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Improving Speech, Language and Orofacial Outcomes in Russell–Silver Syndrome: A Case Study on Integrated Pediatric Care

Varun Singh[1], Mansi Doshi[2], Nandini Tiwari[3]

[1]Assistnat professor, Department of Audiology and Speech Language Pathology, Dr Shakuntala Misra National Rehabilitation University, Lucknow
[2]Assistant professor, Department of Audiology and Speech Language Pathology, Dr Shakuntala Misra National Rehabilitation University, Lucknow
[3]Audiologist & Speech Language Pathologist Grade II, Department of Audiology and Speech Language Pathology, Dr Shakuntala Misra National Rehabilitation University, Lucknow

Abstract

Background

Russell–Silver Syndrome (RSS) is a rare genetic imprinting disorder characterized by intrauterine and postnatal growth restriction, craniofacial dysmorphism, and variable neurodevelopmental outcomes. Communication difficulties, particularly in expressive language, are underreported despite affecting nearly half of the affected individuals.

Case Presentation

This case report describes a 3-year, 11-month-old female with genetically confirmed RSS due to maternal uniparental disomy of chromosome 7 (mUPD7). The child exhibited global developmental delays, particularly in expressive language and motor skills, along with oromotor anomalies including a high-arched palate and velopharyngeal dysfunction. Standardized assessments revealed a marked receptive–expressive language gap and mild intellectual disability.

Intervention

A multidisciplinary, personalized therapy program was carried out over 12 months. It included language facilitation strategies, PROMPT and oral placement therapy, augmentative and alternative communication (AAC) supports such as PECS and core vocabulary boards, and parent-mediated social communication training. Therapy sessions were held three times a week for 30 minutes each, with close coordination among the interdisciplinary team.

Outcomes

Post-intervention assessments demonstrated significant improvements in receptive (from 20–22 to 30–33 months) and expressive language (from 14–16 to 27–30 months). Functional two-word combinations emerged, hypernasality reduced, and joint attention and social engagement improved. Cognitive function remained stable, and parent reports noted enhanced functional communication and reduced frustration behaviors.

Conclusion:

This case underscores the importance of early genetic diagnosis and tailored, interdisciplinary interventions in treating speech and language delays in RSS. Well-structured, multimodal therapy

methods, caregiver participation, and regular outcome monitoring greatly improve communication results in rare neurodevelopmental disorders.

Keywords: Russell–Silver Syndrome, expressive language delay, PROMPT therapy, oromotor intervention, augmentative communication, case report

INTRODUCTION

Russell–Silver syndrome (RSS) is a clinically diverse imprinting disorder characterized by intrauterine growth restriction (IUGR), short stature after birth, distinctive craniofacial features, and feeding difficulties. First identified independently by Silver in 1953 and Russell in 1954, the syndrome has an estimated prevalence of 1 in 30,000 to 1 in 100,000 live births worldwide (Wakeling et al., 2017). The genetic causes of RSS vary: about 35–50% of cases involve loss of methylation at the imprinting control region 1 (ICR1) on chromosome 11p15.5, 5–10% result from maternal uniparental disomy of chromosome 7 (mUPD7), and rare cases are due to other chromosomal anomalies, such as duplications or deletions on chromosome 6 (Eggermann, 2010; Abu-Amero et al., 2008; Begemann et al., 2015). The phenotypic features include a triangular facial shape, body asymmetry, fifth-finger clinodactyly, limb-length differences, and a high-pitched voice, along with nonspecific neurological and cognitive outcomes.

Although the somatic and growth characteristics of RSS are well documented, its neurodevelopmental and communicative profile remains underexplored. Early oromotor dysfunction—attributable to high-arched palate or micrognathia—may hinder babbling and speech sound development (Price et al., 2002). Several studies report a high rate of speech and language delays: Wakeling et al. (2017) found that up to 50% of children with RSS show significant delays in expressive language, while a retrospective cohort study by Haug et al. (2020) observed that 40% of subjects exhibited receptive–expressive language discrepancies at school age. Morison and Reeve (2003) found that 60% of individuals showed mild to moderate intellectual challenges, which were linked to a smaller vocabulary and slower development of sentence structure. Cassidy and Allanson (2010) also noted that behaviors like difficulty focusing on shared activities or engaging in imaginative play can make communication even tougher for these individuals.

