

Understanding Rubinstein-Taybi Syndrome:

A GUIDE FOR FAMILIES AND PROFESSIONALS



Emma enjoying the sun on the playground

This booklet has been made possible by the generous support of the Dr. Jack Rubinstein Foundation.





From left to right: Rhett, Levi, Ryan-Dylan, Lexi, Madison, David and Myliege

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A GUIDE FOR FAMILIES AND PROFESSIONALS



*This booklet has been made possible by the generous
support of the Dr. Jack Rubinstein Foundation.*

*This booklet is dedicated to Dr. Jack Rubinstein
who had a lifelong passion for understanding
and improving the lives of children and adults with
Rubinstein-Taybi syndrome (RTS) and their families.*

Table of Contents

Introduction..... 6

The History of Rubinstein-Taybi Syndrome (RTS)..... 7

RTS Diagnosis 8

Developmental Milestones 11

 Cognitive Development 11

 Physical and Motor Development..... 13

 Communication and Language Development..... 13

 Social and Emotional Development..... 14

Care Coordination and Navigation..... 17

What to Expect During Each Life Stage 18

 Infancy..... 18

 Early Childhood..... 26

 School Age 30

 Adolescence..... 35

 Transitioning to Adulthood 39

 Adulthood and Aging..... 43

What to Expect Medically..... 49

 Anesthesiology 49

 Cardiology 49

 Dermatology 50

 Gastroenterology 50

 Genitourinary 50

 Neurology 51

 Oncology and Hematology..... 51

 Ophthalmology 52

 Orthopedics 52

 Respiratory/Ear, Nose and Throat..... 52

Caregiving and Respite 57

Conclusion 60

References 61



*Dr. Wiley and Hamza's family
at the museum*

Introduction

The goal of this booklet is to provide families with accurate, up-to-date information about how to support their child with Rubinstein-Taybi syndrome (RTS) and to equip them with the tools to educate family members, friends, colleagues, professionals, and the general public about the syndrome.

The stories included in this booklet are meant to emphasize the beauty and joy of sharing life with an individual with RTS. We want families to feel empowered to anticipate what to watch for and attend to, in order to create a healthy, happy, safe, and productive life for their loved one in the community.

This RTS information booklet was a collaborative effort of the University of Cincinnati Center for Excellence in Developmental Disabilities (UCCEDD), the Division of Developmental and Behavioral Pediatrics and the Division of Genetics at Cincinnati Children's Hospital Medical Center (CCHMC) and the Rubinstein-Taybi Syndrome Ohio-Kentucky-Indiana (RTS OKI) Family Support Group. Huge thanks go out to the physicians at CCHMC who have expertise working with individuals with RTS and have been an invaluable resource in the process of creating and editing this booklet. Another big thank you goes out to all the individuals with RTS and their families. Each of them gave personal time and provided valuable input, allowing us to share this important and relevant information in an easily accessible and understandable way.

The History of RTS

Dr. Jack H. Rubinstein, a developmental pediatrician at the University of Cincinnati (UC) Medical Center, and Dr. Hooshang Taybi, a pediatric radiologist at the University of Oklahoma, were introduced by a mutual colleague who had noticed they were both observing unrelated children with similar physical and intellectual disabilities. Upon meeting, the two decided to combine their efforts and research. Together in the early 1960's the pair identified the characteristics that make up what was originally named "broad thumb-hallux syndrome" and is now called Rubinstein-Taybi syndrome (RTS).

Rubinstein-Taybi syndrome (RTS) is a rare genetic condition that affects approximately 1 in 100,000 to 125,000 newborns each year worldwide^{1,2,3,4}. In 50–60% of cases, it is the result of mutations in the CREBBP gene on chromosome 16p13^{1,2,3,4}, also referred to as *RTS Type 1*. Individuals with this form of RTS have many of the following physical characteristics: large, angular thumbs and broad first toes, a distinct angular nose, small stature, thick hair and eyelashes, downward slanting eyes and a narrow palate^{1,2,3,4}. Sometimes at birth, infants show a small reddish-pink mark on their forehead. Intellectual disability is common and can range from mild to severe^{1,2,3,4}.

An even rarer genetic variation that affects 3–8% of diagnosed individuals, occurs on the EP300 gene on chromosome 22^{4,5}. This is also referred to as *RTS Type 2*. These individuals have some of the same features seen in RTS Type 1, but facial features may be milder, thumbs are less likely to be angled outward, and intellectual disability may be milder in some cases².

Genetic testing, which evaluates the genes by sequencing and deletion testing, may confirm a diagnosis of RTS. In about 30% of cases, children with typical features of RTS do not have an identified mutation in CREBBP or EP300. In those individuals the cause of RTS is not yet known³.



Dr. Taybi and Dr. Rubinstein, respectively, at a conference

In about 30% of cases, children with typical features of RTS do not have an identified mutation in CREBBP or EP300. In those individuals the cause of RTS is not yet known³.



RTS Type 1

Sawyer is all dressed up and ready to go



RTS Type 2

Myliege, "Myles" is ready for school

RTS Diagnosis

The diagnosis of RTS can occur in different life stages, depending on the individual. Some families receive a diagnosis at birth when health issues arise. Others may show the physical signs of RTS, prompting physicians to investigate further, possibly through genetic testing.

During genetic testing, a blood sample is obtained from your child and sent to a laboratory offering the specific testing. Many geneticists will first order a test called genetic micro array on children with a suspected syndrome. If that testing is normal, the next step is to order specific genetic testing for the CREBBP and EP300 genes. Results can take several weeks to a few months to be returned. In most cases the results will be straightforward and confirm whether the child has the RTS diagnosis or not. Occasionally, however, testing will show Variants of Unknown Significance (VUS) which represent a "gray zone" and can be difficult to interpret². Your doctor or genetic counselor can help interpret these results for you.

The diagnosis for a relatively healthy child may not come until later in childhood or early adolescence^{1,2}. This may be especially true for children who have RTS Type 2, where the physical features may be more subtle and a diagnosis may only come after genetic testing is done^{1,3}.

Literature has shown that the average age of diagnosis is about 15 months old¹⁵; however, this may change as awareness increases and genetic testing becomes more common. Families can request a referral from their primary care doctor or pediatrician for genetic testing. Tests may not be covered under all insurance plans. Prior to scheduling an appointment with a geneticist, contact your insurance company to see if the testing is covered and, if not, discuss financial options. Families can also contact their local, state, and regional disability resources to explore if other funding options are available.

RTS can be difficult to diagnose, and at times may not be identified by even the most extensive genetic testing³. On some occasions, families may receive a “clinical diagnosis” based on the physical characteristics of the syndrome versus a medical diagnosis, which is confirmed through genetic testing^{6,7}.

After diagnosis, families often want to know what “caused” RTS in their child. Was it something that the parents did or didn’t do and are they likely to have additional children with RTS? An individual who does not have RTS cannot be a “carrier^{1,7}.” Based on current research, a typical parent has a less than 1% chance of having more than one child with RTS^{7,8}. This is because mutations on the genes and chromosomes that cause RTS happen “de novo,” meaning they are new and occur spontaneously^{1,2,5}. However, studies have shown that an individual who already has RTS has a 50% chance of passing the condition on to a biological child⁷.

Parents cope with the new diagnosis of RTS in many different ways⁹. Some parents experience grief after receiving the diagnosis. Others are relieved that they finally have a diagnosis. It is helpful for parents to reach out for support during this time. The families involved in the RTS OKI Support Group have found that connecting with other families and professionals who understand the challenges and beauty of spending life with someone who has a developmental disability is imperative. These families and professionals can provide information, resources, and tools. They can also help you learn about the services and supports that are available through your local healthcare, behavioral health, and developmental disability systems.

When researching your child’s RTS diagnosis, it is important to be aware that the information available in this booklet, online, or from other sources may or may not pertain to your child. While there are characteristics and conditions more common to individuals with RTS, each person is unique.



Example of large, angular thumbs



Example of broad first toe



Natalie and Morgan snuggling

Developmental Milestones

Individuals with RTS will have developmental delays, but this will look different for each child. Just like typically developing children, children with RTS can vary in the age at which they meet developmental milestones. The chart below shows common developmental milestones and the ages at which they typically occur^{1,10}.

Table 1. Developmental Milestones & Skills^{1,10}

Skill	Range for Typically Developing Children (mos)	Range for Children with RTS (mos)	Average for Children with RTS (mos)
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 Bicycle	36–48	42–246	68

Cognitive Development

A child’s cognitive development begins prior to birth and continues at a rapid pace during their early months and years^{11,12}. Early milestones in this area of development include tasks like following objects with their eyes or expressing the need for a change in activity or position by vocalizing or fussing. In later months, infants begin to recognize and differentiate between familiar faces or reach for objects and bring them to their mouths. Toddlers begin to understand that objects have names and they are able to copy behaviors, like waving. This type of cognitive development continues throughout early childhood and broadens into social, emotional and language development in childhood, adolescence, and adulthood^{11,12}.

There is currently no research that specifically explores learning, thinking, and problem-solving in individuals with RTS. We do know, however, that children with RTS experience varying degrees of delays in their cognitive development when compared to their typically developing peers. These delays can be assessed once the child is school age and may be identified as having an intellectual disability. The American Association on Intellectual and Developmental Disabilities (AAIDD) defines intellectual disability as, “characterized by significant limitations in both intellectual functioning and in adaptive behavior, which covers many everyday social and practical skills.” Children and adults with RTS may have limitations in intellectual functioning, such as reasoning, learning, and problem-solving^{1,6}. The impact of these delays is different for every child and every family. Because cognitive development begins early in life, it is best for those with RTS to seek intervention early, when the brain is most adaptable¹¹. The brain development that happens between birth and three years of age lays the groundwork for learning, behavior, and health. Over time, it becomes more difficult, but not impossible, to change the connections in the brain that create the paths for this development¹². Early Intervention (EI) services (typically birth–3) are provided in every state through such agencies as the Department of Health or Department of Developmental Disabilities.

Research-based organizations, like the Centers for Disease Control and Prevention (CDC), provide information on typical cognitive milestones (e.g., CDC’s “Learn the Signs. Act Early.” campaign)¹¹, leading to a better understanding of your child’s growth, needs, and progress over time. Talk to your child’s healthcare provider about expected milestones and suggested intervention services to improve developmental outcomes for your child¹¹. Early intervention, followed by school-based and community-based services, can support your child and teenager to reach their full potential.

Children and adults with RTS may have limitations in intellectual functioning, such as reasoning, learning, and problem-solving, as well as in adaptive behaviors, including everyday social and practical skills^{1,6}.

By learning about the cognitive development of individuals with RTS, and understanding your child's unique challenges as they grow, you can identify and seek out the most effective strategies to meet specific needs across their lifespan. No matter if they are a teen, a young adult or aging adult, each stage will bring new experiences and challenges for your loved one, as well as you, their caregiver, and family. Because adults with RTS may experience a decline in cognitive ability as they age, they may benefit from specific interventions and supports to address this^{1,2,13}.

Physical and Motor Development

Most children with RTS will have some physical and motor development delays, often due to low muscle tone^{1,6}. This, in conjunction with intellectual disability, may affect things like balance and control, which can impact rolling over, sitting up, walking, feeding, toileting, and self-care¹⁰.

Many families seek out early intervention services, such as therapy and play-based interventions to promote their child's development^{6,11}. Families can look for these options themselves and can also talk to their pediatrician or physician about the types of services for which they may need a referral. Early Intervention services allow your family to set goals and provide you with ways to reinforce these goals through activities and play in the community. As your child progresses, he/she should grow in self-determination and want to achieve the goals set out for him/her.

Communication and Language Development

Over 90% of children born with RTS experience substantial speech delays^{1,6,14,15}. One early sign of a potential speech delay may be if a child has difficulty eating. When a child is eating, there is a certain coordination of the mouth and tongue, which is also needed when trying to make speech sounds. If your child is experiencing a language delay, their speech is not easy to understand, or they are non-verbal, start a conversation with your pediatrician or physician early.



*Jason showing off his
Pokemon tattoo*



Cooper and his mom using an iPad to communicate

They may be able to provide a referral for a speech-language assessment to identify the best ways to support your child.

Based on the outcome of the assessment, support recommendations may include: speech therapy, sign language, picture communication boards, Picture Exchange Communication System (PECS), assistive technology, and/or augmentative communication devices. These supports could be provided by the school, in the community, or both. Getting your child involved in speech and language supports may help develop appropriate and preferred communication methods, and may help minimize future communication issues¹⁶.

Supports and therapies are often continued through school age and adolescence. Goals may be adapted to address new stages of development or areas of concern. Families may work with speech, communication, and assistive technology specialists at school and in the community, in addition to working with private therapists on the approaches that best support their child's growth. It is beneficial to keep private and school-based therapists informed of one another's work. This can be done by signing a release so they can collaborate and create informed therapy plans which allow for a focused effort to increase impact.

Social and Emotional Development

Some studies show that children with RTS are more socially and emotionally developed than their typically developing peers. In one study¹⁷, children with RTS performed better than their typically developing peers in the areas of direct physical contact and social interest. Even more specifically, this research showed that children with RTS scored higher in other related areas, like need for physical touch, keeping eye contact while speaking, seeking emotional comfort when upset, initiating playtime with others, expression of various emotions, and looking up when being spoken to. Most children with RTS do best when you have their attention and make eye contact. They may need gentle physical contact to encourage focus and attention¹⁷.

