

## Research Research

# Neurodevelopmental Changes and Emotional Burden of the Primary Caregiver: A Case Report of Robin Sequence, Corpus Callosum Dysgenesis and Comorbidities

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**Abstract**

The aim of this study was to describe the evolution of the neurodevelopment of an atypical Robin Sequence clinical case and the impact of emotional burden on the primary caregiver. Female participant, age 01 years and 08 months, with Robin Sequence, typical characteristics and global developmental delay. Two development assessments were carried. The results showed an evolution of adaptive behavior in the domains of interpersonal socialization, personal autonomy, receptive and expressive communication. Five psychiatric indicators of body activity showed an improvement. Acquisition of sentence comprehension and beginning of primitive imitation activities. The caregiver's emotional burden showed a reduction in indicators and degrees of intensity. The child's repertoire of skills, although showing a restricted evolution, showed the importance of early diagnosis for rehabilitation and minimizing the impact on the family routine and the stressful condition of the primary caregiver.

**INTRODUCTION**

The Pierre Robin sequence (PRS), is characterized by the clinical triad of congenital micrognathia, glossoptosis, and airway obstruction frequently associated with cleft palate [1-4]. This condition is highly heterogeneous, with a wide range of pathogenic and phenotypic differences [1]. Most cases are associated with syndromes of chromosomal origins or additional abnormalities, showing systemic involvement, but they can also occur as an isolated entity [3]. PRS has an estimated prevalence of 1 in 8,500 to 14,000 live births, although recent studies reported an increased incidence of 1 in 3,000 to 5,600 [4]. This condition was known as "Pierre Robin Syndrome" for almost half a century, however, since the same clinical findings could result from the identification of multiple etiologies, the term "syndrome" has been replaced by "sequence" [2]. Cases of syndromic origin are frequently more severe due to involvement of respiratory

airways and associated dysphagia, among other disorders involving neurodevelopment [1,5].

The complexity and severity of pediatric genetic conditions pose significant challenges to families, especially due to inconclusive diagnostics, the necessity of constant care throughout life, and the search for effective health services and treatments. These situations can lead to "side effects" on the parents' physical and psychosocial well-being [6]. As a rule, parents are required to alter their work patterns, affecting income and household responsibilities, as well as to dedicate themselves extensively in order to acquire specialized health knowledge of care, skills and resources and be able to meet their child's needs [7].

Studies show that early identification (e.g. during prenatal care) of possible anomalies effectively improves interdisciplinary assistance, including psychological counseling [8].

In light of the limitations that the exceptionality might entail, interferences in the education process of children are frequently observed, sometimes leading to polarized actions of rejection and overprotection [9]. Children with craniofacial and neurological anomalies tend to manifest impairments in development, which can be evidenced by the difficulty in the acquisition of certain skills [5,10], which often concerns families and lead to negative psychological effects.

According to Razera, Trettene, and Tabaquim [11], the impact of atypical clinical conditions on the parents and/or caregivers is frequently observed and is characterized by an emotional overwhelm resulting from the child's chronic condition. Assessing the resources of children with PRS is crucial for implementing strategies that will promote the adjustments needed to acquire neurodevelopmental skills, as well as for the adequate care for parents in order to reduce negative psychological effects and, thus, maintaining the appropriate family balance needed to improve the quality of life of the population with exceptional and their caregivers.

Given the above, the objective of this study was to describe the evolution of the neurodevelopment of an atypical PRS case and the impact of its associated emotional burden on the primary caregiver.

## METHODS

This is a longitudinal, retrospective, and descriptive clinical case report with quantitative and qualitative approach of a female child diagnosed with PRS, observed at 11 and 20 months of age.

### Clinical history

Unintended and unwanted pregnancy. The mother had type 2 diabetes, with difficulties in controlling glycemic indexes (insulin dependent). On the 4th month of gestational age (GA), it was observed that the placenta had a poor blood flow; morphology ultrasound performed at the end of the 5th month of GA indicated that the baby could have problems. The study subject had a premature birth with 36 weeks of GA by cesarean section, with birth weight, length, and head circumference of 1,774 kg, 28,5 cm, and 34 cm, respectively. Due to unstable oxygen saturation, the baby was admitted to the Neonatal Intensive Care Unit (NICU) for 30 days; this was followed by another 40 days in the Special Care Unit (SCU), due to breathing and feeding difficulties, during which a gastrotomy was performed. Also during this time, the baby was diagnosed with PRS after the identification of micrognathia, glossoptosis, airway obstruction, incomplete post-foramen fissure, and moderate to severe dysphagia. Other craniofacial and body deformities have been described, including: asymmetrical skull, anteverted nostrils, frontal bossing, bilateral grade III microtia, bilateral external auditory canal atresia, mild microcephaly; and malformation of lower limbs, shortening of upper limbs, bilateral pseudocamptodactyly of the 1st, 2nd and 3rd digits, pre-axial polydactyly (feet), sacral agenesis, grade-4 femoral deficiency (vestigial tibia). Cardiac alterations were present in the form of a 6.1 mm *ostium secundum* atrial septal defect. Pulmonary hyperflow was detected, with direct hemodynamic repercussion and mild dilation of the right chambers. Regarding neuropsychomotor development, sagittal balance occurred around the 7th month; sitting with support at 10

