Rubinstein-Taybi Syndrome (RTS) is a rare genetic condition, affecting about 100,000 to 125,000 newborns each year worldwide\(^1,2,3,4\). In 50–60% of cases, RTS is the result of CREBBP gene mutations on chromosome 16p13\(^1,2,3,4\), also sometimes referred to as RTS Type 1. Individuals with this form of RTS have many of the following 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\(^1,2,3,4\). A rarer genetic variation that affects 3–8% of diagnosed individuals is the result of EP300 gene mutations on chromosome 22\(^4,5\), which is sometimes 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 and thumbs are less likely to be angled outward\(^2\).

Genetic testing is done to confirm a “medical diagnosis” of RTS most of the time\(^6,7\). Testing can be done at any age, but the average age of diagnosis is 15 months\(^15\). However, in about 30% of cases, individuals with typical features of RTS do not have identified CREBBP or EP300 gene mutations, meaning the cause of RTS is unknown\(^3\). In these cases, a geneticist might make a “clinical diagnosis” based on presenting physical characteristics of RTS, rather than genetic testing confirmation\(^6,7\).

**Things to Think About During Each Life Stage**

**INFANCY:** Low muscle tone; difficulties with feeding and drinking\(^1,6,7,14\)

**EARLY CHILDHOOD:** Weight gain; sleep issues due to breathing obstructions; gastrointestinal concerns; tethered cord; keloids\(^1,2,6,14,15,24\)

**SCHOOL AGE:** Social and engaged; continuing weight concerns; dental concerns\(^1,2,6,14,15,22,23\)

**ADOLESCENCE:** Puberty starts; behavior concerns; increased need for sleep; continuing weight concerns; discussions about pregnancy/pregnancy prevention\(^6,14,15,23\)

**TRANSITION TO ADULTHOOD:** Transition from pediatric to adult healthcare providers; employment and day programs; community living

**ADULTHOOD AND AGING:** Spine curvatures; vision issues; keloids; tumors; increased behavior concerns; frequent check-ins with specialists; aging and end-of-life planning\(^6,13,18\)

*these lists are not comprehensive

For more information, check out our RTS booklet, [www.bit.ly/2We6EUR](http://www.bit.ly/2We6EUR) or contact us at rts@cchmc.org, 513-636-4723

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25. Morgan has taught us to appreciate the smallest of victories. Every new little skill that he learns brings us a bit of hope.” Morgan’s mom and dad.