Sometimes, children and adults with RTS express their emotions or physical discomfort through acting out^{1,15,17}. These behaviors are often triggered by frustration with the inability to communicate their wants, a need for attention, a response to doing something they do not like, etc. Behavior issues may occur during life changes, (e.g. different schedule, new caregiver) as well as with medical problems.

Many developmental professionals, including mental health counselors and psychologists, specialize in treating challenging behaviors. The approaches to addressing challenging behaviors may include evaluations by a professional, such as a Functional Behavior Assessment (FBA) to determine the cause of the undesired behavior and a Behavior Intervention Plan (BIP) to identify the desired behavior and the steps necessary to achieve it. You can request evaluations from school as part of your child's Individualized Education Plan (IEP), in the community by a local mental health specialist, or both.

Behaviors common in individuals with RTS can be similar, but are not limited to, those behaviors associated with Obsessive Compulsive Disorder (OCD) and Autism Spectrum Disorder (ASD)^{1,6,13,17}. Such behaviors may include self-stimulation or repetitive motions (e.g. flapping hands, rocking body, head shaking, etc.), hyper-focus or getting “stuck” on certain things, or needing a routine and struggling when the routine is not implemented. It is helpful to address these behaviors early with a mental health provider, physician, psychologist, or a Board Certified Behavioral Analyst (BCBA), especially when the behavior is unsafe and/or interfering with day-to-day activities. Research shows that behavior issues in individuals with RTS can increase with age, so it is recommended to continue intervention if the behaviors return or there are new behaviors^{13,17,18}.

Most children with RTS do best when you have their attention and make eye contact. They may need gentle physical contact to encourage focus and attention¹⁷.



Hannah getting a check-up at the doctor's office

Care Coordination and Navigation

Life at any stage can be complicated! Between juggling doctors' appointments, school schedules, monitoring feeding, or even getting enough sleep, it all can seem overwhelming. In spite of this, it is crucial to identify a formal or informal care coordination system to minimize stress. No matter what stage you're in now, it is never too late to start this process.

Healthcare systems, especially in pediatrics, are developing care coordination plans to offer more family centered care^{19,20}. A care coordination plan allows a group of different doctors and specialists to work together to meet the needs of the individual, using the best practices known. This team approach helps your family navigate multiple providers in various departments and settings, with support from a primary care coordinator²⁰. With this empowering approach to care, families are treated as part of the medical decision-making team, and are better able to advocate for their loved one's needs.

A personal care notebook is an important part of a care coordination plan. The notebook provides a system for tracking the care of an individual. If a care coordination plan is not offered to you by your doctor, you can use the care notebook to help you act as the individual's care coordinator. Care notebook templates are usually free in print and online. One helpful resource in this area is Guiding People Through Systems²¹, a website that offers tools, resources, and information on care coordination plans and care notebooks. You may also find it helpful to search key words or phrases on the internet such as "medical care notebooks" for additional resources and templates. It is recommended that you find one that best fits your style and needs, so you can maintain it easily, and bring it to all of your child's appointments. This will ensure that all doctors, therapists, and other clinicians on the individual's team are on the same page when it comes to caring for and treating them.

A care coordination plan allows a group of different doctors and specialists to work together to meet the needs of the individual, using the best practices known.

What to Expect During Each Life Stage

**Note: the medically related items in the Life Stage sections are described in more detail in the “What to Expect Medically” section later in this booklet.*

Infancy

Welcoming a new baby in your home is a very special time! You are probably feeling happy and excited, while also anticipating less sleep and maybe some struggles ahead. It might feel a bit overwhelming when you add the unknown special healthcare needs or developmental disabilities of your child with RTS. During this time, it is important to create a support system of professionals and loved ones who can care for you so that you can meet the needs of your newest family member.

It is common for infants with RTS to have low muscle tone. Because of this, they may have difficulty with feeding, including sucking, drinking, and advancing to baby and table foods. This may indicate dysphagia, a motor coordination problem of the mouth and swallowing muscles^{1,6,7,14}. Sometimes an infant will choke or gag, which might mean that food or formula has gone down the wrong way and gotten into the lungs. This is called aspiration. Frequent aspiration can cause injury to the lungs, sometimes leading to pneumonia and respiratory problems^{1,2,6}. Talk to your pediatrician or physician immediately if you start to see these issues.

Some children with RTS have a smaller jaw and/or narrow palate, which at times leads to difficulty latching on and swallowing when breastfeeding, but breastfeeding is still possible⁶. If you would like to breastfeed, it is helpful to talk to a lactation consultant and your child’s physician. Professionals can take your child’s specific needs into consideration and help develop a breastfeeding regimen. Pumping and/or bottle-feeding are also options. Those who bottle-feed may need to try different nipples and

It is common for infants with RTS to have low muscle tone. Because of this, infants with RTS may have difficulty with feeding, including sucking, drinking, and advancing to baby and table foods.

ways to hold their baby when feeding. It is important for you to determine, with proper guidance and support from professionals, what is best for you and your baby.

Though infants with RTS are generally of typical birth weight, sometimes, due to feeding difficulties, they can lose weight or have difficulty gaining weight. They may also not grow in height at the same rate as typically developing infants^{1,23,24}. Your pediatrician will monitor your baby's growth, but if you are concerned, talk to them and share the RTS growth charts included in this booklet. Sometimes a feeding tube is recommended for these babies to be able to get enough nutrition. Conversations with your physician and/or specialist (geneticist or gastroenterologist) can help you make an informed decision about what is right for your child⁶.

Children with RTS often have differently structured airways^{1,6,7,14}. As a result, respiratory infections can sometimes turn into something more serious, like pneumonia. It is important to keep a close eye on your child when they have a respiratory infection or when considering anesthesia (See Anesthesiology section). Because of the structure of their airway, if your infant has repeated episodes of difficulty breathing or persistent wheezing, it is recommended that you speak with your pediatrician about reflux or possible aspiration⁶.

Some parents report that their baby with RTS is quiet, sleeps often, and for long periods of time. This can often be seen as a blessing, but it is important to monitor when your child is sleeping and eating, especially if the baby weighs less than they did when they were born. Many infants with RTS sleep much of the day and night. However, if your infant is not gaining weight, you may need to wake him/her up for feeding. Check with your child's pediatrician on how much time your infant should be awake based on his/her age.

Testicular issues are common in boys with RTS^{1,6,14}. Often, one or both testicles do not descend properly. If your son's testicle(s) are not descended at birth or shortly after, consult with your child's pediatrician, who can continue to

monitor this. If your son's testicle(s) do not descend into the scrotum (sac) between 6 and 12 months, make sure to bring this up again. Surgery by a urologist may be required to bring the testicles down into the scrotum. This is usually corrected without complications, but work with your pediatrician and specialist to determine when it is medically safe for your child to have anesthesia.

Just like with any infant or typically developing child, you want to work closely with your child's pediatrician to monitor how they are progressing toward the expected developmental milestones. Each child with RTS can vary in the age at which they meet these. If necessary, the pediatrician can help you access appropriate resources and possible interventions and therapies to keep your child on track.

Parent Tips

- Use therapeutic sippy cups or straws that are designed to reduce dysphagia. Liquids are released more slowly than in typical cups in order to reduce the risk of aspiration.
- Quickening, fast flow, or "y-shaped" nipples make latching on easier for bottle-fed babies.
- Using sign language while you speak can help with language development.
- You might notice that your child is oversensitive to auditory stimulations. If loud noise or "white noise" starts to bother your child to the point of anger or tears, using noise-canceling headphones might do the trick.
- Monitor developmental milestones by using the CDC's Act Early website. It's truly family friendly and has great information.
- Create community connections. Being part of both typical and special needs playgroups, parent groups and community groups is important for kids to socialize and parents to make connections, making families more resilient.
- Introduce educational and sensory toys to encourage development and self-soothing skills.
- Continue with annual check-ins with any specialist you have seen for diagnosed medical concerns, like cardiology, to be sure nothing new arises.

(These tips are shared from other parents of children with RTS. Always consult with your child's pediatrician or specialist prior to implementing these tips.)

Meet Morgan

He is 21 months old and lives with his mom and dad in Maryland.



Morgan and his dad enjoying some special one-on-one time

Morgan is an only child, but loves to visit his aunts, uncles, and cousins. Even though some days are a blur of doctors' appointments and therapies, it's nice to just take him on a walk or to run a simple errand. We also enjoy story time at the library or bookstore. Above all, my husband and I just enjoy doing our everyday routine with Morgan. My husband works from home, so a few times a day Morgan and I "visit daddy" at work. I enjoy playing with Morgan and making him laugh. He brings us so much joy and encouragement each day.

Morgan has taught us to appreciate the smallest of victories. Every new little skill that he learns brings us a bit of hope. It is easy to take things for granted in life, and he helps us to enjoy each day. Morgan teaches us patience and perseverance, and we've learned to be content with where he is presently. Even though much is unknown about his future—when he'll walk or even speak, he's a happy baby and that keeps us going.

Through all the sickness, surgeries, and sleepless nights, Morgan smiles and laughs through it all. That amazes us!

To newly diagnosed families, our advice is to seek a support group, whether that be an RTS specific group or other families that you can share your concerns, fears, and joys with. It can be lonely and scary at times to have a child with a rare disorder, especially when doctors haven't even heard of RTS before.

Finding people who support you and your child make it a little more manageable. Also, don't forget to take care of yourself! Parents often get so caught up in appointments, therapies, and the day-to-day that we often forget to take care of our mental and physical health.

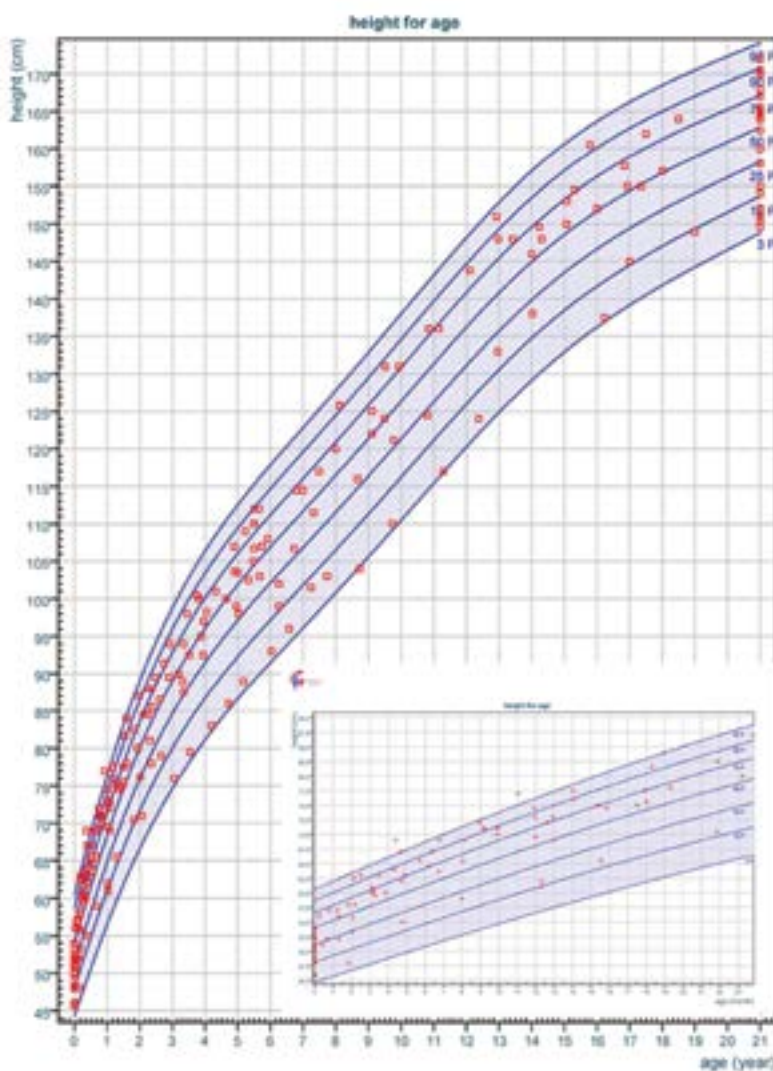
Don't forget to treat yourself sometimes!

