

CASE REPORT

Addressing Menstrual Management in a Child with Rare Syndrome: A Caregiver's Perspective

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ABSTRACT

In Malaysia, rare genetic disorders affect fewer than 1 in 4,000 individuals, including complex chromosomal 10 disorder, which causes severe developmental delays and cognitive impairments. This case report explores the challenges faced by the mother of "Q", an 11-year-old Malay girl with this condition. Q is dependent on a wheelchair for mobility, nonverbal, and presents with profound cognitive impairments. She recently began menstruating, leading to increased energy levels and irregular sleep patterns, which have added to her mother's stress. Scales reported by the mother indicated the presence of behavioural difficulties in the child and elevated levels of stress within the parent-child relationship. Counselling focused on stress management, problem-solving, and establishing routines, alongside collaborative caregiving and peer support for both Q and her mother. Following the counselling sessions, Q's sleep patterns remained irregular; however, her mother's stress levels significantly decreased. Q has now resumed physiotherapy and speech therapy, with further assessments planned to evaluate the effectiveness of these interventions. This case highlights the urgent need for caregiver support, accessible daycare services, and systemic reforms to improve outcomes for parents and children with disabilities in Malaysia.

Malaysian Journal of Medicine and Health Sciences (2026) 22(SUPP2):173-175. doi:10.47836/mjmhs.22.s2.27

Keywords: Rare genetic disorders, Parenting stress, Menstrual management, Caregiver support, Supportive counselling

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INTRODUCTION

Rare disorders are diseases that present significant challenges in both diagnosis and treatment, affecting between 0.65% and 1% of the population, as reported by the World Health Organization (WHO) and supported by Malaysian data (1). In Malaysia, there is currently no standard definition for rare diseases due to limited prevalence data; however, their recognition and management are increasingly acknowledged as important health priorities (1). Rare genetic disorders, which form a subgroup of rare diseases, predominantly manifest during childhood, with estimates suggesting that a large proportion affect children (2). Approximately 70–80% of rare diseases have a genetic origin, and many involve the nervous system, resulting in developmental, cognitive, and motor impairments (2). These impairments create substantial long-term challenges for both affected children and their caregivers, with additional care de-

mands emerging during developmental transitions such as puberty (1, 3, 5).

Managing puberty in children with such conditions adds further complexity, especially for females who begin menstruating. Developmental, cognitive, and motor limitations can make independent menstrual hygiene management difficult, while communication impairments – common in many rare genetic disorders – can prevent the child from expressing discomfort or specific needs (2-3, 5). This often results in increased physical demands, emotional strain, and financial pressures on caregivers - who may already face high stress levels due to the overall caregiving burden (4-5). Despite these challenges, little research in Malaysia has examined the lived experiences of caregivers navigating menstruation in children with disabilities. This case report addresses this gap by highlighting these specific burdens and suggesting interventions to support caregivers.

CASE REPORT

Q is an 11-year-old Malay girl diagnosed with a rare complex chromosomal 10 disorder, a genetic condition

caused by structural abnormalities in chromosome 10 that can lead to significant developmental, cognitive, and motor impairments (1–2). Chromosome 10 is among the human chromosomes that harbour genes crucial for cell growth, nervous system development, and metabolic regulation; disruptions in such genetic material can contribute to a wide spectrum of health issues (2). Such chromosomal abnormalities often cause lifelong functional limitations, requiring high levels of care and ongoing multidisciplinary support to meet daily living needs (1-2, 5). Q is dependent on a wheelchair for mobility, nonverbal, and presents with profound cognitive impairments, all of which substantially increase her dependency on caregivers for both basic and complex activities of daily living.

Q is the youngest child of a 47-year-old housewife and a 58-year-old businessman, to whom she is closely attached. Q's mother - Madam Y, is the second wife to Q's father, whom she married after her first husband passed away in 1999 due to a motor vehicle accident (MVA). Q has a 19-year-old brother who is also diagnosed with a rare genetic disorder called cri-du-chat, which caused him to be immobile and nonverbal. Q is currently staying with her mother and brother, while her older half-siblings from Madam Y's first marriage are all living separately (see Fig. 1 for family genogram). Having two families to take care of, Q's father takes turn to be with Q and her family. Despite these circumstances, Madam Y maintains a good relationship with her husband, children from the first marriage, and stepchildren, who are affectionate towards both Q and her brother.

Following the onset of puberty, Madam Y reported that Q showed increased energy, irregular sleep, and heightened distress, alongside longstanding dependence for daily self-care and immobility. Q's nonverbal status complicated the identification of menstrual-related needs and hygiene management, requiring diaper use and frequent lifting by her mother. This physical demand has contributed to Madam Y's deteriorating health, including a slipped disc, while frequent diaper changes pose a significant financial burden given limited income and reliance on assistance.

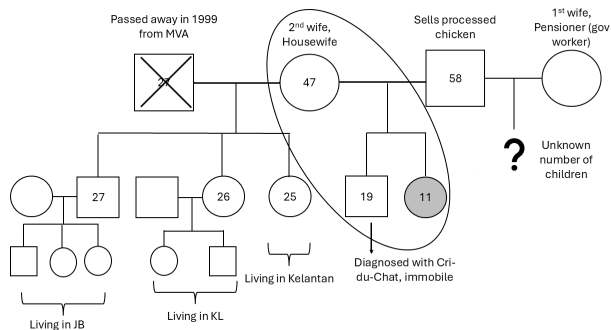


Figure 1: Q's Family Genogram. Q is highlighted in grey and has an older brother also diagnosed with a rare genetic disorder, cri-du-chat.

