CASE REPORTS

The impact of Potocki-Lupski syndrome on sibling relationships

Impacto da síndrome Potocki-Lupski na relação entre irmãos

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ABSTRACT

Introduction: Potocki-Lupski syndrome is a condition characterized by cognitive, behavioral, and medical manifestations caused by 17p11.2 duplication encompassing the RAI1 gene. Due to the significance that sibling relations acquire throughout life, changes in the health and functioning of one sibling affect the others.

Case report: A 14-year-old boy**,** healthy sibling of a girl with Potocki-Lupski syndrome, attended the Child and Adolescent Psychiatry consultation due to maternal concerns about the impact of the sister's disease on his well-being. On observation, despite looking relatively well adapted to the sister's condition, the boy showed sadness and shame regarding some of the sister's behaviors, particularly when occurring in public places, and great guilt towards them.

Discussion/conclusion: The impact of the intellectual disability of a child on healthy siblings comprises negative and positive experiences. Therefore, it should be assessed in light of a network of influences and idiosyncrasies characteristic of family relationships.

Keywords: behavior problem; healthy sibling; intellectual disability; Potocki-Lupski syndrome

RESUMO

Introdução: A síndrome de Potocki-Lupski caracteriza-se por manifestações cognitivas, comportamentais e clínicas causadas por duplicações da região 17p11.2 englobando o gene RAI1. Devido ao significado que as relações entre irmãos adquirem ao longo da vida, as mudanças na saúde e funcionamento de um irmão têm impacto nos restantes.

Caso clínico: Um rapaz de 14 anos de idade, irmão saudável de uma rapariga com síndrome de Potocki-Lupski, foi observado em consulta de Pedopsiquiatria a pedido da mãe. Apesar de se apresentar relativamente bem adaptado à condição clínica da irmã, referiu tristeza e vergonha relativamente a alguns dos comportamentos exibidos por ela, principalmente quando estes ocorriam em locais públicos, bem como grande culpabilidade relativamente aos mesmos.

Discussão/conclusão: O impacto da debilidade intelectual de uma criança nos irmãos saudáveis compreende vivências negativas e positivas, devendo por isso ser considerado à luz de uma rede de influências e idiossincrasias próprias das relações familiares.

Palavras-chave: alteração comportamental; debilidade intelectual; irmão saudável; síndrome Potocki-Lupski

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INTRODUCTION

Potocki-Lupski syndrome (PTLS) is a neurodevelopmental disorder characterized by cognitive, behav-ioral, and medical manifestations caused by a microduplication, a 3.7 MB copy number variant, mapping within chromosome 17p.11.2 and encompassing the dosage-sensitive RAI1 gene.[1-4] Despite the extreme variability of presentations, the main clinical manifestations of PLTS comprise speech delay, intellectual disability, and behavioral disturbances.[2-4]

Medical conditions commonly found in this patient population include hypotonia, congenital heart dis-ease, hypoglycemia associated with growth hormone deficiency, mildly dysmorphic facial features, epi-lepsy or electroencephalogram epileptic anomalies without clinical manifestations, brain magnetic reso-nance imaging anomalies, hypermetropia, sensorineural hearing loss, short stature/low weight, gas-troesophageal reflux, chronic constipation, oropharyngeal dysfunction, and obstructive and central sleep apnea. (5,6) Regarding cognitive performance, most individuals show early signs of developmental delay, meeting the criteria for moderate or severe intellectual disability later in life.[4] Behaviorally, attention deficits and signs of hyperactivity, withdrawal, and anxiety may be observed. In addition, some individu-als fulfill the criteria for autism spectrum disorder and exhibit behavioral problems.[7]

The diagnosis of a developmental condition is a relevant event in the family context in ways that are difficult to predict.[8] Although parents' adaptation to a chronically ill or mentally handicapped child has been well studied, considerably less is known about the adaptation of healthy sibling(s).[9]

Sibling relationships typically represent the longest relationships that people have in life and are cen-tral to children's everyday lives. They constitute the first relational experience after parents and are one of the greatest influences for later social relationships and personality development. Sometimes, they represent the only intimate and daily relationship with peers that children keep until preschool age. They have multiple roles across the lifespan, including the provision of companionship, nurturance, support, and learning opportunities. Accordingly, they have a substantial impact on children's development and behavioral and psychosocial adjustment.

When chronic illness appears, it often forces the healthy sibling to assume a new position in the phra-try and to play unknown roles with greater responsibility at a stage of life in which he/she demands indi-vidualized attention. This raises concerns, as active participation in age-appropriate daily routines is es-sential for the healthy development and emotional well-being of healthy siblings.

The research on how having a sibling with intellectual disability affects a child's socio-emotional and behavioral outcomes has mixed findings.[10] The child's experience is often marginalized in the process of planning clinical services for the developmentally challenged sibling.

This study aimed to investigate the real impact of intellectual disability and behavior problems of a sib-ling on a child's experience.