—Natalie, Morgan's mom

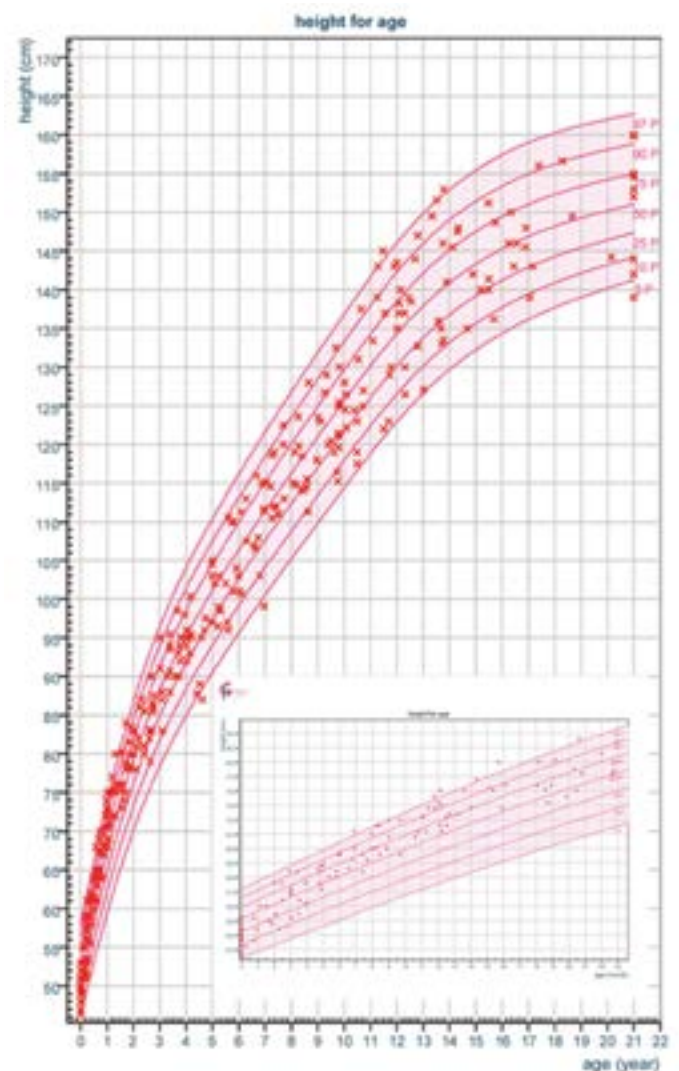
RTS Growth Charts

There are two growth charts for individuals with RTS worth mentioning. The growth charts from “Growth in the Rubinstein-Taybi syndrome”, Stevens et al, (1990) used typical growth data from the 1970’s, compared to 50 individuals with RTS from the United States and 45 from the Netherlands²². The diagnosis of RTS in this study was confirmed by clinical diagnosis, but did not include genetic confirmation. It is important to note that growth charts have been updated based on changes in health and cultural differences. For example, The Centers for Disease Control (CDC) updated growth charts in 2000 for typically developing individuals in the United States.

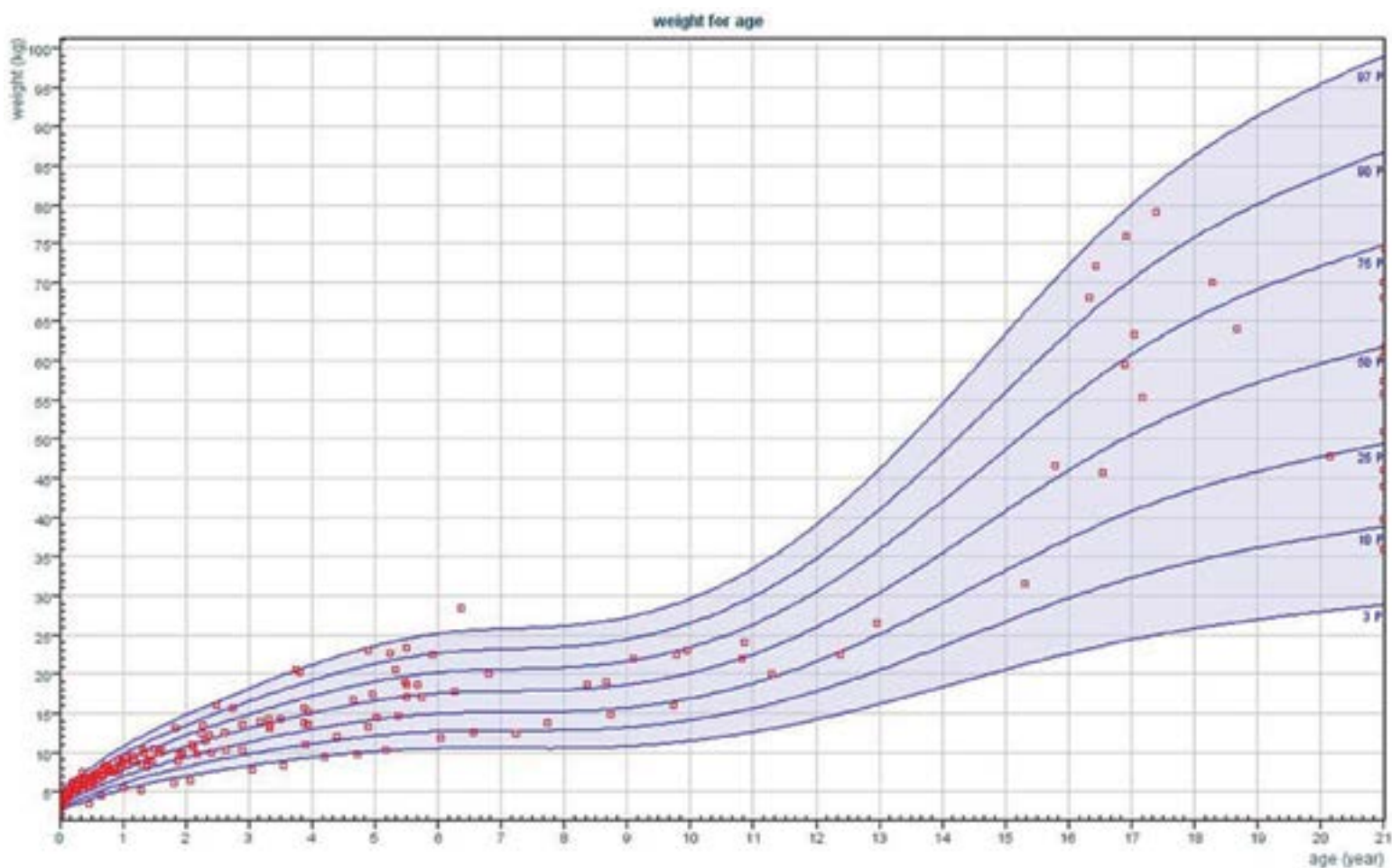
The second set of charts published in “Growth Charts for Individuals with Rubinstein-Taybi syndrome”, Beets et al, (2014) were formulated with individuals from a variety of countries. They included only individuals with confirmed diagnosis by genetic evaluation (clinical and/or molecular). The charts are shown here²³.



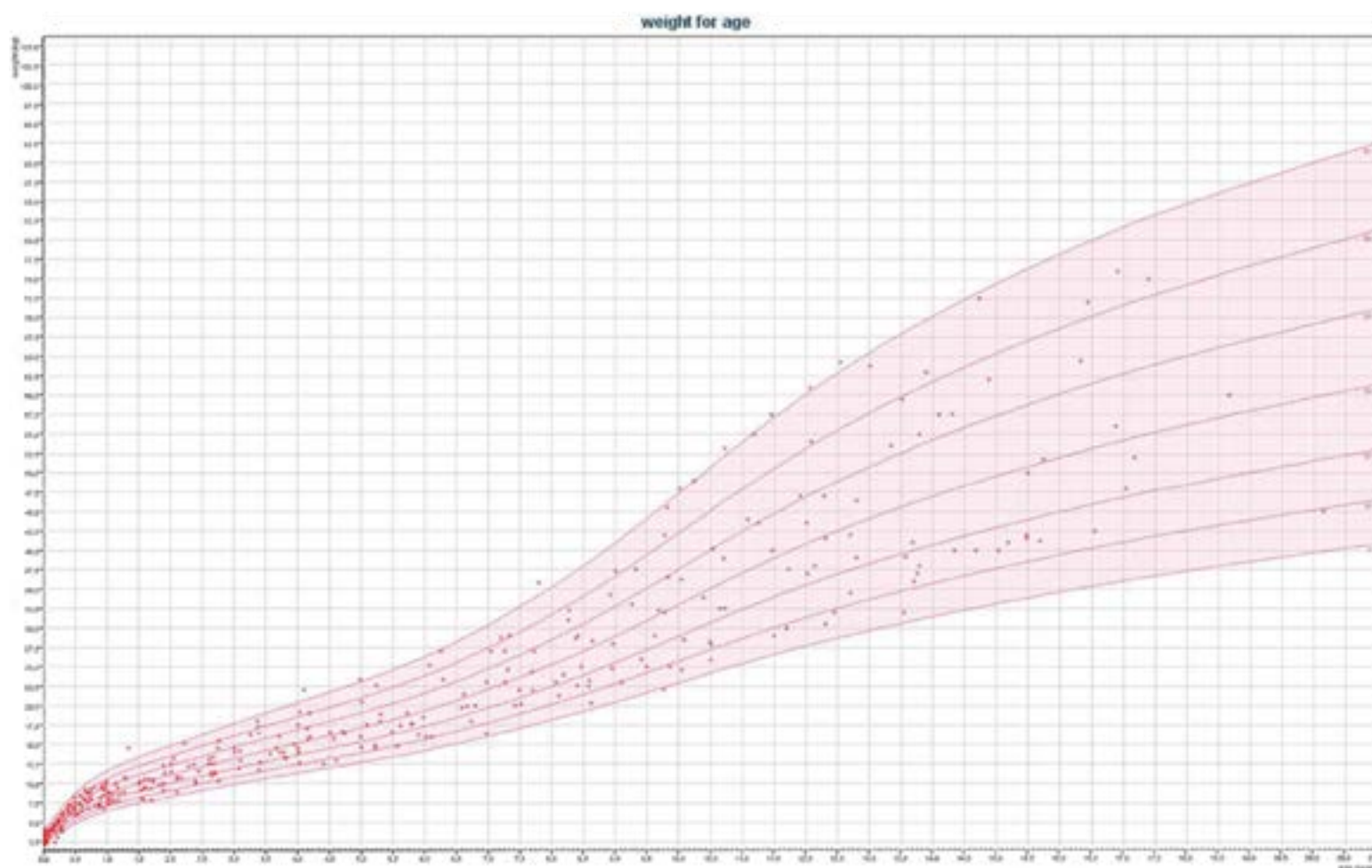
Growth in height for boys with RTS, ages 0–21 years, showing individual data points. The inset shows an enlarged graph of length/height for boys 0–24 months of age²³.



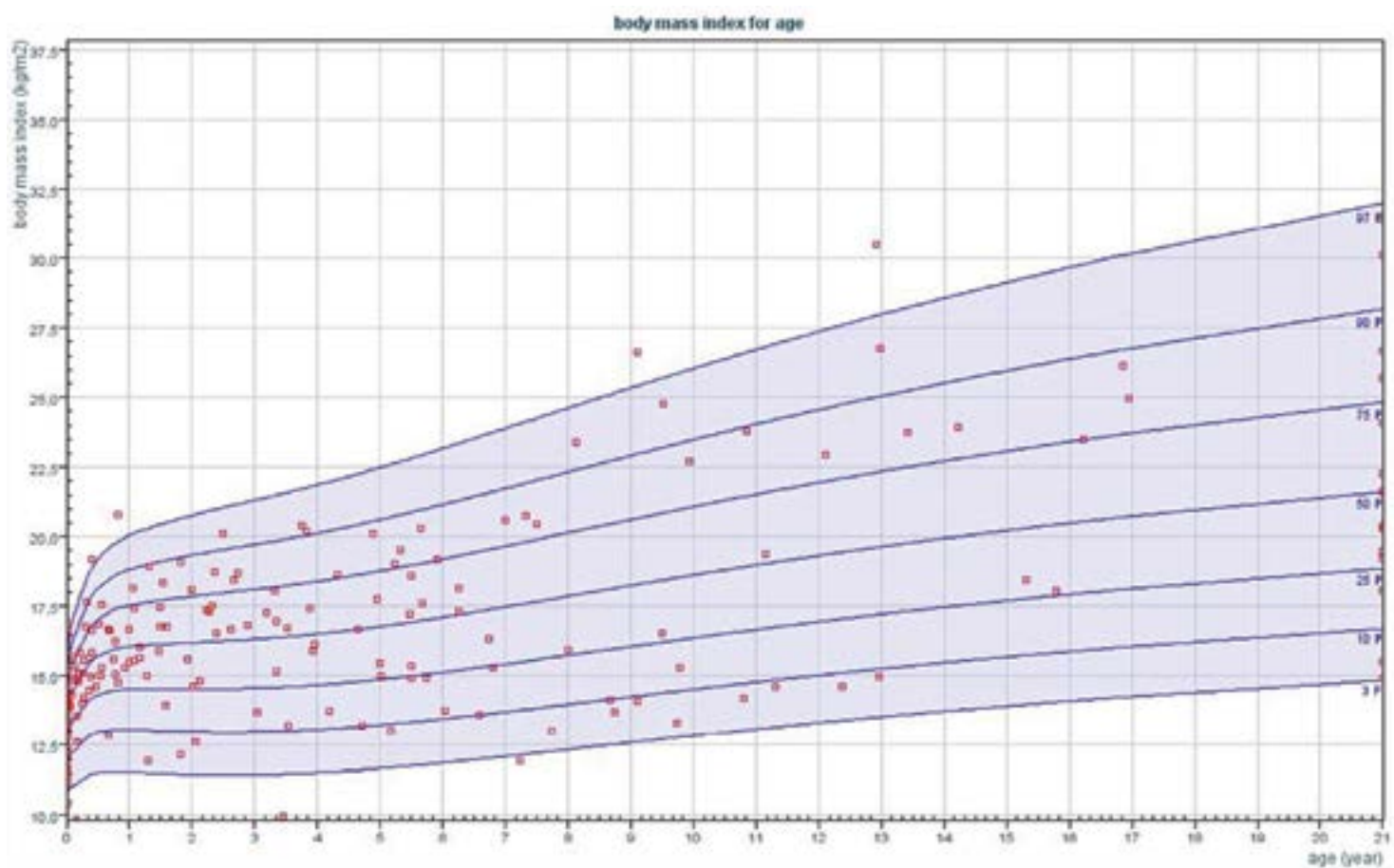
Growth in height for girls with RTS, ages 0–21 years, showing individual data points. The inset shows an enlarged graph of length/height for girls 0–24 months of age²³.