months; grabbed objects with 6 months, although reflex activity (palmar grasp reflex) was still noticeable; and, around the same time, attempts to turn the body to both sides. Communication is deficient, only beginning to emit sounds.

Cardiological care and follow-up have been carried since birth. There is an indication for surgical intervention that has not been performed yet. The cleft palate has not yet been repaired due to health restrictions, and aggravates the feeding and respiratory difficulties, even during sleep time.

Audiological evaluation by means of brainstem auditory evoked potential (BAEP) showed an increased absolute latency for the age group of the wave V in the right ear, at an intensity of 90 dBnHL. Using the same intensity, there was no neural response in left ear. At the electrophysiological threshold of the right ear, the responses were inconclusive (with air and bone conduction at 80 and 40 dBnHL, respectively), indicating central hearing loss of brain stem origins and non-peripheral. The baby has physiotherapy and speech-language treatments twice a week; nutrition therapy (monthly); and regular medical follow-up with neurologists, cardiologists, and orthopedists.

### Procedures

This study was developed in accordance with Resolution 466/2012 of the National Commission for Research Ethics (CONEP), and approved by the Committee for Ethics in Research (CEP) of Bauru School of Dentistry - University of São Paulo (FOB-USP), registered at CAAE n° 42356815.1.0000.5417. The responsible family member signed the Informed Consent Form (ICF).

The assessment tools used to investigate and characterize the child's conditions and its impact on her caregiver (mother) were: medical history; Vineland Adaptive Behavior Scale [12]; Alarm Distress Baby Scale (ADBB) [13]; The Informal Caregiver Burden Assessment Questionnaire [14]; Family Behavior Assessment procedure [15]; Operational Portage Inventory (OPI) [16]; Denver Developmental Screening Test II (DDST-II) [17]; Early Language Milestone Scale (ELM Scale) [18]; Gesell Developmental Assessment (GDA) [19]; MacArthur-Bates Communicative Development Inventories (CDIs) [20].

The data obtained through the scales were recorded in spreadsheets using Microsoft Office Excell 2007®. Quantitative results were submitted to inferential statistics for correlation analysis. Qualitative analysis was performed based on the available literature.

## RESULTS

Developmental milestones from birth to age 1 were evaluated at 11 months (2016) and at 20 months of age (2017). The comparative results are shown in Table 1.

Comparatively, in 2016 the child's repertoire was 22.12% versus 50.95% in 2017, an increase of 43.4%. Individual analysis of each domain of development showed an increase of 20% in Interpersonal Socialization, 16.67% in Personal Autonomy, 30% in Receptive Communication, and 20% in Expressive Communication. There was no record for Motor skills (fine or gross).

**Table 1:** Comparative results between two assessment periods.

VINELAND ADAPTIVE BEHAVIOR SCALE												
Domains	Motor Skills			Communication			Socialization			Daily Living Skills		
	2016	2017		2016	2017		2016	2017		2016	2017	
	>1 y/o	>1 y/o	1 y/o	>1 y/o	>1 y/o	1 y/o	>1 y/o	>1 y/o	1 y/o	>1 y/o	>1 y/o	1 y/o
Gross	12,5	50	0									
Fine	12,5	37,5	0									
Receptive				50	60	30						
Expressive				30	50	20						
Interpersonal							56	87,5	20			
Play and Leisure							16	41,67	0			
Personal										0	30	16,67
Domestic												0
Σ %	12,5	43,75	0	37,5	55	25	39,3	64,58	10	0	30	8,33
										22,12	50,95	10,83

In the ADBB, eight indicators were analyzed. In 2016, six “possible indicators” (score of 1) were observed on the pathological character of the child’s reactions versus five in 2017, indicating an improvement in Activity Level; while the pathological doubt aspect remained for facial expression, eye contact, self-stimulation, vocalizations, and relationship with the observer. The parameters of briskness of response and attraction to stimuli were preserved in both observational periods, indicating the absence of pathological factor in these two categories. Thus, the overall results pointed to a positive response from the child to environmental stimulation, since in the score in the most critical categories (2, 3 and 4), was zero (Table 2).