Recent studies using brain imaging and neurophysiological techniques suggest that children with Russell-Silver Syndrome (RSS) often exhibit unique brain development patterns, including differences in the structure of the two brain hemispheres. These differences may make it more difficult for them to process language and sounds effectively (Sato et al., 2019). For example, Yamamoto et al. (2022) used MRI scans and found that the protective coating around nerves in the temporal and parietal lobes—key regions for understanding language and memory—develops more slowly in children with RSS. This could explain why they have difficulty grasping language and retaining information. Similarly, Henkin et al. (2021) conducted brain activity tests (called ERP studies) and observed delays in a brain signal called P300, indicating challenges in shifting attention and remembering spoken information. Together, these findings demonstrate a biological basis for the language delays in RSS and underscore the importance of early testing of cognitive and language skills.

Because many factors are involved, identifying these issues early and adopting a team-based approach is essential. Tools like the Receptive–Expressive Emergent Language Scale (REELS), Developmental Screening Test (DST), Vineland Social Maturity Scale (VSMS), and COM-DEALL provide a solid starting point for developing personalized therapy plans (Bzoch et al., 2003; Jethwani et al., 2012). Malin's Intelligence Scale for Indian Children (MISIC) is also useful for gaining a more complete understanding

of a child's cognitive abilities (Malin, 1971).

Still, there are few detailed studies that combine genetic, cognitive, and language data to show how interventions work for kids with RSS. This report aims to fill that gap by sharing the story of a child with genetically confirmed RSS, providing a comprehensive speech and language evaluation and a customized, team-based intervention plan. In doing so, it contributes to the growing understanding of how to support communication development in children with this syndrome.

Case Description

Patient Profile and Birth History

The subject of this case study is a 3-year, 11-month-old girl born to non-consanguineous parents from a middle socioeconomic background in an urban Indian setting. The antenatal history was largely uneventful, with routine prenatal scans and maternal nutrition within normal limits. However, fetal growth restriction was first noted during the third trimester, raising concerns of intrauterine growth restriction. The child was delivered via an elective cesarean section at term with a birth weight of 1.9 kg, classifying her as small for gestational age (SGA). Apgar scores were 8 and 9 at one and five minutes, respectively. The neonatal period was mainly characterized by transient feeding difficulties, which resolved with minimal intervention. There was no history of hypoxia, seizures, or admission to a neonatal intensive care unit.

Developmental Milestones

Developmental milestones showed a global delay, especially in gross motor and language skills. Head control was achieved around 8 months, independent sitting at 24 months, and walking without support only at 36 months. Fine motor skills like holding a crayon and self-feeding appeared by approximately 3.5 years. Social milestones such as social smiling and stranger anxiety emerged by 9 and 18 months, respectively. Language development was notably delayed. Canonical babbling started at 9 months but did not develop into variegated babbling. Her first meaningful word, "mama," was spoken around 2.5 years. At the initial evaluation, her expressive vocabulary consisted of fewer than 10 single words, and she mainly used gestures to communicate.

Genetic and Medical Findings

Due to the presence of dysmorphic features (triangular face, micrognathia and limb asymmetry), delayed growth, and speech-language concerns, a genetic evaluation was performed at 5 months old. Cytogenetic microarray analysis identified a heterozygous duplication (22.418 Mb) at 6p25.3p22.3 and a deletion (1.209 Mb) at 6p25, both classified as pathogenic. Karyotyping with GTG banding (450-band resolution) revealed 46, XX, 6p+inv(9), indicating a pericentric inversion of chromosome 9 and a structural abnormality on the short arm of chromosome 6. These findings, along with the clinical presentation, confirmed a diagnosis of Russell-Silver Syndrome. There was no family history of genetic disorders, learning disabilities, or developmental delays. Audiological and ophthalmological evaluations were unremarkable.

Parental consent for publication and clinical documentation was obtained by the institutional ethical guidelines for pediatric case reporting.

Speech & Language Assessment

A comprehensive speech and language assessment was conducted over two sessions using a combination of standardized tools and clinical observation. The Receptive–Expressive Emergent Language Scale (REELS-3) was administered to assess language functioning. Receptive language abilities were found to

be at the developmental level of 30–33 months, while expressive language skills corresponded to an age equivalent of 14–17 months, highlighting a significant receptive–expressive discrepancy. The child demonstrated comprehension of simple commands and familiar object names but had difficulty following two-step instructions and understanding abstract concepts.