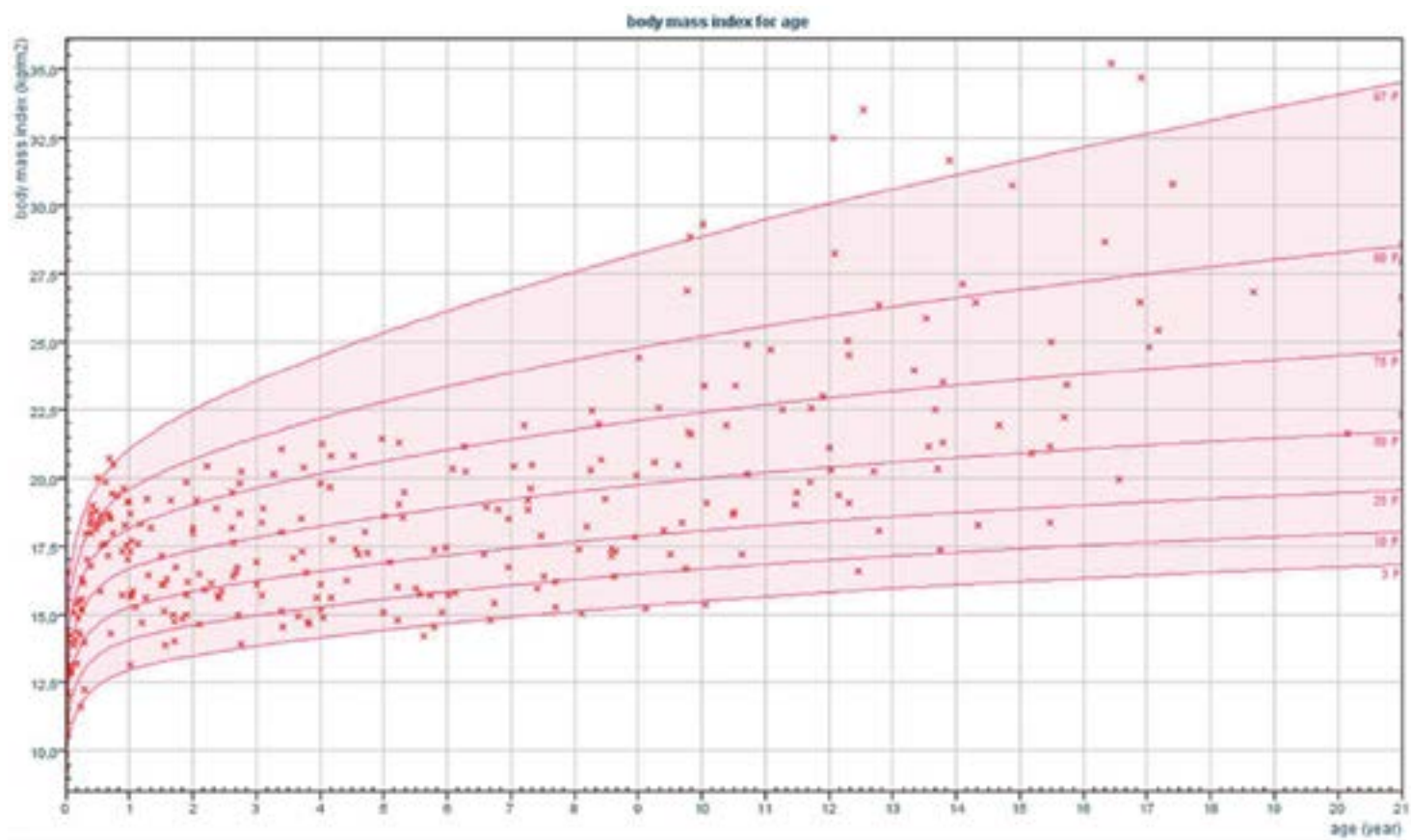
Weight in males with RTS, ages 0–21 years, showing individual data points²³.



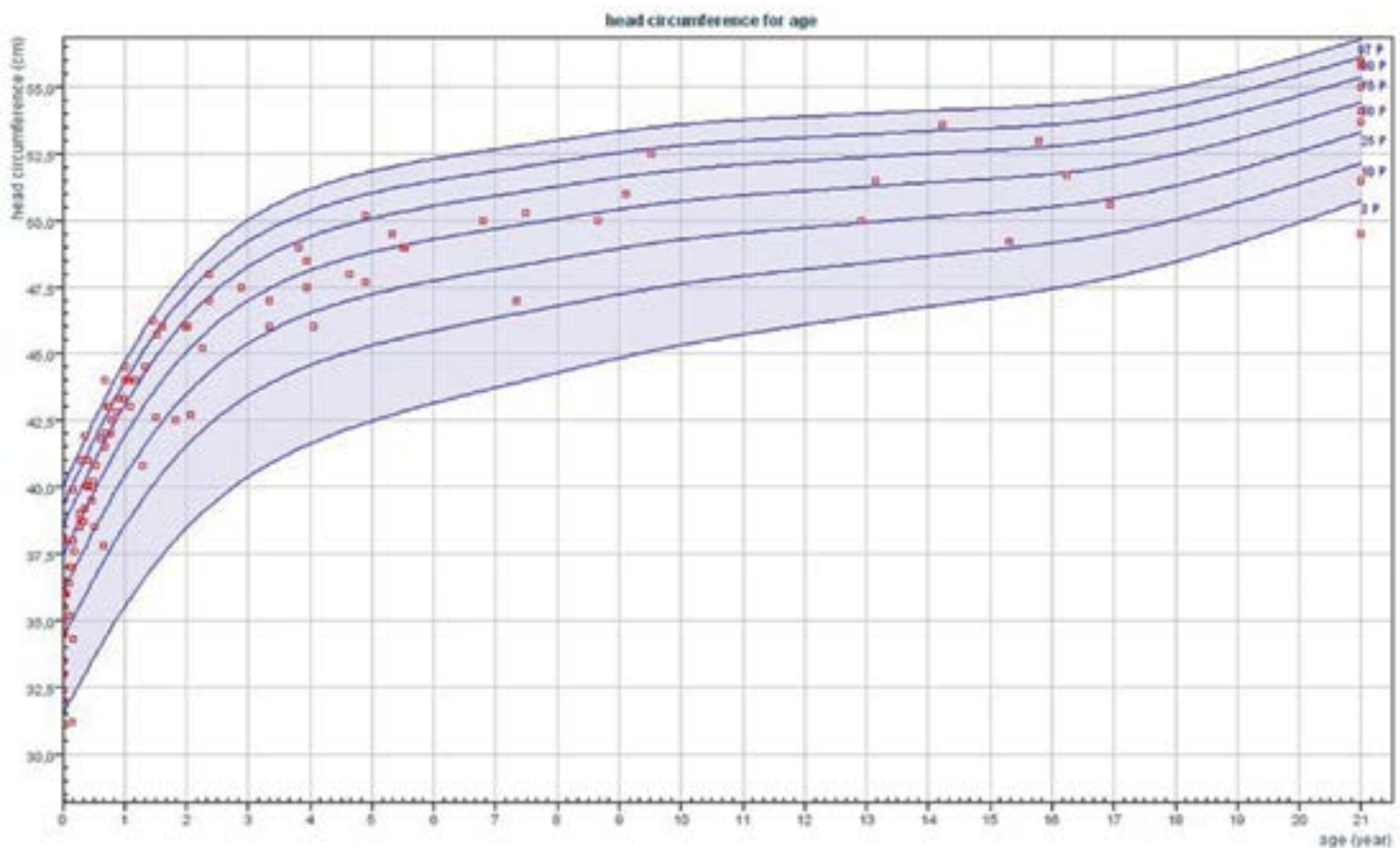
Weight in females with RTS, ages 0–21 years, showing individual data points²³.



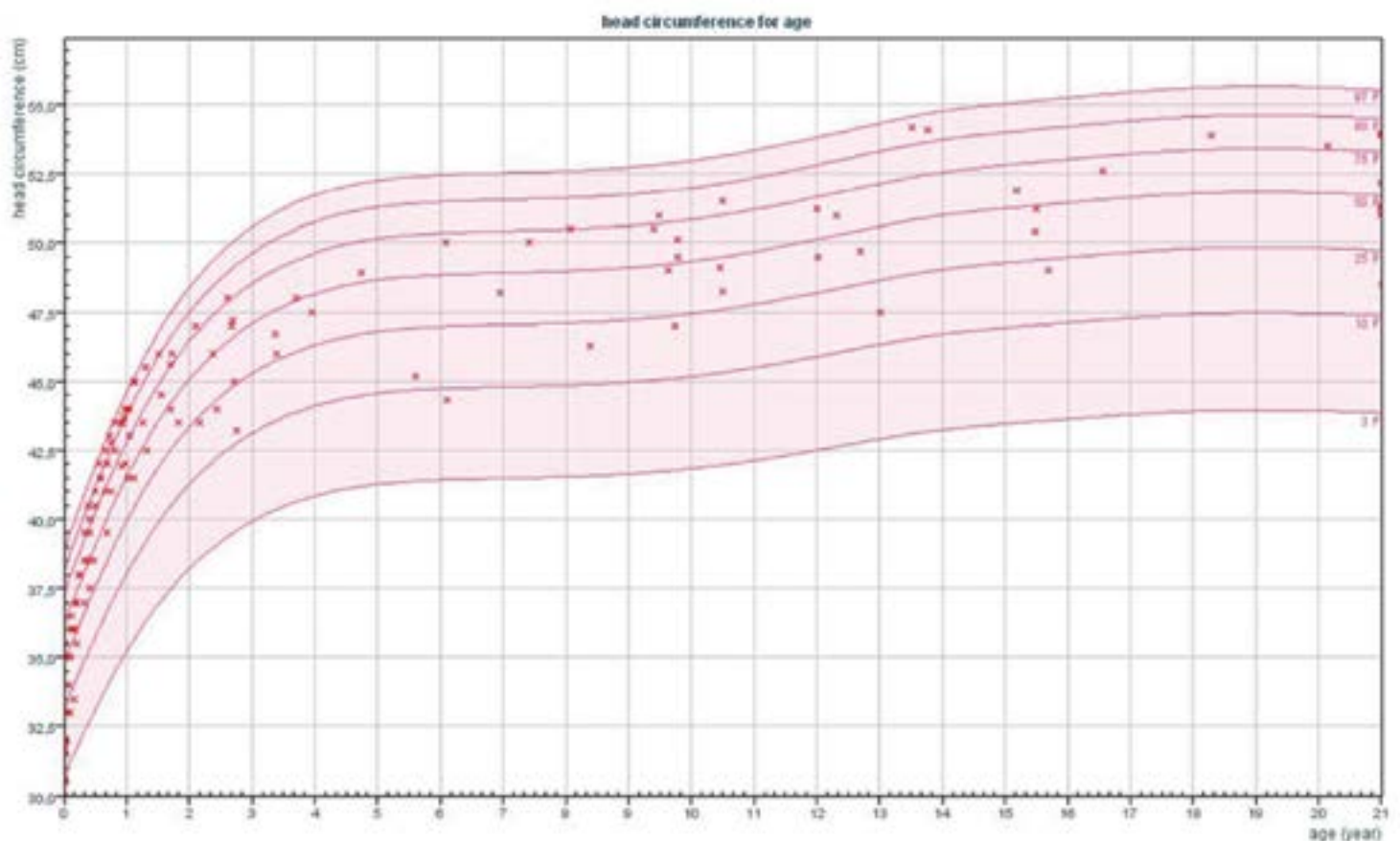
BMI in males with RTS, ages 0–21 years, showing individual data points²³.



BMI in females with RTS, ages 0–21 years, showing individual data points²³.



Head circumference in males with RTS, ages 0–21 years, showing individual data points²³.



Head circumference in females with RTS, ages 0–21 years, showing individual data points²³.

Syndrome specific growth charts, when used in comparison to those of typically developing children, are helpful in understanding if a child is making appropriate gains in areas such as height and weight, keeping in mind their genetic condition. If your child is not tracking along an appropriate growth trajectory, your doctor can use these to help guide expanded evaluations of medical and social conditions that may contribute to your child’s difficulty with growth.



Hannah smiling as she awaits a visit from her doctor at Cincinnati Children's

Early Childhood

This is the time when you will start to see your child's personality start to take shape. You may see fewer issues with eating and swallowing, but could see some new health issues. It is important to remain in touch with your doctors and specialists to ensure that your child is happy and healthy.

Healthy sleep is important for growth and development. Sleep issues, such as loud snoring, restless sleep, and abnormal breathing might occur at this age^{6,15}. Consult your physician if your child experiences these sleep issues, as they could be a sign of sleep apnea (an obstruction of the airway during sleep). Ongoing breathing obstruction can lead to high blood pressure and hypertension issues^{6,15}. If your child is diagnosed with sleep apnea, their physician should monitor their blood pressure regularly.

A common occurrence in individuals with RTS are keloids, or excess scar tissue²⁴. Keloids are large reddish-pinkish bumps that typically show up on the chest and back, but can appear any place where there is irritated skin. The skin could be irritated as a result of something big like a surgery, or as small as an insect bite. Keloids are benign (non-cancerous) but can be itchy and uncomfortable²⁴. Consult your physician or dermatologist for treatment options to reduce the effect keloids have on your child.

