DOCUMENT SUMMARY This 2019 study on Silver-Russell syndrome provides powerful evidence for the "no normal brain" concept, showing that two different molecular causes for the same clinical diagnosis result in vastly different autistic and cognitive profiles. Critically, the research found

no association between the severity of autistic traits and intellectual ability, debunking a common ableist myth. The paper also highlights the unreliability of standardized assessments by documenting a discrepancy where parent reports of autistic traits (SRS-2) were not always confirmed by clinical observation (ADOS-2), reinforcing the need to value lived experience over single-point-in-time testing.

FILENAME `Lane_2019_RESEARCH_research_article_Silver-Russell_Syndrome_autism_IQ.md`

METADATA **Primary Category:** RESEARCH **Document Type:** research_article **Relevance:** Core **Update Frequency:** Static **Tags:** #neurodiversity, #genetics, #no_normal_brain, #autism, #IQ, #standardized_testing, #assessment_bias, #ADOS, #SRS-2, #Silver-Russell_Syndrome, #comorbidity **Related Docs:** `Kalb_2022_RESEARCH_research_article_ADOS-2_BIAS_racial_sex.md`, `Maddox_2014_RESEARCH_dissertation_ASD_SAD_assessment_eye-tracking.md`

Autistic Traits and Cognitive Abilities Associated With Two Molecular Causes of Silver-Russell Syndrome

Why This Matters to Enlitens

This paper is a scientific cornerstone for two of our most fundamental arguments. First, it is a perfect illustration of the "no normal brain" principle. By showing that the

same clinical diagnosis (Silver-Russell Syndrome) stemming from two different molecular causes (matUPD7 and 11p15 LOM) produces dramatically different cognitive and autistic profiles, it demolishes the idea of monolithic diagnostic categories. This is proof from the genetic level that diversity is the rule, not the exception.

Second, and most critically, this study provides definitive evidence to debunk the pervasive and ableist myth that autism is simply a function of intellectual ability. The finding that there was

no association between IQ and the severity of autistic traits is a powerful tool for advocacy and education. It validates our position that autism is a distinct neurotype, not a cognitive deficit. Finally, the documented discrepancy between the parent-report questionnaire (SRS-2) and the "gold-standard" ADOS-2 allows us to critique the validity of these tools, arguing that a clinical snapshot can easily miss the lived reality of a child's experience at home and school.

Critical Statistics & Findings for Our Work

- **Autism and IQ Are NOT Linked:** In both genetic subgroups, there was no significant association between the severity of autistic traits (measured by both the parent-report SRS-2 and the observational ADOS-2) and overall intellectual ability (IQ/GCA score). This finding directly contradicts the theory that the presence of autistic behaviors is increased by the degree of intellectual disability.
- **Different Profiles for the Same Diagnosis:** The two genetic subtypes of Silver-Russell Syndrome presented very differently:
    - **matUPD7 Group:** Associated with a significantly higher prevalence of autistic traits and lower intellectual ability. The mean IQ was **79.86** (borderline intellectual functioning). On the ADOS-2, **33.33%** of this group scored in the autism spectrum range. On the SRS-2, **37.50%** scored in the *severe* range for autistic traits.
    - **11p15 LOM Group:** Associated with a lower prevalence of autistic traits and intellectual ability in the average range. The mean IQ was **98.56**. On the ADOS-2, **11.11%** of this group scored in the autism range. On the SRS-2, only **10.64%** scored in the severe range.
- **Discrepancy in Testing Tools:** There was a mismatch between the parent-report and the observational test. All participants who scored above the cutoff on the ADOS-2 also scored above the cutoff on the SRS-2, showing good reliability in that direction. However, the reverse was not true: "some participants scored above clinical cut-off on the SRS-2 but clinically significant autistic behaviors were not observed during the ADOS-2".
- **Uneven Cognitive Profile NOT Found:** Despite significant differences in overall IQ between the groups, there was no evidence of an uneven cognitive profile (e.g., specific strengths or weaknesses in verbal vs. nonverbal vs. spatial skills) associated with either genetic cause.

The "No Normal Brain" Evidence

This study is a powerful case study in neurodiversity originating at the molecular level. It demonstrates that a single clinical label, "Silver-Russell syndrome," encompasses at least two distinct neurobiological pathways (11p15 LOM and matUPD7) that lead to profoundly different cognitive and behavioral outcomes. The matUPD7 subtype is associated with a high prevalence of severe autistic traits and borderline intellectual functioning, whereas the 11p15 LOM subtype is characterized by average-range IQ and lower prevalence of autistic traits. This genetic evidence directly supports our core tenet that there is no single "normal" or "abnormal" brain; rather, there is a wide spectrum of human neurological variation driven by complex biological factors.

The Failure of Standardized Testing

The study's results reveal a critical flaw in the assessment process. While the authors interpret the discrepancy between the SRS-2 (parent report) and ADOS-2 (clinical observation) as "reduced sensitivity of the SRS-2," we can frame this as the

failure of the ADOS-2 to capture the reality of a child's life. A one-hour, structured, and artificial observation (ADOS-2) may not elicit the same behaviors that parents (via the SRS-2) observe daily across multiple natural environments. This discrepancy validates our skepticism of

"gold-standard" observational tools and reinforces our belief in the primary importance of lived experience (i.e., parent/self-report and clinical interview) in assessment.

Quotes We Might Use

- **On the independence of autism and IQ:** "There was no evidence... of an association between autistic traits and intellectual ability."
- **On screening implications:** "This demonstrates that individuals with Silver-Russell syndrome should receive screening for ASD, regardless of their overall intellectual ability."
- **On the different neuro-profiles:** "Despite the same clinical diagnosis, it is clear that there are some differences in the cognitive and behavioral phenotypes associated with 11p15 LOM and matUPD7."
- **On the failure of one test to confirm another:** "...it is important to note that some participants scored above clinical cut-off on the SRS-2 but clinically significant autistic behaviors were not observed during the ADOS-2, indicating reduced sensitivity of the SRS-2 in this population."
- **On characterizing different syndromes:** "Use of autism-specific standardized assessments enables autistic traits to be characterized in detail, rather than relying upon the prevalence of clinical diagnoses alone."

Clinical Implications (The Enlitens Way)

The study's conclusions, when viewed through our lens, strongly support our approach to care.

- **Individualized Assessment is Essential:** The authors conclude that "appropriate educational strategies should be considered on an individual basis" due to the variability observed. This rejects a one-size-fits-all approach based on diagnosis alone.
- **Screen for Autistic Traits Regardless of IQ:** The finding that autism and IQ are not linked leads to the crucial recommendation that clinicians should screen for ASD regardless of a person's intellectual ability. This combats ableist assumptions that high-IQ individuals can't be autistic or that autistic traits in low-IQ individuals are just part of their intellectual disability.
- **Support Should Target Specific Needs:** The finding that the matUPD7 group had greater difficulty with Restricted and Repetitive Behaviors (RRB) "demonstrates the need for support with RRB in individuals with matUPD7," highlighting that support should be tailored to specific traits, not just a general diagnosis.