Expressive language output was limited to approximately 10–12 functional single words (e.g., "mama," "ball," "milk") and several context-bound vocalizations. No spontaneous two-word combinations were observed. Pragmatic skills were also underdeveloped: the child rarely initiated joint attention, showed limited response to name, and preferred one-on-one adult interaction over peer play. Reynell's developmental levels placed her at Level 3 (single-channel attention), indicating an early pre-linguistic stage of communicative engagement.

An oral peripheral mechanism examination revealed intact articulators; however, the palatal vault was high-arched, and the soft palate exhibited reduced elevation during phonation tasks. Mild dental malocclusion and spacing were noted. Diadochokinetic rates (AMRs/SMRs) for bilabial and alveolar sounds were within age-appropriate norms, suggesting relatively preserved speech motor planning. However, hypernasality was detected during speech tasks, indicating velopharyngeal insufficiency that could compromise speech intelligibility.

In sum, the speech and language profile indicated a mixed receptive–expressive language delay with contributory oral–motor anomalies. These findings were consistent with the communicative phenotype described in RSS literature. The assessment outcomes informed a targeted intervention plan involving oromotor strengthening, early language facilitation strategies, and augmentative and alternative communication (AAC) supports.

Oro Motor and Articulation Examination

An oral peripheral mechanism examination revealed intact and symmetrical external structures, including lips, jaw, and tongue. However, intraoral examination showed a significantly high-arched palatal vault, which is often associated with syndromic craniofacial anomalies and may contribute to resonance disturbances. The soft palate displayed hypomobility with reduced elevation during voluntary phonation and reflexive tasks (e.g., gag reflex and blowing), suggestive of velopharyngeal dysfunction. The uvula appeared short and slightly bifid. Tongue mobility was within functional limits, with normal lateralization, protrusion, and elevation.

Dentition was characterized by mild anterior open bite and generalized spacing of upper and lower incisors, which may influence articulatory placement. Bite alignment was suboptimal, suggesting a mild Class II malocclusion. These structural variations could contribute to compensatory articulatory strategies and reduced clarity of speech.

Diadochokinetic (DDK) testing for alternating motion rates (AMRs) and sequential motion rates (SMRs) revealed bilabial (/pa-pa-pa/) and alveolar (/ta-ta-ta/) patterns within expected range for age, with good rhythm and consistency. However, palatal sounds (e.g., /ka/) were produced with reduced force and increased nasality. Speech tasks involving pressure consonants (e.g., /p, b, t, d/) demonstrated nasal air escape, confirming incomplete velopharyngeal closure during articulation.

Resonance assessment during connected speech revealed mild to moderate hypernasality, which became more pronounced during production of longer utterances. Nasal emissions were perceptible during repetition of high-pressure phonemes. Although articulation errors were minimal due to the child's limited phonetic inventory, imprecise production and weak oral pressure contributed to reduced speech intelligibility.

These findings underscore the presence of velopharyngeal insufficiency likely contributing to nasality and reduced oral resonance, which are commonly reported in children with RSS. The oromotor deficits observed necessitate focused intervention to strengthen soft palate function and improve intraoral air pressure regulation, which is critical for maintaining speech clarity.

Cognitive and Socioadaptive Function

The child underwent cognitive assessment using the Developmental Screening Test (DST) and the Vineland Social Maturity Scale (VSMS), both standardized tools commonly used in developmental pediatrics and speech-language pathology in the Indian context. The composite developmental quotient (DQ) obtained from the DST indicated a mild intellectual disability, with an estimated DQ ranging between 50–69. This score reflected significant developmental lags in reasoning, problem-solving, and task persistence, particularly in the context of structured play and attention-based activities.

The VSMS revealed a mild delay in socioadaptive behavior, consistent with the cognitive findings. The child's social age was approximately 30 months, as compared to a chronological age of nearly 48 months. Specific delays were noted in the domains of self-help dressing, self-direction, and occupation (e.g., using tools or engaging in age-appropriate tasks like stacking, coloring). The communication and socialization subdomains highlighted difficulties in initiating interactions, making age-appropriate requests, and using symbolic gestures, although the child responded well to familiar adults and routine-based environments.

During clinical observation and play-based interaction, the child demonstrated parallel play but rarely engaged in cooperative play. She responded to social stimuli such as smiling and facial expressions but exhibited limited sustained eye contact and inconsistent name response. Behavioral characteristics included short attention span, high distractibility, and a preference for solitary or adult-facilitated activities. Mirror recognition and object permanence were present, indicating intact basic cognitive schema. Simple turn-taking routines could be introduced with prompting, but generalization was limited.