Table 3 shows the results obtained in the two assessments (at 11 and 20 months) regarding ELM, DDST-II and GDA.

As for MacArthur-Bates CDI, the subject scored 66.6% of the “first signs of understanding” item in 2016 and 100% in 2017. In the “phrase comprehension” item, the scores were 14% in 2016 and 67.5% in 2017. She did not score in any of the semantic categories in 2016, nine months later in the reassessment, she scored 8.33% in “understanding and speech for sounds of things and animals”, and comprehension for “toys” (40%), “food and drink” (9.37%), “body parts” (10.52%), “people” (38.88%) and “action words” (3.57%).

The OPI results did not reach the recommended minimum (at least 80% of the tasks) in the domains of “socialization”, “language”, “self-care” and “motor” in neither of the assessments. At the 20 months assessment, the subject obtained 100% in “cognitive development” for the age of 0-1 year and 22.2% for the age of 1-2 years.

The analysis and comparisons of the results obtained shows the development of multiple skills, especially linguistic ones, with emphasis on auditory-receptive and visual functions. The results also show that motor development is severely affected, which was expected given the complexity of upper and lower limb malformations.

With regards to the emotional burden of the caregiver, the first assessment (2016) showed that 50% of the indicators had moderate to severe intensity (scale 3); which is representative of the many challenges of dedication and care, imposed by the specificity of the child’s diagnostic condition. In the second assessment, there was a reduction in indicators (33%) as well as intensity (scale 2 and 3) of signs of overwhelm (Table 4).

The overall numbers of emotional burden assessment, that implicate in psychological aspects, indicated “mild intensity” for emotional and financial overload, personal life implications, and response to the child’s need of special care. However, a “high impact” (scale 5) was observed on the perception of their effectiveness, family support, and satisfaction with the role of caregiver (Table 5).

According to the Family Behavior Assessment procedure mediated by the mother, 84.62% of actions indicators were classified as systematic positive parenting practices, reflecting the appropriate use of reinforcers as a mean of improving the child’s social and affective behavior.

## DISCUSSION

The present case-report study identified, through a longitudinal follow-up, the developmental achievements of a child with severe sequelae, as well as the psychological burden faced by the main caregiver, and compared them to those expected for the first year of life.

The birth of a child with disabilities might have impacts at different levels; among other factors, the moment of diagnosis is crucial for the history and evolution of the case [8]. This case showed that, despite having an early diagnosis and multidisciplinary support for the child’s rehabilitation, the evolution was slow, indicating an importance of intrinsic trophic factors for the stimulated functions, development, and learning. As for maternal support, it shows that families, especially the main caregiver, can display a wide range coping results, both in their available resources to deal with unexpected situations, as well as in the way they face the challenges of a potentially stressful

**Table 2:** Comparative results between two assessment periods for sustained relational withdrawal using the ADBB.

Items	ALARM DISTRESS BABY SCALE									
	0		1		2		3		4	
	2016	2017	2016	2017	2016	2017	2016	2017	2016	2017
Facial Expression			X	X						
Eye Contact			X	X						
Activity Level		X	X							
Self-stimulation			X	X						
Vocalizations			X	X						
Briskness of response	X	X								
Relationship with observer			X	X						
Attraction to stimuli	X	X								
	2016	6 points = 81% of possible sustained relational withdrawal, without psychiatric factor								
TOTAL	2017	5 points = 62,5% of possible sustained relational withdrawal, without psychiatric factor								

**Table 3:** Comparative results for ELM, DDST-II and GDA for assessments at 11 and 20 months of age.

	Áreas	Faixa etária de desempenho	
		11 months	20 months
ELM	Auditory Expressive	4 months	8 months
	Auditory Receptive	4 months	11 months
	Visual	10 months	18 months
DDST- II	Personal-social	2 months	5 months
	Fine motor adaptive	3 months	7 months
	Language	6 months	8 months
	Gross motor	3 months	4 months
GDA	Adaptative	9 months	11 months
	Gross motor	3 months	4 months
	Fine motor	3 months	7 months
	Language	6 months	8 months
	Personal-social	2 months	5 months

**Table 4:** Comparative results between the two assessments.

N	Difficulties with the child's care	Intensity							
		0		1		2		3	
		2016	2017	2016	2017	2016	2017	2016	2017
1	How much does it affect you?			X	X				
2	Self-care dependency								
3	Slow development						X	X	
4	Limited self-expression	X	X						
5	Mood swings (frequent crying)	X	X						
6	Highly dependent				X			X	
7	Need of intensive care							X	X

situation like having a child with chronic health problems or disabilities.