Constipation can be an issue for individuals with RTS of all ages, but especially in childhood and adolescence^{1,2,6,14,15}. It is helpful to talk to your physician or specialist about the options available to aid in this issue. Your child may communicate their discomfort in different ways, including behavior. It is important, especially after they are toilet trained, to develop discreet and respectful ways to monitor and track toileting. Some doctors might recommend a special diet high in fiber or specific medications (e.g. laxatives) that could help avoid or relieve constipation^{1,2,6,14,15}.

A tethered cord, which happens when there is stretching and tension of the spinal cord, can occur in children with RTS^{6,14,15}. Problems associated with this tend to occur

Both males and females with RTS have a tendency to gain excess weight, starting at an early age into the teenage years. It is important for your child to eat a balanced diet and stay active.

during periods of rapid growth²⁵. Signs of a potentially tethered cord include: severe or worse than normal constipation; toileting issues after a child has been potty trained; difficulty sitting upright; changes in how your child walks (crouched gait, tip toes, trouble with balance), and lower back and leg pain. To determine if a child has a tethered cord, doctors will typically do an MRI of the lower spine with cinematography (ability to see if the cord moves). Sometimes the scans are inconclusive and other testing is helpful. This may include urodynamic testing, which assesses how the bladder and urethra are performing their job of storing and releasing urine. After testing is complete, if there is concern that your child has a tethered cord, it might be helpful to see a pediatric neurosurgeon with experience in this area^{25,26}.



*Griffin visiting
Cincinnati Children's*

Parent Tips

- Discuss any breathing issues and sleep apnea with your pediatrician. He/she may recommend having tonsils and/or adenoids removed.
- Share early and often with your pediatrician and specialists the concerns about children with RTS and anesthesia. “Rubinstein-Taybi syndrome medical guidelines” by Wiley, Swayne, Rubinstein, Lanphear and Stevens (2003) is a recommended professional reference.
- Begin to help your child learn how to make friends and negotiate social situations. Work to build skills that will help them succeed in preschool.
- Foster relationships with typical peers, too. Having friends who model milestones, academic goals, and proper behavior is really helpful. Making sure your child can manage the “playground curriculum” is as important as academics, at times.
- Have expectations like helping with simple chores and self-care to increase their success in school and their community. Our daughter loves helping and we see a real difference in her trying hard things because she believes she is capable.
- Manage behavior and emotional outbursts with clear and consistent boundaries. Do not lower your expectations of appropriate behavior from that of their siblings or typical peers. Don’t hesitate to seek help from developmental and behavioral specialists!

(These tips are shared from other parents of children with RTS. Always consult with your child's pediatrician or specialist prior to implementing these tips.)

Meet Jaymin

*Jaymin is 6 and a half years old
and lives with his mom and dad.*



Jaymin enjoying the freedom of a fall day

Jaymin was a healthy baby boy with an amazing head of jet black hair. We noticed his angled thumbs right away, but were told not to worry too much about it. The first three months were totally glorious—tough of course since we were new parents—but he was a great baby. He ate wonderfully, slept well, and was happy most of the time. He was a total delight.

At Jaymin's three month check-up, the pediatrician we usually see was out, but the replacement insisted that we see a geneticist for his thumbs. This of course made no sense to us, but my husband, John took him. After we got the test results, we cried a lot and freaked out. We researched what we could about RTS, but there wasn't much to find. When we got the actual RTS diagnosis, in a lot of ways it was a total relief. There wasn't a question anymore, there was just the future.

Jaymin is now in 1st grade at an amazing charter school where $\frac{1}{3}$ of the kids have IEPs and the other $\frac{2}{3}$ are "typical." He loves school. He's great at math and loves to count. He also loves to recite his favorite books and adores reading.

He is completely obsessed with all sports, and loves to play them! His other current obsessions include dinosaurs and his Snoopy collection, which he is extremely proud of. He got a new bike for Christmas and is quickly learning to ride. We love to go to museums as a family on the weekends, especially the Museum of Nature and Science, where Jaymin can walk through the Botanic Gardens. He also participates in gymnastics class on Saturdays and looks forward to it all week.

Jaymin is a delightful boy who makes friends easily, smiles virtually all the time, and is extremely caring.

—Samantha and John, Jaymin's mom and dad

During early childhood, parents often notice that their child with RTS is not growing in height as quickly as their typically developing peers and that they may be gaining weight^{5,22,25}. The average height for males is around 5 feet, and about 4 feet 9 inches for females^{5,22,23}. Both males and females with RTS have a tendency to gain excess weight, starting from an early age and into the teenage years. It is important for your child to eat a balanced diet and stay active. Finding enjoyable activities is a helpful way to promote exercise and could include swimming, dancing, walking, or playing a sport. Activities like these can be done in the community with their typically developing peers, as well as supported activities through Special Olympics or your local developmental disabilities agency. If additional attention is needed regarding your child's weight, families might consider working with a nutritionist for guidance.

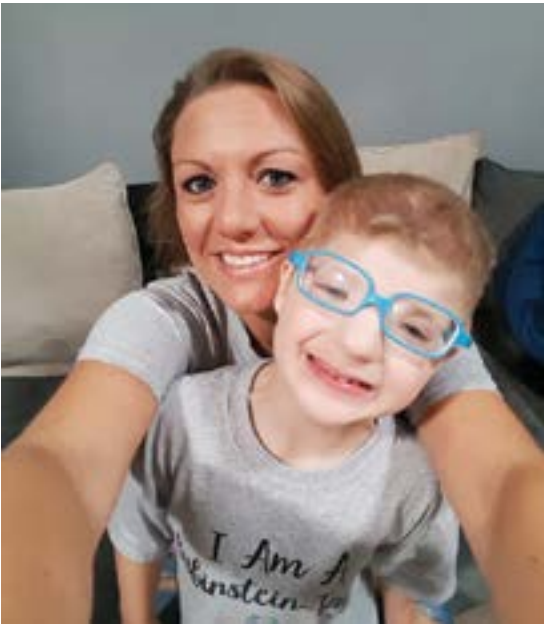
Dental health is important for all children, including those with RTS²⁷. The recommended age at which kids with RTS should see a dentist varies, but a focus on brushing should help with visits and care later on⁶. It is tempting at this age to put children down to bed or a nap with their sippy cup or bottle, but this can lead to cavities and additional problems.



This is what Sawyer's sister Shelby has to say about living with her brother with RTS:

"I like when Sawyer laughs. He has a cute laugh... My favorite time is when we are dancing to music together. He really likes it and I like it too!...This summer, we made a special, secret handshake. He always remembers how to do it and it makes us both happy... When we are older I don't think we will argue as much about silly stuff like the iPad!"

Sawyer and his sister Shelby having fun together



Jett and his mom taking a selfie on World RTS Day

School Age

Seeing your child attending school, working on homework, and engaging with their peers will be rewarding. Most parents with children who have RTS say that their child loves school and looks forward to going every day. Your child's version of school might look different depending on their needs and abilities, but regardless of the classroom setting, parents are usually amazed at what their child can do! As your child becomes involved in school and extracurricular activities, there are some potential health concerns to be aware of, addressed below.

Continuing attention to dental care is important. Because individuals with RTS typically have crowded teeth, a high arched palate, and small mouth opening, brushing is sometimes difficult^{1,2,6,14,15}. It is recommended that you supervise your child to make sure they are thoroughly brushing their teeth. They may also need some encouragement when they do not feel like brushing their teeth. A large percentage of children with RTS have at least one, and often two, talon shaped teeth. Some children have this in their first set of teeth ("baby teeth"), but in most cases, these occur in permanent teeth. These teeth do not typically impact dental care or require additional procedures^{14,15}.

At this age, males, if not closely monitored, may start to gain noticeable weight. Females tend to gain more weight during adolescence^{1,6,22,23}. Again, a well-balanced diet and exercise are important.

There are ways in which you can help support your child to reach his or her full potential in an academic setting. This might include advocating for your child by asking teachers and staff members for school based interventions, therapies, and/or augmented communication devices.

Children with RTS almost always require some level of special education services in school. Assessments and evaluations can be done to identify the individual needs of your child and are then written in an Individual Education Plan (IEP). An IEP typically includes the following: educational goals designed to close the skill gap between

your child and their typically developing peers, supports to access the curriculum, ways that your child can become more self-sufficient in everyday life, and means by which your child can better communicate and advocate for themselves.

This is a good time to be certain you understand the rights afforded to you and your child through the Americans with Disabilities Act (ADA) and the Individuals with Disabilities Educational Improvement Act (IDEIA). This can be done by connecting with organizations that support families and individuals with intellectual and developmental disabilities, like your statewide and local Developmental Disabilities (DD) agencies or the local Arc chapter.



Ryan-Dylan enjoying time at the park

Parent Tips

- Maintain communication with specialists (including geneticists) even when things are going well. This has helped us make more informed decisions during challenging times.
- Using an electric toothbrush might be more helpful than a standard toothbrush for more thorough, easy brushing. Consult with your child's dentist prior to implementing this tip.
- Be an active player in your child's IEP process. Decisions are being made that impact your child's trajectory in life. Learn as much as you can about your child's abilities, their rights, and your school to best help your child.
- Find resources for advocating for your child's needs, like "From Emotions to Advocacy" by Pete Wright.
- Encourage your child to be as much a part of the community where you live as possible. Create opportunities for them outside of the disability system by combining community activities with typical friends and supported activities with peers with special needs. This helps them develop lifelong relationships.
- When your child has difficult behaviors in public, fight the urge to keep them home. Get help from your child's school, therapists, and behavior specialists to work past the difficulties so your child can continue to be a part of things.
- Begin to help your child think about the future. Think about what you want for them and what they need to accomplish this. Share that dream with others. Some of your child's best supports are family and friends who share your vision.

(These tips are shared from other parents of children with RTS. Always consult with your child's pediatrician or specialist prior to implementing these tips.)

Families who have children with significant developmental delays often look to community-based therapies and interventions, in addition to those provided by the school. Your child is growing and learning significantly during this time and these additional supports can maximize progress in areas where they might be experiencing delays. Input from experts, in your child's specific areas of need, is the best way to determine if this approach can assist in your child's progress.

If your child with RTS experiences speech delays, an augmented communication device or assistive technology may be used. These supports can help a child communicate and demonstrate what they know and need in school, and may help to curb behavior challenges. It also allows them to connect with their peers to create friendships.



Inara is Mia's little sister and shared some thoughts about their relationship:

"I love everything about my sister! She makes me laugh-cry by saying silly words. We like to play hide and seek. We like to pretend that I'm a dog and she tells me what to do. This summer we learned how to swim together, and I loved it! I like to just spend time with her... because I love her very much!"



Mia and Inara enjoying a meal together; playing on the playground

Meet Mia

Mia is 10 years old, and lives with her dad, step-mom, and 5 year old sister.



Mia at the playground

We live in a rural Midwestern community where Mia has become quite the celebrity. She wants to be a teacher when she grows up and her favorite pastime is holding class for her baby dolls. As a family, we enjoy going to the park where Mia loves to swing.

Mia is in 4th grade, attends a special needs class at school, and is a social butterfly! Mia loves school and it plays an important role in her personal development. Mia didn't start talking until kindergarten, but now her vocabulary is huge and she is reading up to about 75 sight words! She works on self-care at school, such as learning how to brush her hair and use dry shampoo. Mia is very capable, but sometimes relies on people to do things for her that she is able to do herself. We're continually working on her self-reliance.

Mia teaches our family about patience and unconditional love. She has surpassed any expectations that we had for her when she was younger. One of Mia's greatest attributes is that she loves with all of her heart. She is in a great mood 98% of the time, but does get sad occasionally. We let her cry for 5 minutes or so, then she will say, "Mia happy now!"

If there was a piece of advice we'd give to a newly diagnosed family, it would be to be brave and push yourself to not give up. Always remember that you know your child best. If something doesn't seem right and a test points to nothing, keep pushing and working with your doctors to figure out what's wrong—advocate for his or her needs. One of the hardest things we have had to deal with is having a child that is non-verbal and figuring out what is ailing her. Finally, don't become complacent with your child. They deserve to be the best they can be.

There are times we're so happy with Mia's progress that it feels okay not to push her as much, but that's not fair to Mia. She deserves to reach her full potential just like any other kid.

—Jamie and Aimee, Mia's dad and step-mom

Meet Jason

He is 12 years old and lives with his mom, dad, and older brother.



Jason sharing his contagious smile

We are a typical upper middle class family living in a suburb of Ohio. My husband Rob and I both work and our kids go to public school. We enjoy hiking and being together as a family. Jason has a busy schedule and is involved with Cub Scouts, church, and other school related activities.

Jason rides the bus to and from school. He's in 6th grade and attends class in a separate, modified middle school setting. Jason's with typical kids for some subjects, but has a modified curriculum. He's in a smaller class with an Intervention Specialist for reading and math. His favorite part of the school day is lunch because he gets to sit with his best friend and then go swing outside. Even though his comprehension is a bit delayed he can almost read at grade level, and knows his math facts pretty well! Using an iPad for homework has really been good for Jason.

Jason is a delight. Everyone in our community knows him for his “million dollar smile,” and it's been great to see all of the people that are committed to helping him succeed.

We are proud of Jason for many reasons. He does fairly well academically and has received awards for compassion and perseverance, as well as a student nominated award for “friendliness and helping others.” He recently learned how to ride his bike and loves it. Jason participated in the summer Special Olympic games and stayed in the dorm with his team. We were so thrilled with his independence!