CLINICAL CASE

A 14-year-old boy, brother of a 17-year-old girl diagnosed with PTLS, observed at the Child and Adoles-cent Psychiatry consultation due to maternal concerns about his lack of motivation for daily activities. The mother described a change in the boy's functioning and behavior. The boy was previously a happy, active, joyful, popular, and well-adapted kid and progressively became increasingly isolated, reserved, and ad-dicted to cellphone/computer use and less enthusiastic about previously pleasurable activities, also losing interest in academic tasks. The disinvestment in music lessons that he used to love was what raised the mother's attention.

The boy attended consultation with the mother. In the beginning of the interview, he let the mother expose her concerns without speaking, adopting a listening attitude. The mother's speech evidenced the weight of guilt and how it clouded her perception. She associated the son's change in behavior and func-tioning to a lack of attention and insufficient care during his development due to the sister's increased demands and needs.

After an initial joint interview with the mother, the boy was given some time alone to express his indi-vidual perception of the issues addressed in consultation. When asked to comment on his mother's con-cerns about the time he spent on electronic devices, he promptly disagreed and argued with the pan-demic, stating that it narrowed other possibilities. The boy was also asked to describe in his own words his previous and current personality and to evaluate if there had been any changes in his current behavioral pattern. He initially described himself as insensitive to the well-being of others, with difficulty engaging in empathic feelings towards others, and even though being capable of nourishing positive affection for some individual people, he "did not care if they were not okay". At school, the boy was well adjusted and mentioned an established group of friends with whom he used to play during and after school. At home, he was more passive and avoidant, spending most of the time in his room with little or no time shared with his family. Despite having difficulties in exploring some emotional factors and biographic events, he was able to collaborate in a logic and organized manner.

On clinical assessment, the boy showed no signs of clinical depression or anxiety-related symptoms, nor self-harming thoughts. It seemed that the family daily routines were organized around the sisters' particular and individualized needs, which required adaptation and mobilization of the whole family sys-tem. At first, there was some evident reluctance of the boy in addressing this particular topic (possibly due to a silent sense of guilt underlying his feelings), but later, after normalization of various emotional responses and empathic reflections regarding the difficulties of having a sibling with cognitive disability, he started acknowledging some regret for having to adjust his life to the sister's health condition. Alt-hough he considered himself perfectly attuned with the sister's needs, he also felt the need to have his own space and routines in the household without involving her condition or having less discriminate at-tention

by his parents, managing to build a well-structured and predicable daily routine.

He also disclosed (with evident discomfort) feeling particularly embarrassed with some of the sister's behaviors, particularly during psycho-motor agitation episodes and emotional outbursts, triggered by overcrowded environments, which discouraged him to engage in family plans outside the household.

Overall, the changes observed and valued by the mother seemed to be a normative process of ado-lescence of building one's identity that, due to the family's circumstances and sister's illness, was leading to some distancing from the cohesive family dynamics centered on the sister's needs. Therefore, the boy began developing a somewhat early process of identification and proximity to his peers, assuming a cen-tral and leading role in that context (in contrast to the more secondary and support role played within the family environment). No clinically significant symptomatology was observed that justified a more special-ized intervention. The concern that was amplified by maternal projections of insecurity and guilt was suc-cessfully worked in later sessions.

DISCUSSION

The pioneering research with siblings of children who are mentally handicapped focuses on the potential adverse effects of growing up with an unhealthy child. It assumes that having a child with intellectual dis-ability has a negative impact on the other children in the family.[11] That research concluded that healthy siblings are at risk of psychological effects, such as diminished self-concept, poor psychological adjust-ment, increased psychopathology and emotional disturbances manifested in resentment, aggressive behavior, confused thinking, guilt, embarrassment, and feelings of neglect.[11-13] More recent research reported that siblings of people with intellectual disability tend to be well-adjusted and generally indistin-guishable from their peers.[11-14]

Research in the area tends to follow one of two theoretical frameworks. The first describes a deficit model that assumes that the presence of a sibling with disability is a risk factor for the healthy adjustment of the sibling without disability. The second suggests that having a sibling with disability makes a positive contribution to the psychosocial development of the healthy sibling, through personal growth imputed by exposure to adverse circumstances.[10]

The literature suggests that having a disabled sibling is not necessarily a stressor and that the ad-aptation of the healthy sibling to this reality depends on his/her personal resources and life history and to the meaning that the disability assumes and is shared within the family system, which leads to greater variability in the child's adjustment.[12,13]

Given the multiple variables that contribute to the variability of responses of healthy siblings (re-sources, child and family characteristics, family history), results should not be generalized, but instead understood within the network of influences and idiosyncrasies of family relationships. The quality of family relationships, communication, existence of a support and care network, individual characteristics, coping strategies, and disability features are factors that should be considered when evaluating the im-pact on healthy siblings.[15]

CONCLUSIONS

The way in which healthy siblings cope with the disability of another sibling is unique. The disparate findings reported in studies reflect the great versatility of reactions and adaptations to the disease pro-cess, which are not necessarily psychopathological.

AUTHORSHIP

Marta P. Antunes - Conceptualization; Writing – original draft; Writing – review & editing

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