Survey of Behavior across Sex and Lifespan in Individuals with Rubinstein-Taybi Syndrome

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1. Background

RSTS Overview: Rubinstein-Taybi syndrome (RSTS) is a rare congenital disease affecting approximately 1 in 100,000-125,000 individuals.

- **Characteristics:** developmental and intellectual disability, broad and angulated thumbs and halluces, and a characteristic facial dysmorphism.[1]
- **Types:** Type 1 is due to pathogenic variants *CREBBP,* and Type 2 is due to pathogenic variants in *EP300.*[2] Some individuals do not have a variant in either gene but are diagnosed with RSTS through clinical criteria.

Behavioral Phenotype: Children with RSTS demonstrate a variety of behaviors known to impact quality of life.

- **Reported behaviors include**: attention problems, hyperactivity, self-injury, repetitive behaviors, and aggression[3]. These behaviors may emerge when children begin puberty[4].

Purpose of Study:

- Assess changes in OCD-type, anxious, abnormal-type, and daily living skills behaviors by sex and age among patients with RSTS to provide anticipatory guidance about behavioral expectations and guide medical management recommendations.

2. Methods

Participants, including family members and caretakers of individuals with RSTS, provided written consent either on the phone (online form) or in-person (paper form). Participants were recruited through the CCHMC epigenetics clinic and through RSTS Facebook support groups. We emailed four validated surveys to participants and the data was stored in REDCap.

Surveys Used:

- Yale Brown OCD Assessment (Y-BOCS) for OCD symptoms
- Spence Anxiety Scale (SCAS) for anxiety
- Aberrant Behaviors Checklist 2 (ABC-2) for abnormal behaviors
- Vineland-3 Assessment (Parent Report form) for Adaptive Daily Living Skills

Data Analysis:

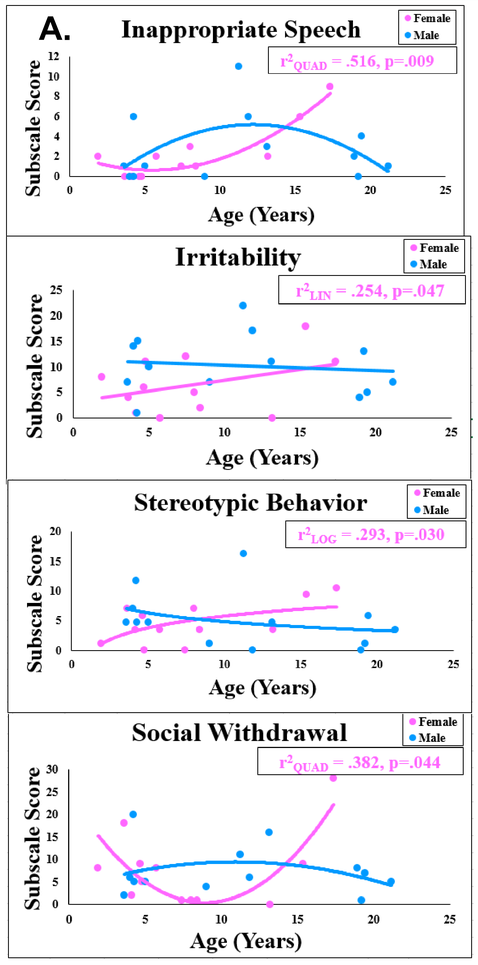
- ANOVA models to examine differences based on Age Group, Sex, and RSTS Type
- Linear and non-linear regression models to examine change in behavior with age as a continuous variable

3. Results: Demographic Information

Total Participants: 38 (age range 1.5 y.o. – 61 y.o.)

- **Sex**: Males = 21; Females = 17
- **Race**: White = 35; Multiple Races = 3
- **Ethnicity**: Hispanic = 3; Non-Hispanic = 34, Not reported = 1
- **Diagnosis**: RSTS1 = 14; RSTS2 = 10; Clinical = 14

4. Results: Age-Related Changes in Behavior By Sex



Age-related regression models showing significant behavior differences within subdomains between males and females.

A: ABC-2 assessment.
B: Y-BOCS assessment.
C: Vineland-3 assessment.
D: SCAS Assessment.

5. Conclusions

- **Females:** The severity of certain behaviors including **obsessions, compulsions, irritability, and anxiety** increase with age, especially during the post-pubescent period.
- **Males**: There is less of a predictable trajectory of behavior changes over time.
- **RSTS Type:** No differences were identified between Type 1 and Type 2; however, this may be due to small sample size.

All Individuals with RSTS: Our findings suggest anticipatory guidance should be provided to patients and their caretakers about age-related and sex-specific differences in behaviors among patients with RSTS. In addition, providers should continue to follow management guidelines and use this data as a resource when considering future interventions.

6. Future Direction

1. Include larger number of older individuals with RSTS to further learn about symptoms throughout the lifespan.
2. Replicate the study with longitudinal data to account for any cohort-related effects present.
3. Investigate how interventions (i.e. therapies, pharmacological interventions, clinical support) impact behavior changes in individuals with RSTS.

References

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