Our advice would be to enjoy each day, and milestone, whenever it comes. You can't know the future. Focus on what you and others can do to help your child reach his or her full potential. Let your child determine that potential, and try not to measure against other kids, even those with RTS. Your child is unique, and wonderful, and will show you things about life you never imagined.

We have been fortunate that Jason has had few medical and behavior issues. He is just a really sweet boy who loves Pokémon.

—Karen and Rob, Jason's mom and dad

Adolescence

This stage can be a fun, yet trying time for any parent regardless of the child's ability level. You will begin to see physical changes in your child and may even notice a variability in emotions as hormones change. As your child with RTS hits puberty, there are a few things (besides patience) to keep in mind.

Puberty for individuals with RTS starts around the same age as their typically developing peers, between 9 and 12 years of age^{6,14,15,23}. You are encouraged to begin conversations with your son or daughter before the signs and changes of puberty begin. These can include learning the appropriate formal words for body parts and processes, and the importance of good and effective hygiene routines. Help your child develop easy to follow routines for the morning like showering, dental care, and shaving. Explain menstruation, nocturnal emissions, erections, and extreme mood changes, where applicable. Do this at an appointed time, in a relaxed setting. Take into account your child's learning style using supports that work best for them like repetition, pictures, simplified facts, and stories about social expectations and experiences.

In most girls, periods start about 1–2 years after breast development begins¹⁴. Having a conversation with your daughter before her period begins is important. Remind her that this is normal, and that though she is bleeding, she is not hurt. Answer all her questions to avoid fear or embarrassment. In case your daughter starts her period when you are not around, help her practice the conversation she might have with appropriate adults. You can also teach her how to use a pad and pack supplies, so she feels confident and ready. It might also be helpful to talk to your child's pediatrician, physician, or a gynecologist about how to best address menstrual cycles, especially if it seems to cause pain and/or irritation.

You may also want to prepare for other conversations related to body hair, private parts, erections and emissions. These conversations should be fact-based, but will also reflect your family's value system. Explain to your child

that these topics are private, but it is good to identify who they can talk to about these things—parents, doctors, or the school nurse. It is important to distinguish what parts are private, and what is appropriate and inappropriate in public. However, the touching of private parts could indicate irritation or infection. If your child is insistent or the behavior gets in the way of other activities, you should consult your physician.

There is no research indicating that males or females with RTS have challenges with fertility, so your family may think about how to address the topics of pregnancy prevention and pregnancy^{6,7}.

Adolescents with RTS have a typically developing sexual drive and all associated feelings^{1,6,7}. Teens with RTS are just as interested in developing intimate relationships as anyone else. It is important to teach your child about appropriate and safe relationships with others. If your child is interested in a relationship with a peer, support them with clear guidance.

Parents will probably see an increased need for sleep in their teen with RTS¹⁰. Sleep cycles in typically developing teens tend to shift to going to bed later and sleeping in later, due to a difference in melatonin cycles²⁸. There is not much data on sleep related to RTS, but it seems to be consistent with typically developing teens.

This is the age when you might start to notice your child with RTS gaining weight, specifically females^{1,6,7,22,23}. Males typically start gaining weight earlier (around 6–12 years, see School Age section). A well-balanced diet and exercise are recommended.

Puberty for individuals with RTS starts around the same age as for their typically developing peers, between 9 and 12 years of age^{6,14,15,23}.

In typical adolescent development, new behaviors may arise. Your child with RTS is no different. These behaviors (see Social and Emotional Development section) are varied, might catch you off guard, but often times aren't harmful to anyone. It is a good idea to talk about what to expect when going into a new or unfamiliar place ahead of time. Especially, if you know that it will be different from normal routines. Things like how noisy the place will be,

how many people will be there, and what they might see and experience, are important to discuss. Creating visual schedules and having a conversation about any expectations in behavior, might be helpful. It is recommended to consult a behavioral specialist or psychologist on positive coping strategies during stressful times or when behavior begins to negatively impact your child's participation in daily activities. Individuals with RTS at this age can be friendly and outgoing. However, at times they may be overly social. Working with your child on how to be socially appropriate in all situations is important (e.g. not hugging strangers or understanding personal boundaries).

Identifying your teen's likes and dislikes, interests, preferences, and/or skills will help frame what they might like to do in the future as far as employment and/or volunteer work. These topics can be discussed with your child's school team and written into their IEP (until age 21). All of this will help your teen develop a vision on how to live a healthy, happy, safe, self-determined, and productive life in the community.



Ethan is Emma's older brother. Ethan and Emma are very close. Ethan shared what he likes about his sister and how he plans for her future:

"I love Emma's outgoingness! She is very affectionate and likes to hug on me. It means a lot that she cares so much and that she shows it...Even when I do not live at home I am still going to come see her. I know that I am going to help to take care of her in the future. I am fine with that. By being a part of her life as an adult, I will know that she is safe and she will always have people around her that love her."

Ethan and Emma attending a performance

Meet Cooper

Cooper is 15 years old and lives with his mom, dad, and dog, Nosey.



Cooper loves getting out and about in his hometown

Our family of four lives on a quiet street in an urban community. We are blessed to be surrounded by a large support system and spend a lot of time with our loved ones. During the work week, Cooper spends a lot of time with his community living support staff, at the park, playing basketball or Wii, at the library, and listening to music. On the weekends, we like to cook, watch movies and Cooper's favorite, watch YouTube videos!

Cooper is a sophomore at a public high school. He's in a self-contained classroom with peer mentor support and opportunities for mainstreaming with typical peers in elective classes.

Cooper and his Content-Based-Instruction (CBI) instructor go to a local grocery store once a week for career exploration, where he is learning to stock shelves. Cooper also goes with a group to retail stores and restaurants where they apply practical skills like self-care, safety, organization, and practical math. He takes the bus to and from school, and has many friends with and without disabilities.

Cooper encountered many physical challenges early in his life, including multiple surgeries. He also had many infections and severe asthma. Now, Cooper is playing basketball, walking two or more miles, and taking stairs independently! He is nearly asthma free, and very healthy! Cooper has also had to learn how to communicate nonverbally. He learned sign language, then progressed to the picture-exchange communication system (PECS), and, now uses augmentative communication devices, including ProLoQuo on an iPad. Now Cooper is communicating unprompted, in sentences, typing his own thoughts, and having conversations with everyone he meets!

One piece of advice I'd give is to breathe. This will take time to understand and accept. Be patient with yourself and your child. Re-frame your perceptions and expectations. Reading "Welcome to Holland" helped us. Reach out to other RTS parents and support groups. You are not alone!

—Joe and Jennifer, Cooper's mom and dad

Transitioning to Adulthood

The transition to adulthood can be exciting and overwhelming. There are lots of choices for youth with disabilities, so it is important to consider your teen's strengths and challenges, their likes and dislikes, their interests, dreams, and desires. Getting in touch with local, state, and regional resources can help determine steps in the transition process⁶. It is important to work with your teen, their school team, and the state disability resource systems to determine the skills your child needs to take their next step, whether they plan to participate in independent daytime activities or supported programs.

The transition process happens across several years and includes numerous decisions. Typically, in the United States, it begins at age 14 with the Transition IEP (Individualized Education Plan), which is designed to formulate a timeline for a child to transition from school-based to adult services. It is helpful to have a conversation with the school team prior to the first transition IEP meeting. During this conversation, you can discuss your vision for your son or daughter's life and what skills they have so that you can create a plan on how you will work together to get there. Topics of conversation could include: school based services after the age of 18, post-secondary programs, a job in the community, volunteering, internships, college-supported program, etc. The plans you make and the skills your teen needs to be successful are then built into the IEP as transition goals.

There are also financial and legal decisions that you will need to learn about as your teenager approaches the age of 18. These include decisions about eligibility-based Supplemental Security Income (SSI) and Social Security Disability Insurance (SSDI). This could also be a good time to discuss an ABLÉ account if you don't already have one. It is also important to talk about where they might need support in making life decisions after turning 18, by learning and discussing the options of Conservatorship, Supported Decision-Making, Powers of Attorney, and Guardianship.



Xen (13) from the UK is focusing while completing his artwork



Another drawing by Xen reminds us that every child has talents and abilities, no matter their challenges.



*Devin taking his work seriously
and giving it his all*

It is crucial to talk about how, when, and where your young adult might transition from a pediatric to an adult healthcare provider, both in general medicine and specialty areas of care. You can check with your pediatrician and other specialists as to how long they will follow your child past the age of 18. You might also consider an earlier transition to a family practitioner, as they will follow them across the life span. These decisions can be daunting, but if conversations about transition occur early and often, the transfer to adult care may be smoother. Given the medical complexity many individuals with RTS experience, it is important for you to track your child's medical history. This is sometimes done through a care notebook (see Care Coordination section) and can be used to inform new adult providers. Their pediatrician, physician or specialists should be able to provide you with healthcare summaries and transition checklists to prepare for the transition to adult healthcare.

Every person has the right to live in their community, doing what they love with the people they care about. During this transition period, you, your young adult with RTS, and your family may discuss and make decisions about what this means for your family. Your son or daughter might participate in life much like your other family members, supported by family and friends in the community where you live, or they may choose to be part of a community-based supported day or employment program, or maybe something in-between. You will want to consider whether your family member will live at home, in a home or apartment in your community, or in a supported environment. Each community is different, and has different options and resources. It is important to assess and reassess your options to determine what is best for you and your transitioning adult. Getting in touch with local, state, and regional resources can be a huge help in determining next steps. Talking with other families who have made decisions for housing, transportation, and community living may also help you decide what is best for your family.

By law, at 18, your child is considered an adult and is legally responsible for making decisions for him/herself. Protecting your young adult's autonomy and personal independence, while allowing them to make safe and/or supported decisions is crucial for them to live a good life²⁹. In most states, there are various options for supporting your son/daughter to make decisions for themselves. These may include, but are not limited to, General Power of Attorney or Durable Power of Attorney in a specific area (financial, medical, educational, etc.), Representative Payee for Social Security benefits, Supported Decision Making, Conservatorship or Full Guardianship.

Often, conversations about these topics begin at the IEP table as part of your child's transition planning. There are no hard-and-fast rules to determine whether or not your child needs you to take guardianship at 18 years of age, or whether allowing some decision making in all or specific areas with supports is more appropriate. You may be told that guardianship is necessary, but there may be other options that would promote the health, safety, and independence of your child. Simply stated, it is important to know that guardianship, though at times an appropriate choice, it is not the only choice. It is recommended that you do thorough research about the options in your state. Ask questions of your state's Developmental Disability agency, your University Center for Excellence in Developmental Disabilities (UCEDD), Developmental Disabilities Council, Protection and Advocacy agency, or an attorney who understands all the guardianship options. Take time to become informed on each of these choices.

Every person has the right to live in their community, doing what they love with the people they care about. During this transition period, you, your young adult with RTS and your family may discuss and make decisions about what this means for your family.

Meet Sophia

Sophia is 21 years old and lives with her mom and dad.



Sophia taking a brisk walk with her parents and dog, Gabriel

We live in the suburbs with Sophia's lovable but unruly golden retriever, Gabriel, and three cats that she thinks are "so cute." Our town is small, and everyone knows and loves Sophia. Much of what we do, we do together. Sophia is an adult now, and we are in a phase of trying to figure out how to give her independence while still supporting her to make good decisions. She keeps active by swimming at the YMCA. She also loves thrift stores, Chipotle, and Starbucks.

Each day, Sophia rides the bus to an adult workshop environment, which she loves. Sophia likes structure in her routine, from breakfast to how she gets ready. She also participates in Stepping Stones' Young Adult Club.

Our family has highs and lows just like anyone else. However, having a person in our family with RTS has allowed us to meet many people we likely wouldn't have met otherwise. And because Sophia is so outgoing, we get to know people on a different level!

Sophia is funny, dramatic, and loves people. We sometimes struggle with her behavior, but if she can get beyond her anxiety, she is deeply empathetic. She is a tremendous soul and we are constantly amazed at her resilience, joy, and excitement by life and her surroundings.

Dr. Rubinstein said it to us best when he told us to "go live your life". While there is strength in connecting with others in the RTS community, the real beauty happens when you just start to live and not let RTS define your every moment. Don't be afraid to reach out to your family and friends early on to make them a part of your extended support network—it's so important to your family's ongoing success.

—Bill and Melissa, Sophia's mom and dad

Adulthood and Aging

Information in the medical literature on adults with RTS is limited, which can make care challenging for medical professionals and caregivers. Typically, issues that individuals with RTS face during childhood can continue to impact them as they age (weight gain, spine curvatures, vision issues, keloids and tumors, eating challenges, etc.)^{6,13}.

Most adult providers are unfamiliar with RTS and the issues associated with the syndrome. It is recommended that you continue check-ins with the specialists your loved one sees now (this may include orthopedics, gastroenterology, cardiology, endocrinology and neurology), while also scheduling time with your geneticist. These regular check-ins will allow you to share important information and research about RTS and how it may pertain to your adult child, ultimately forming a team to address any challenges in aging^{6,13}.

Behavior issues may worsen with age^{13,18}. These behaviors may include getting and maintaining attention, impulsive or disruptive behaviors, and repetitive, self-stimulating behaviors, which are common in other developmental disabilities and mental health diagnoses like ADHD, ASD, anxiety or OCD^{1,6,13,18}. If your loved one shows behaviors like these, work with his/her physician to identify whether they may benefit from educational, behavioral, and/or pharmacological therapies. These behaviors may also indicate a new physical or medical condition and may warrant further investigation or a medical assessment^{2,6,18}.