Menstruation-related behavioural changes further increased caregiving stress and emotional exhaustion, particularly Q's distress when her father is unavailable. Assessment using the Strengths and Difficulties Questionnaire (SDQ) and Parenting Stress Index (PSI), conducted with informed consent, indicated borderline conduct issues, elevated hyperactivity, and low prosocial behaviour on the SDQ, interpreted cautiously due to Q's disability. The PSI showed high stress in the Parent-Child Dysfunctional Interaction domain, reflecting challenges in understanding Q's needs and managing caregiving demands.

This case can be visualised using the Double ABCX model – a family stress theory that explains how families respond to and adapt to crises over time (4). In this model, A represents the initial stressor, B the family's existing resources, and C the family's perception of the stressor, which together influence the level of crisis (X) experienced. The “double” aspect of the model recognises that, after the initial crisis, families often face additional stressors (aA), acquire new resources (bB), and develop evolving perceptions (cC) that affect their long-term adaptation – either maladaptation (poor adjustment) or bonadaptation (positive adjustment) (4).

Using this framework, caregiver stress in Madam Y's situation was examined (Fig. 2). The initial stressor (A) was caring for two children with genetic disorders. Her existing resources (B) included receiving financial aid from the government, while her perception of the stressor (C) was that “this is a test from God”, reflecting acceptance of her circumstances. The onset of puberty for Q acted as the crisis (X), leading to a build-up of demands (aA) such as behavioural changes in Q, both parents' advancing age, and Madam Y's own health problems. Additional resources (bB) included family support and assistance from NGOs. Over time, Madam Y's perception (cC) shifted towards more negative thoughts, such as “This is too much; I can't handle it anymore”. These factors resulted in a mixed adaptation outcome (xX): she had trouble regulating her emotions (maladaptation) yet also engaged in therapy and counselling to seek support (bonadaptation).

A structured management plan was introduced to ad-

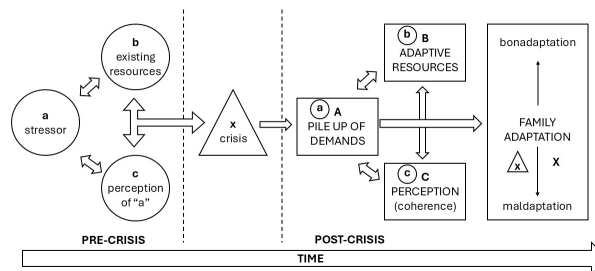


Figure 2: The ABCX Model, adapted from (4). This model encapsulates the adaptation to stress and crisis by Madam Y

dress maternal stress and strengthen caregiving strategies, as psychosocial support and targeted interventions can reduce caregiver burden in families of children with developmental disabilities (5). Supportive counselling encouraged emotional expression and coping, consistent with family adaptation models that emphasise problem-solving and emotional support (4). Relaxation techniques and self-care practices were incorporated to mitigate the physical and emotional strain associated with long-term caregiving (5). Establishing predictable daily routines helped improve caregiver organisation and sense of control, supporting more consistent care delivery (4). Collaborative caregiving was promoted by increasing the father's involvement, and exploring day-care services aimed to provide respite opportunities, both of which have been shown to alleviate caregiver stress (5). Finally, developmental goal setting encouraged Q's active participation in speech and physiotherapy, aligning with the recognised need for multidisciplinary rehabilitation to address functional challenges in children with complex genetic disorders (1-2).

After four months of fortnightly sessions, post-intervention reports showed reduced maternal stress and improved caregiving strategies. Madam Y demonstrated better emotional expression and confidence, particularly in managing Q's menstrual hygiene and behavioural changes. Although daily challenges became more manageable, Q's irregular sleep due to restlessness persisted. Daycare was considered to channel Q's energy but deferred due to reduced government financial aid, which would increase caregiving costs. While caregiver stress improved, ongoing support remains important to sustain Madam Y's resilience.

DISCUSSION

Developmental, cognitive, and motor impairments complicate menstrual hygiene management in girls with disabilities, increasing physical, emotional, and financial strain on caregivers, most often mothers (3-5). This case reflects existing literature and underscores the need for stronger systemic support in Malaysia, where research on menstrual hygiene challenges in rare diseases remains limited. Using the Double ABCX model, puberty functioned as a crisis that intensified caregiving demands, while family support and multi-agency interventions facilitated adaptive coping, although persistent stressors such as financial constraints and limited respite care remain.

This case highlights a clear gap in Malaysian literature and services, as specialised caregiver support and structured respite care are scarce. Although interventions reduced maternal stress, ongoing psychosomatic symptoms indicate the need for additional measures, including behavioural management plans, affordable daycare services, and policy reforms to improve equitable access to support (1,5). Future research and advocacy should focus on long-term, context-specific interventions for

nonverbal children with rare genetic disorders, particularly in relation to menstrual hygiene management and caregiver well-being.

CONCLUSION

This case highlights the significant challenges faced by caregivers managing puberty in children with rare genetic disorders, emphasising the physical, emotional, and financial burdens involved. While structured interventions were effective in reducing maternal stress and improved caregiving strategies, systemic gaps in Malaysia continue to hinder comprehensive support. Policy reforms should include: (i) expanding access to affordable respite and daycare services, (ii) providing financial subsidies for menstrual hygiene products, and (iii) developing training programmes for caregivers on menstrual and behavioural management.

ACKNOWLEDGEMENT

We sincerely thank Madam Y for her willingness to share her experiences, which provided invaluable insights into the challenges of caregiving for a child with a rare genetic disorder. We also extend our gratitude to the healthcare professionals and therapists involved in supporting Q's development and well-being. Lastly, we acknowledge the guidance and feedback from our colleagues, which have strengthened the development of this manuscript.

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