On the Vineland Adaptive Behavior Scale, the child showed acquisition of gross skills compatible with the expected for first year of life (0 to 1 years). Comparing the repertoires of both assessments events, there was an increase in the number of achievements; however, the numbers were still closer to the

initial age group (0 to 1 years), showing a limited development, given that this period is characterized by quick and constant changes in motor skills; this can be justified by the severe sequelae.

According to the World Health Organization (WHO), the global prevalence of developmental disturbances and mental and behavioral disorders in childhood is 10 to 20%. Still, accurate

**Table 5:** Results for the indicators of emotional burden.

Caregiver burden – GENERAL AND PSYCHOLOGICAL ASPECTS					
Level	1	2	3	4	5
Sobrecarga emocional	100%				
Implicações vida pessoal	100%				
Sobrecarga financeira	100%				
Reações às exigências	100%				
Percepção sobre a eficácia					100%
Apoio familiar					100%
Satisfação com s/ papel					100%

diagnosis has been a great challenge in clinical practice, given the heterogeneity of clinical conditions and the diagnostic particularities for different medical, psychological, social, and educational specialties. In the present study, the ADBB was used as an instrument for psychiatric indicators; the results showed no signs of association between the child's impaired neurological development and pathological profile of mental disorder. The significant deficiency observed, initially permeated by great family anguish, was better understood at a latter moment, as noted by the change in expectations (between the two moments of assessment) regarding the slower processes in the various motor and social responses; this contributed to foster the relationship of attachment and favor the child's mental health.

Thus, the decrease in the signs of caregiver burden observed in the second evaluation moment, directs the comprehension about the psychological organization reflected in daily practice, such as in the organization of day-to-day activities and in the guidance of primary and secondary caregivers. Studies of the caregiver's burden by Pimenta [21], showed that this role is often given to females and the responsibility is not always spontaneous; this is influenced by affectiveness, closeness of kinship, availability, among others.

In this case study, the primary and responsible caregiver was the child's mother. She was responsible for making decisions and organizing the child's daily activities, as well as guiding the "secondary" caregiver, the maternal grandmother. This collaboration, as a support network, is essential for the family to organize itself in a positive way and to form bonds that will structure the child's life through affective self-esteem, and cherish development. The overall burden, in psychological aspects and coping with the situation of special care, was classified as having a "mild intensity" for emotional and financial aspects, because, thanks to this supportive network, access to medical monitoring and necessary care for the child was guaranteed.

The implications of emotional overwhelm were evidenced in personal life aspects and in reactions to the needs of constant care; it was also observed when recognizing whether or not the caretaker was being effective, and meeting family and personal expectations. The break in the stereotype of the desired child, previously idealized and built in the parents' imagination, can be compensated by mechanisms of full dedication to the care of the child and by the high expectations for perfect performance of daily activities. According to Oliveira and Poletto [22], this mechanism, in its transient form, can be positive, however,

chronicity, as a rule, culminates in psychological dissatisfaction and suffering.

According to the Family Behavior Assessment procedure mediated by the mother, actions indicators were classified as systematic positive parenting practices, reflecting the appropriate use of reinforcers as a mean of improving the child's social and affective behavior. According to Gomide, Salvo, Pinheiro, Sabbag [23], parental monitoring is a set of parental behavior that includes attention to the activities and ways of adaptation of their children, which favor the development of prosocial behaviors. The favorable indexes of psychological overwhelm observed, combined with the positive monitoring, reflect a stable family condition that enables verifying the child's real limitations, and the positive possibilities of multidisciplinary rehabilitation work as a protective factor for development.

In the caregiver emotional burden assessment results, there was a significant percentage of indicators with moderate to severe intensity, which referred to the challenges of dedication and practical care. In the psychological aspects of coping with the situation of care for the child, the indicators were classified as "mild" for emotional and financial burden, personal life implications, satisfaction with the role of caregiver, family support, perception of their effectiveness and control, as well as reactions to the child's needs.

The results show the importance of an early diagnosis to the adaptations of the family routine; they also highlighted the role community support between the primary and secondary caregivers, which was capable of minimizing emotional burden and contributing to the organization of the cooperative routine, complicity, and socioemotional integration of their relationship. This cooperation, as a support network, proved to be important for forming a vital bond in the child's structuring, based on the affection and love that the mother and grandmother showed in the relationship with the child.

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