespan



Example of large, angular thumbs



Example of broad first toe

Skill	Range for typically Developing Children (months)	Range for Children with RTS (months)	Average for Children with RTS (months)
Roll Over	2-5	2-24	7
Sit Up	5-8	6-30	11
Crawl	7-10	8-30	15
First Word	9-13	6-57	25
Walk	11-15	15-54	30
Potty-Trained	24-27	30-216	63
Word Phrases	14-24	24-156	65
Ride Bicycles	36-48	42-246	68

Developmental Milestones & Skills Chart from "Understanding Rubinstein-Taybi Syndrome"

RTS Support Group

- The regional Ohio – Kentucky – Indiana Rubinstein - Taybi Syndrome Support Group (OKI-RTS) has been supported by the Divisions of Developmental and Behavioral Pediatrics (DDBP) at the Cincinnati Children's Hospital (CCHMC) since the late 1990s
- Started with quarterly, two hour-long lunchtime meetings on Saturdays at DDBP with medical provider presentations and for socialization with other RTS families.
- Basic childcare provided for children/adolescents with RTS so parents/caregivers have time with other adults and medical providers.
- Why?
 - Challenges in getting RTS Diagnosis
 - Limited knowledge about RTS
 - Families often feel alone
- Despite positive feedback on meetings, attendance dwindled over time
- Revamping needed to be done for the RTS support group, taking a family-centered, individual empowerment and community focus approach



2018 RTS event at the Cincinnati Art Museum

- The RTS support group shifted their quarterly meeting to all-day events, within the community, keeping medical presentations and adding community-based information (e.g. financial planning, guardianship, etc.)
- Outreach expanded by disseminating information via Facebook pages, webpages, listservs, etc.
- Attendance at events increased and families were willing to participate in community-based events



The Kamalmaz family at the 2018 Family Support Group Museum Event

- Family leaders began to emerge and connect to the work and purpose of the support group

Inclusion and Belonging

RTS-OKI FAMILY IMPACT

- Broaden vision of a life in the community for their loved ones
- Empower families to participate in planning events
- Families take on leadership roles as presenters



The Walczak family on RTS 2018 World Day



Mia enjoying her time at the Gorman Farm event 2018

- Families organically initiating group projects like World RTS Day T-Shirts design and dissemination
- Opportunities created for families to participate in research activities both as part of research team and as participants
- Families changed their vision of these events and felt empowered to include siblings and extended family in their RTS-OKI experiences



Kiara and Annika work together on an art activity at the 2018 RTS Zoo event

Inclusion and Belonging cont.'d

RTS-OKI COMMUNITY IMPACT

- Increase in number of families involved and engaged in activities and projects
- Increase in regional reach and distance traveled by families to participate
- Increase in the resources available to families (i.e. RTS booklet, webpage, and medical guidelines)



David with his sister Cynthia at the RTS Art Museum Event 2018

What's Next?

- Release of RTS booklet "Understanding Rubinstein Taybi Syndrome: A guide for families and professionals"
- Development & dissemination of educational RTS video modules for family members and professionals
- Research study on needs of families of children & adults with RTS to improve positive life outcomes
- Update of RTS medical guidelines
- Continuation of community-based RTS meetings and activities



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Thank you to all the families who engage with us, take risks, challenge us and are involved in the process of moving to greater inclusion and belonging to increase positive outcomes for children and adults with RTS.