The overall cognitive and socioadaptive profile was indicative of a global developmental delay with a significant impact on communication readiness. These deficits underscored the need for structured, routine-based, and visually supported learning strategies that could scaffold higher-order language, play, and adaptive behaviors. Parental counseling emphasized the importance of enriched language exposure, predictable routines, and task-specific reinforcement strategies to enhance social learning and cognitive flexibility.

Intervention

Based on the comprehensive assessment findings, a multidisciplinary, individualized intervention plan was developed to address the child's unique speech, language, oromotor, cognitive, and social needs. The overarching goals of intervention were to enhance receptive and expressive language abilities, improve velopharyngeal function, promote joint attention and play-based communication, and support the child's overall developmental trajectory.

Therapy commenced on 09 May 2024, with a frequency of three 30-minute sessions per week, delivered by a certified speech-language pathologist. Therapy sessions were designed to be child-centered, goal-specific, and family-inclusive, using evidence-based practices.

1. **Language Intervention**

- **Developmental Language Facilitation:** Focused on modeling, expansion, and recasting to increase vocabulary and sentence length. Structured routines were embedded into sessions (e.g., snack time, toy play) to provide contextual cues.

- **Milieu Teaching Techniques:** Included incidental teaching and time delay strategies to create communicative temptations and encourage spontaneous verbalizations.
- **Use of Visual Supports:** Picture schedules and thematic vocabulary boards were used to enhance comprehension and reduce cognitive load.

2. **OroMotor and Resonance Therapy**

- **PROMPT (Prompts for Restructuring Oral Muscular Phonetic Targets):** Applied to address motor speech planning and improve articulatory precision. Targets focused on labial and lingual sounds with gradual complexity.
- **Oral Placement Therapy (OPT):** Included blowing, lip rounding, tongue elevation, and soft palate stimulation exercises to enhance intraoral strength and mobility.
- **Resonance Training:** Emphasis on improving velopharyngeal closure through auditory feedback, tactile cueing, and minimal pair practice involving pressure consonants.

3. **Augmentative and Alternative Communication (AAC)**

- **Picture Exchange Communication System (PECS) - Phase I & II:** Introduced as a parallel support system to promote functional requests and reduce communicative frustration.
- **Core Vocabulary Boards:** Provided for use at home and in therapy to encourage consistent symbolic communication.

4. **Play and Social Communication Training**

- **Joint Attention Routines:** Activities such as turn-taking games, rolling a ball, and book-sharing were used to build social reciprocity.
- **Symbolic Play Development:** Pretend play with dolls, animals, and everyday objects was encouraged to scaffold abstract thinking and language generalization.
- **Parent-Implemented Strategies:** Caregivers were trained to use modeling, parallel talk, and contingent responding during daily routines.

5. **Interdisciplinary Collaboration**

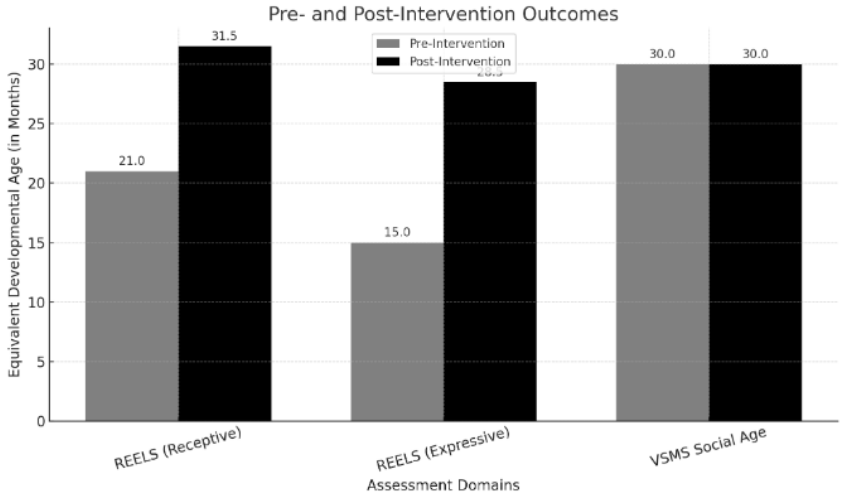
- Weekly coordination was maintained with a developmental pediatrician (for growth, feeding, and medical monitoring), a genetic counselor, and an occupational therapist (for fine-motor coordination, ADLs, and sensory integration).
- Monthly review meetings were held with the family to reassess goals and therapy outcomes. The intervention approach emphasized consistency, reinforcement, and generalization across settings, with a strong focus on caregiver involvement to support long-term developmental gains.