Some families of adults with RTS have reported a decline in cognitive ability, stamina, mobility, vision, and hearing¹³. To possibly lessen this decline, it is important to stay integrated and active in the community. One suggestion may include connecting with both typically developing peers and peers with disabilities, with similar interests. Social interaction, emotional support, and physical/mental wellness help people avoid boredom, depression, and negative behaviors.

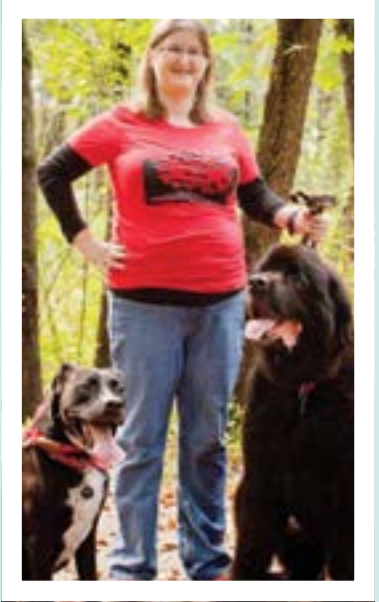
We all make decisions about how to age, including where to live, how to spend time, and with whom to spend time. These decisions need to be discussed and adjusted periodically. Not only are you thinking about these things for your adult child, but you are also making decisions about your own life as the parent or caregiver of an adult with RTS. Where and when will you retire? Will you sell the family home for something smaller? Will you move close to grandchildren? It is also important to think about what will happen when you are no longer able to care for your adult child and to have a plan in place. Is there a sibling who is interested and willing to take care of your son or daughter with RTS, or other relatives? Have you set aside funds to support your adult child? A plan for aging well (for both you and your child with RTS) allows you to anticipate change by initiating conversations for future planning, helping to avoid crises created by illness or death.

If you have not already, you will need to start thinking about supported decision-making and guardianship beyond yourself. Who will help your adult child with RTS continue making decisions when you are no longer able to? Have you discussed and formalized end of life decisions (for you and your adult child who has RTS), including advanced directives? Have you identified times to update plans with the appropriate family members? Have you considered the financial needs of your adult child with RTS? Have you finalized your will and discussed a successor guardian, if applicable? Have you explored finances, special needs trusts, assets, ABLE accounts and other ways to support your adult child with RTS? All these conversations are important and if you have them early, it will reduce the stress of tackling them during moments of urgency and crisis.

A plan for aging well (for both you and your child with RTS) allows you to anticipate change by initiating conversations for future planning, helping to avoid crises created by illness or death.

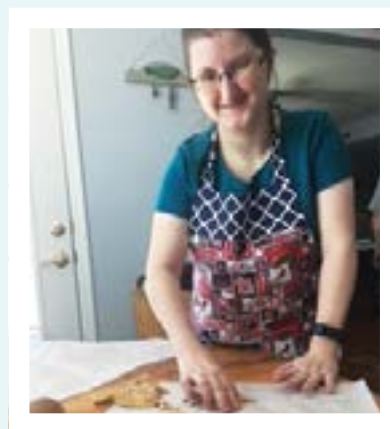
Meet Jessica

*Jessica and
her dogs Annie
and Yadi*



Jessica is from St. Louis and started her own company, “Yadi’s Yummies”. She began this in her home and sold her treats at craft fairs, special events, expos, and online. Thanks to a lot of hard work and support from her customers, community, and family, she recently opened up her own shop in the heart of St. Peters. Jessica has proven that with hard work and determination, no task is too big.

“Hi, my name is Jessica and I have two dogs Annie & Yadi and two cats Joey & Mickey. I am a huge sports fan and especially LOVE cheering for the St. Louis Cardinals and the St. Louis Blues. I enjoy going to sporting events, horseback riding, and spending time with my family and friends. I started Yadi’s Yummies for two reasons. First, I love dogs and enjoy making dog treats. Second, I wanted to find a way that I could help some of the abandoned and abused dogs I see in the shelters that don’t have families to love and care for them. Through your support I can donate to the medical care of these dogs so together we can make a difference.”



Above: Jessica at her new store in St. Peters; Jessica baking her homemade dog treats; Jessica takes Yadi’s Yummies on the road, selling her tasty treats at an event in her community.

Meet Devin

Devin is 25 years old and lives with his mom, dad, younger brother, and lab, Bella.

Devin loves biking for fun



We live in a fairly small town near Indianapolis, surrounded by family. We enjoy going out to eat or just spending quality time together at home. Devin doesn't like busy, noisy places, so crowds aren't ideal, but he's very social and knows no stranger.

After receiving the diagnosis of RTS, I asked myself 'what did I do wrong?' and I grieved for the healthy baby we had planned for. It wasn't until we attended our first RTS conference, that we did not feel alone. We had found our new family to support and encourage us.

Devin graduated with his Certificate of Completion in 2012. That year, he was crowned Homecoming King and was also on the Prom Court, which was voted on by his peers.

Currently, Devin attends a day program, where he puts boxes together and packs care kits. A couple times a month, he goes on one-on-one outings, like eating out and shopping. Devin also participates in a Life Skills Training Program, where he works on day-to-day self-care and pre-vocational skills. The rest of the time Devin is at home with me and enjoys a routine.

My advice is, celebrate the little things and brag about the accomplishments, no matter how small. You are the expert on your child, so trust your instincts. My husband and I prioritize making time for each other away from our children, like going on cruises. It's so important to our marriage.

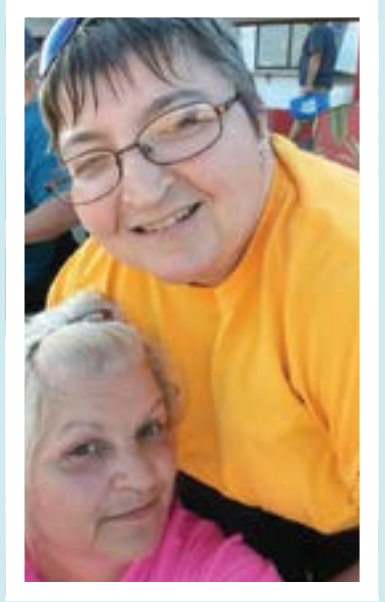
Take it easy, take a breath, and be gentle on yourself; no one is perfect. Parenting is difficult no matter how typical your child is (or isn't). It's okay to have a good cry every now and then. Most importantly, you are not alone. Over the 25 years that we have been blessed to have Devin in our life, he has taught me what it means to love unconditionally, to love someone so much it hurts, and what it feels like when your heart is so full it could actually burst.

—Corina, Devin's mom

Meet Melissa

Melissa is 46 years old and lives in an assisted living facility.

Melissa and her sister enjoying a day together



I live in an Assisted Living Facility with both young and old people. My parents are still alive, but my sisters thought it was time that I live a little more independently. Not everyone here knows that I have RTS.

A typical day for me includes making bracelets and necklaces and talking to other residents! I've been on a huge weight loss journey recently and have lost 19 pounds so far. I take a lot of walks during the day for exercise and watch what I eat! My goal is to live life to the fullest and be an inspiration to those around me.

I have an amazing boyfriend who is totally deaf and has Asperger's Syndrome. He lives in a different state, so we video chat in American Sign Language (ASL). My boyfriend and I love each other very much. My boyfriend doesn't know about me having RTS though because he wouldn't understand and it's complicated to explain.

Having RTS has meant a lot to me and I wouldn't trade it for anything in the world. It has taught me how to love and be loved. I count RTS not as a curse or disadvantage, but a blessing. I've had to face obstacles in my life, like learning difficulties in school. I've had anger issues sometimes, getting in spats with my mom or having an attitude. I usually talked out any issues with my mom, sisters, and daddy. I've been able to overcome anything with God and the close bond my family and I share. My two brothers were always there for me, too. I'm very grateful that my mom and daddy raised me in the church.

My advice is to not think of RTS as a bad thing, but a good thing, a blessing in disguise. Think of it as God's way of saying he has a special plan for you. RTS is a gift. One of my favorite things about having RTS is sometimes I get in a mood where I want to go hug somebody and tell them I love them. I have my flaws just like anyone else, but I am what I am. This is how God made me and wanted me.

I think that's pretty special.

—Melissa



Dr. Manning and Griffin at his appointment

What to Expect Medically

Individuals with RTS may have a variety of medical conditions, but most will have a good quality of life. This is especially true when medical concerns are addressed early and a follow-up regimen is in place.

Anesthesiology: If anesthesia is needed, the medical professional should understand the potential differences in the airways of individuals with RTS, as well as what types of medications have been safely used with your child. Without these considerations, your loved one may be at higher risk for cardiac arrest^{2,5}. Intubation may be necessary if using anesthesia, due to high risk of aspiration during surgery, but could be challenging because of easily collapsible laryngeal walls⁶. You can ensure that your physician and anesthesiologist are well informed by requesting a pre-op anesthesia consult. This strategy is often used, even in the most minor of surgeries, when people have a higher risk of issues with anesthesia.

Cardiology: Children with RTS may be born with congenital heart problems. Congenital heart disease (CHD) occurs in about 30% of individuals with RTS^{1,6,15}. In the United States, hospitals are now screening *all* infants for the possibility of a congenital heart disease. If there are concerns in the newborn screening process, more testing, like an echocardiogram (an ultrasound of the heart to look at the structure and functioning of the heart) is often done.

If CHD screening was not done with your child, you can ask for it. Children with RTS should also have regular check-ups that follow usual healthcare guidelines (checking blood pressure at well child visits starting at the age of 3 years). If heart problems are identified, it may be best if a pediatric cardiologist guides next steps and appropriate intervals of follow-up⁶.

Note: the medical items in this section may also be addressed in the life stage sections earlier in the booklet. It is important to seek professional medical advice for any of the issues or concerns addressed in this section, as well as any concerns not listed.



Example of what keloids look like

Dermatology: Individuals with RTS are susceptible to skin growths called keloids. These are raised, often puffy, excess scar tissue that form on the skin, often because of skin irritation. While keloids most often form on the chest and/or back, they can form on any part of the skin, most often when it is healing from something like surgery, or even as minor as an insect bite, piercing, or vaccination²⁴.

Gastroenterology: Issues with reflux, gagging, and spitting up are common in babies with RTS. Some infants might require a feeding tube if they are not able to chew or swallow properly and are losing weight⁶ (see Infancy section). Most infants grow out of these issues and develop more typical appetites.

Constipation can occur in individuals with RTS of all ages, but especially in childhood and adolescence^{1,2,6}. It is helpful to talk to your child's pediatrician about the options to aid in this issue. He/she may recommend a special diet, such as increasing fiber or fluids, or specific medications like laxatives to help avoid or relieve constipation. It is important, especially after your child is toilet trained, to develop discreet and respectful ways to monitor and track toileting and soiling. Tracking can help identify what might be wrong if you see a change in their behavior or activity.

Individuals with congenital anomaly syndromes such as RTS are also at a higher risk of having malrotated intestines³⁰. This occurs when the intestines (or bowels) do not coil like they should, or become twisted, leading to discomfort and blockage. This typically occurs in early childhood, if at all. If vomiting, extreme stomach pain, or constipation occur as a result of the intestines being malrotated³⁰, surgery will need to take place. If you are concerned that your child could have this problem, contact his or her pediatrician and/or gastroenterologist to request further evaluations.

Genitourinary: Current research shows that more than half of individuals with RTS have kidney problems, which can lead to frequent urinary tract infections. In addition, many males diagnosed with RTS experience incomplete or delayed descent of their testicles, and if they have not dropped by the age of 6 to 12 months, a pediatric urologist should address it⁶.

Neurology: Many individuals with RTS have some neurodevelopmental differences, resulting in developmental delays and cognitive disabilities. Families should reach out to their Early Intervention and developmental disability systems, schools, and pediatric providers to learn more about services and supports, early intervention, school based, and private pay therapies to achieve the best outcomes possible⁶.

As addressed earlier in this booklet, children with RTS may have a condition referred to as a tethered cord^{25,26}. A tethered cord is the result of stretching and tension of the spinal cord. Some individuals are born with a tethered cord, but it most commonly occurs in times of rapid growth in younger children. Signs of a potentially tethered cord include: severe or worse than normal constipation; toileting issues after a child has been potty-trained; difficulty sitting upright; changes in how your child walks (crouched gait, tip toes, trouble with balance), and lower back and leg pain^{25,26}. It is important to seek advice from a healthcare professional if you recognize changes in your child's mobility, behavior, or toileting. Trust your instincts and be sure your provider hears your concerns. A tethered cord can be corrected with surgery.

Oncology and Hematology: There have been several reports of benign (noncancerous) and malignant tumors in individuals with RTS^{31,32}. The most common reports involve meningiomas and pilomatricomas, which are benign. It has not yet been determined through research that malignant tumors occur more commonly in persons with RTS than in the general population. Keep in touch with your team of doctors, specifically a geneticist, to ensure that everyone is keeping a close eye on anything that might seem out of the ordinary³³.



*Hamza playing at an RTS
OKI event*



Example of what talon teeth look like

Ophthalmology: Individuals with RTS can have glaucoma or experience strabismus (crossed eyes) as an infant. Catching these things early is important. It is also common for those with RTS to experience watery eyes or discharge from the eye. If any of these issues persist, treatment might be necessary. Children with RTS are often referred to an eye doctor (ophthalmologist) as early as 6 months old, or at the age they are diagnosed with RTS^{6,13,14}.

Orthopedics: In addition to intellectual delays and intellectual disability, children with RTS may experience problems with the way that bones are formed and how flexible the tendons can be. This can cause a kneecap to slip, hip problems, back pain, and leg pain^{13,14}. It is important to look for signs and monitor your child closely from an early age, to prevent long-term problems^{6,13,14}.

Respiratory/Ear, Nose, and Throat: Some research indicates that individuals with RTS are at risk for mild hearing loss, and frequent ear infections are common¹³. Because individuals with RTS often have small nasal passages and airways, respiratory infections are common and can sometimes turn into something more serious, like pneumonia^{1,6,13}. It is also common for children with RTS to undergo surgery to remove enlarged tonsils and adenoids, especially if obstructive sleep apnea is a concern⁶.

Signs of sleep apnea can include gasping, night sweats, loud snoring, and strange sleep positions. If sleep apnea is not identified and treated over a long period of time, it can cause a strain on the heart and lungs, leading to pulmonary hypertension.

Concerns about any of these issues should be addressed with your medical provider and/or specialist as early as possible. Your doctor may recommend surgery to remove tonsils and adenoids. Discussions about anesthesia should happen prior to these or any other surgeries, due to the risk factors mentioned in the Anesthesiology section. They may also provide a referral for a sleep study in the case of sleep apnea. Untreated sleep apnea can cause a strain on the heart and lungs, which may lead to pulmonary hypertension⁶.



Wally in a field of sunflowers

Suggested Medical Guidelines across the Life Span for Individuals with Rubinstein-Taybi syndrome***6

	Birth–6 M*	6 M	12 M	18 M
Cardiology	All newborns are screened: pulse ox, cardiology, echo and EKG	Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.		
Dental		Begin conversation with pediatrician about regular dental exams.	Crucial to continue 6M check-ups and monitoring for oral health	
Dermatology				
Developmental and Educational Services	Developmental assessments conducted. Referral for Early Intervention (EI) services if concerned.	Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.		
ENT and Audiology	Newborn screening at hospital	Monitor previous areas of concern closely, notify pediatrician of changes.	Begin annual hearing check-ups.	
Gastroenterology	Monitor feeding, constipation and reflux. Share concerns with your pediatrician.		Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.	
Genitourinary	Testicle position of all newborn boys evaluated. Baseline renal ultrasound****.	Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.		
Growth and Nutrition	Use growth charts referenced in this booklet.	Work with pediatrician to evaluate and monitor nutritional status when decrease or increase in weight is of concern.		
Ophthalmology	Assessment for ocular abnormalities conducted	Assessment for ocular abnormalities with visual function tests	Crucial to continue 6M check-ups and monitoring for oral health	
Orthopedics	Spinal ultrasound for tethered cord conducted and assessed****.	Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.		
Respiratory		Assess symptoms of sleep apnea with pediatrician.		

* Abbreviations: M, months

** It is implied that this table continues into teens and adulthood. Continue with these appointments every 6 or 12 months as indicated on the table, or as needed.

24 M		30 M	>3YRs (Annual Exams)	Adolescence**
			Blood pressure tested at check-ups. Share any concerns.	Monitor and share concerns. Primary care provider may make recommendations.
			Regular exams for talon teeth as part of check-ups every 6 months	Monitor and share concerns. Primary care physician may make recommendations.
				Adolescents may develop keloids. Monitor and share concerns with your primary care physician.
			Pediatrician includes systematic developmental assessment tool every 2–3 years****.	
	Annual hearing check-up		Hearing assessment no less than every 3 years.	Monitor and share concerns. Primary care physician may make recommendations.
				Throughout lifespan, continue to monitor weight and height ratio to avoid obesity.
			Assessment conducted no less than every 3 years.	Assessment conducted no less than every 3 years.
Monitor previous diagnosis, assess new risks, and notify pediatrician with concerns.				

*** While these recommendations are made based on supporting literature and research, the medical conditions associated with this table vary from person to person. These recommendations have been made based on generalizations of the complications that most individuals with RTS face at certain times, but is not meant to imply that your child is certain to have these issues. Please use this table as a basic informational guideline, but speak to a physician if problems arise more frequently than this table indicates.

**** It is recommended to monitor with targeted attention due to known issues in this area for many individuals with RTS



Emma and her mom ice skating

Caregiving and Respite

Being the primary caregiver for a family member with a disability, though rewarding, can be challenging and stressful. Research shows that the financial, emotional, and social isolation of parents caring for children with complex needs impacts a parent's emotional and overall health³⁴. We also know that the stress experienced by caregivers of older adults has a negative effect on overall health³⁴. As a parent or primary caregiver of someone with RTS, it is important to allow yourself and your family support system temporary relief or short breaks to step away from this role³⁵. Whether it is grocery shopping alone, going to the gym, or leisure activities with friends, time taken for self-care will benefit all members of your family³⁴.

Because opportunities for formal paid supports are often limited and vary from community to community, families of individuals with developmental disabilities are encouraged to look beyond traditional paid options and to consider alternative supports³⁶. Finding and building trusted supports and caregivers who can establish a close relationship with your child and provide meaningful interactions may contribute to an overall better respite experience. When this time is “personalized and purposely planned, it benefits not only caregivers, but all family members, including siblings or a spouse or partner³⁷.”

When creating opportunities of momentary relief from caregiving, it is important to consider all the possibilities and how you might adjust or adapt these to meet your child's unique situation. Are there places in your community where your child can participate in a planned activity with typical peers? If necessary, could you add a support person through your formal service system or a family member or friend who is willing to share time with your son or daughter? What skills can you help your child build over time to gain independence in these types of activities as he/she grows? Could you ease your worries by checking in with the respite caregiver or friend through a video chat or phone call?

Finding and building trusted supports and caregivers who can establish a close relationship with your child and provide meaningful interactions may contribute to an overall better respite experience.

Identifying the skills that your child with RTS has or needs to spend time in the community without you is one of the first steps to ensure that you are able to take time for yourself.

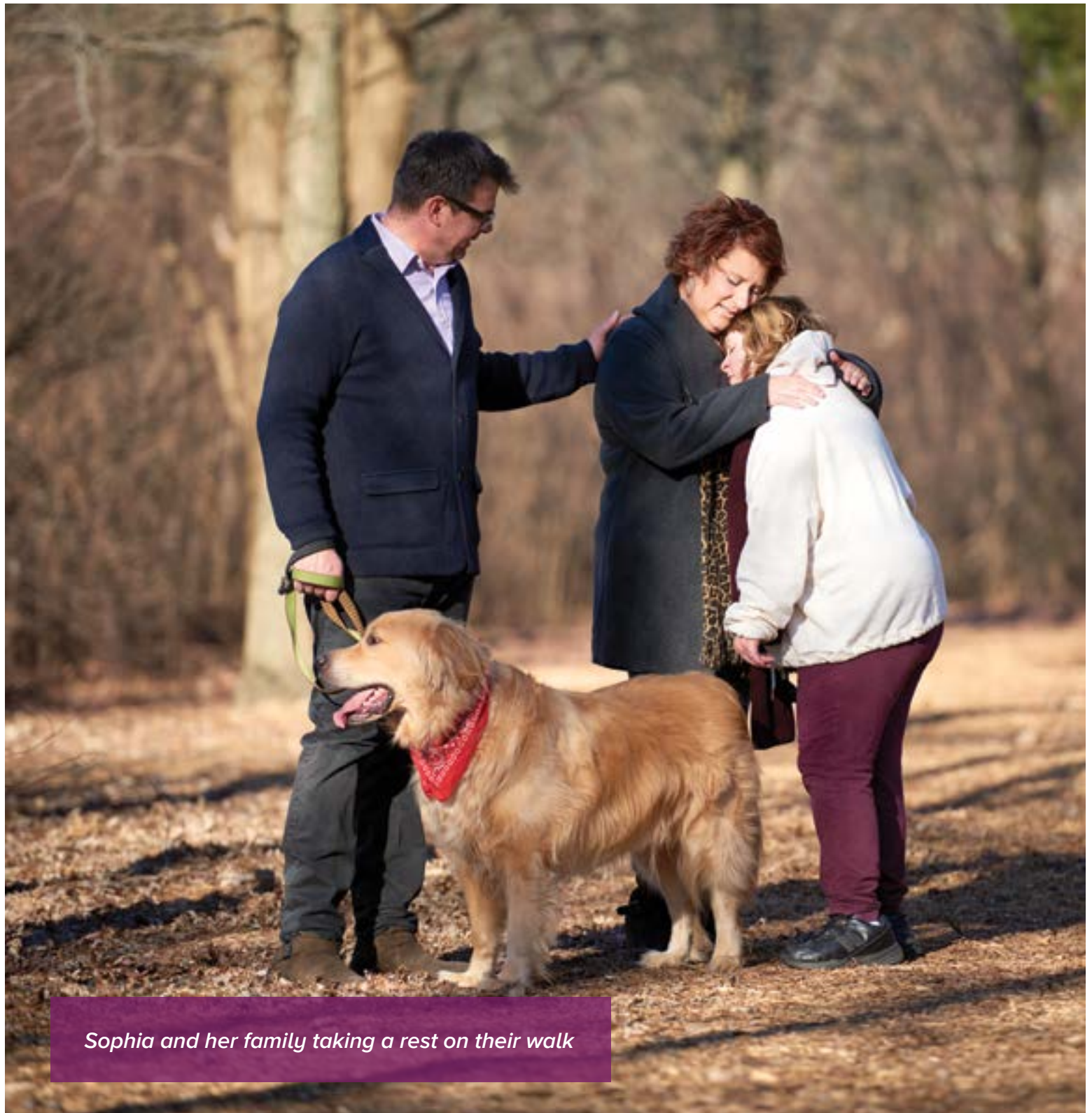
Identifying the skills that your child with RTS has or needs to spend time in the community without you is one of the first steps to ensure that you are able to take time for yourself. Consider your child's strengths, how your child best communicates, and how he/she lets people know what he/she needs. Make sure the caregiver knows what to expect from your child and that he/she is prepared for the activities planned. Understand when your child is most vulnerable and most likely to experience anxiety or express frustration. Identify a space ahead of time where your child can take a break if needed. Thinking through and planning for all of these factors will enhance your child's experience and make these opportunities more successful, creating greater possibilities of respite for you and other family members³⁷.

It might also be helpful to identify people in your life who are already connected or willing to be involved with your child. This is encouraged because building this informal circle of community support will allow your child to be happier, more active, and more independent and self-determined³⁶. As these people become more familiar with your child's needs, wants, and abilities over time, you will be more confident in creating informal community respite opportunities. Each time you step away, you may be able to reduce some of your worry or stress, re-energize, and be better equipped to face the challenges of the day³⁷.

Knowing what's available to ALL in your community and seeing what might be of interest to your child is another way to start creating opportunities for informal respite. For example, community recreation centers or organizations like the YMCA often have fitness or sports activities, art or music-based events for both children and adults. Religious institutions may provide a place to share and support one another, along with opportunities to volunteer. Museums, theaters, and civic organizations may offer community outreach events that are a perfect opportunity to take a break. These are also often offered at little or no cost. Exploring these possibilities in your community, and reaching out to your local disability systems and agencies, will allow you to grow new opportunities outside of the

limits of paid services³⁶. These are just some examples of how to grow opportunities for respite for you and your family, while increasing the quality of life for your child with RTS.

For more information on how to create or access respite, visit the ARCH National Respite Network at www.archrespite.org and www.archrespite.org/consumer-information/lifecourse-tools-for-respite³⁸ or LifeCourse tools for respite at www.lifecoursetools.com/respite/³⁷



Sophia and her family taking a rest on their walk

Conclusion

So much has changed and much has been learned since Dr. Rubinstein and Dr. Taybi met each other and shared their knowledge of RTS. Families now have a body of research, though small, to refer to and to share with their loved one's medical providers for optimal care. There are doctors and geneticists in the United States, Europe, and a handful of other countries, continuing to try to learn more about RTS and interventions that can support individuals with RTS to reach their full potential.

In addition, the advances in technology have improved families' ability to access the most up to date information and to connect with other families who have children or adults with RTS via Facebook, Twitter, the internet, and other social media outlets. Given the rarity of RTS, the ability to reach beyond geographical areas and connect with each other is something that offers families new opportunities to avoid isolation. These connections offer support, information, and informal mentoring.

Families of, and individuals with RTS, are living full lives in their communities. Individuals with RTS contribute to their communities by working, volunteering and spending time enhancing the lives of others who are part of their schools, religious institutions and neighborhoods. With the appropriate supports from their families, community, and experts in the area of disability, the RTS community is thriving!

As family members and parents/guardians, you are encouraged to help your loved one with RTS create and attain their desired best life in their community. It is important to share your experience and knowledge about RTS with medical providers, service providers, other family members, and the general public. Through these efforts you will contribute to a more knowledgeable, accepting, and inclusive society for individuals who have RTS.

Families of, and individuals with RTS, are living full lives in their communities. Individuals with RTS contribute to their communities by working, volunteering, and spending time enhancing the lives of others who are part of their schools, religious institutions, and neighborhoods.

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Contact

For RTS support, contact our Family Support Coordinator at rts@cchmc.org or call 513-636-4723.

For appointments within the Division of Developmental and Behavioral Pediatrics at Cincinnati Children's, call the appointment line, 513-636-4611.



*From left to right:
Lexi, Kiara,
Wally, Lexi,
Rhett, Cristofer
Adrian and
Arleigh*



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