Outcome

After twelve months of intervention:

A visual comparison of pre- and post-intervention developmental age equivalents for receptive and expressive language, as well as socioadaptive functioning, is shown in **Figure 1**. The chart illustrates marked improvement in both language domains, while social maturity remained stable over the intervention period.

Figure 1: Bar chart showing pre- and post-intervention developmental age equivalents (in months) for REELS receptive and expressive language subscales and VSMS social age.



- Receptive Vocabulary increased to approximately 45 words; followed simple two-step verbal commands 70% of the time.
- Expressive Skills: Emergence of consistent two-word combinations (e.g., "want juice," "more ball"); total expressive lexicon approximately 30 words.
- OroMotor Function: Improved velopharyngeal closure noted on /p, b, t, d/ tasks; reduced hypernasality.
- Social Communication: Increased initiation of joint attention (e.g., pointing to objects for requests); sustained eye contact for 3–5 seconds.
- Parents reported improved functional communication at home and daycare, with decreased frustration behaviors.

Table 1: pre- and post-intervention outcomes, based on the qualitative improvements

Assessment Tool	Pre-Intervention Score	Post-Intervention Score	Notes
REELS (Receptive)	20–22 months	30-33 Months	Improved to ~45 words
REELS (Expressive)	14–16 months	27-30 Months	Two-word combinations emerged
DST & VSMS (Composite IQ)	50–69	50-69	Stable cognitive function

Discussion

This case highlights the multifactorial nature of communication delays in children with Russell-Silver Syndrome (RSS), emphasizing the interplay of genetic, anatomical, cognitive, and behavioral factors. The child in this report showed classic signs of Russell-Silver Syndrome (RSS), including being born smaller than expected, distinct facial features, and developmental delays. These traits match what researchers like

Wakeling et al. (2017) and Haug et al. (2020) have seen in larger groups of kids with RSS. The child also had physical challenges, like a high-arched palate, limited movement in the soft palate, and misaligned teeth, which caused a nasal-sounding speech (hypernasality) due to air escaping through the nose during talking. This aligns with what Price et al. (2002) noted about speech issues in RSS. Targeted therapies, like PROMPT (a hands-on approach to guide mouth movements) and exercises to improve voice resonance, helped the child speak more clearly and reduce the nasal tone. These results back up studies, like Schlüter et al. (2009), that stress the need for early speech therapy to address mouth and speech problems in RSS.

The child's language skills followed a common pattern seen in genetic syndromes: understanding language (receptive skills) developed faster than speaking (expressive skills). With therapies like language facilitation and visual aids, the child made progress in both areas. Tools like the Picture Exchange Communication System (PECS) and core vocabulary boards—simple visual tools to support communication—were game-changers, helping the child build verbal skills. This mirrors findings by Girolametto et al. (1997) and Brady et al. (2016), who showed that these augmentative communication methods can boost speaking abilities and even reduce frustration-related behaviors in kids with developmental delays.

The child also got better at paying attention to others, playing imaginatively, and interacting socially, thanks to structured play activities and therapy involving parents. These improvements echo what Koegel & Koegel (2006) found about using natural, everyday interactions to help kids communicate better. Parents played a huge role in making sure these gains stuck outside of therapy sessions, which lines up with research by Roberts and Kaiser (2011) showing that when parents are actively involved in language therapy, kids make bigger strides in speaking.

This case also highlights why catching RSS early and confirming it with genetic testing is so important. Knowing the genetic cause helped the team set realistic goals and coordinate care across different specialists. Regular input from therapists, doctors, and others ensured the therapy kept pace with the child's changing needs. Eggermann et al. (2010) pointed out that early genetic diagnosis helps plan therapies and predict how a child might develop over time.

All in all, this case shows that kids with RSS can make real progress in communication with personalized, evidence-based support that involves a team of experts. Looking ahead, more long-term studies are needed to track progress and better understand how brain development affects communication in RSS, which could guide better care strategies.

Conclusion

This case underscores the value of spotting RSS early, using genetic testing, and bringing together a team to tackle speech and language delays. Tailored therapies—like visual communication tools, mouth movement exercises, and parent involvement—led to big improvements in the child's ability to communicate and connect with others. It highlights the importance of developing therapy plans that encompass a comprehensive view of a child's needs, rather than focusing solely on a single issue. While more research is needed, stories like this are key to shaping early support for rare conditions like RSS.

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