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CASE STUDY: PARENTAL ROLE IN THE HOLISTIC CARE OF A CHILD WITH DOWN SYNDROME

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Abstract

Background: Down syndrome is a genetic disorder caused by trisomy 21 and is associated with developmental delays in physical, cognitive, and adaptive functioning. Family roles, particularly parental involvement, are critical to ensuring optimal development and quality of life for children with this condition. **Aims and scope of paper**: This study aims to explore the holistic role of parents in caring for a child with Down syndrome from pregnancy to school age in a rural Indonesian setting. **Methods**: A qualitative case study was conducted involving one family in Blora, Central Java. Data were collected through observation, interviews, and document reviews from 2017 to 2025. **Result**: The study found that despite limited access to screening, diagnostic resources, and professional therapy, parents played a central role in health care, stimulation, and education of the child. Parents demonstrated resilience, sought information, initiated home-based stimulation, and integrated the child into early education. Challenges included lack of prenatal knowledge, financial constraints, and social stigma. **Conclusion**: Parental involvement is essential in holistic care for children with Down syndrome. Strengthening family-based support systems and improving access to early intervention are critical in optimizing developmental outcomes.

Keywords: Down syndrome, Holistic care, Parental role

INTRODUCTION

Down syndrome (trisomy 21) is a genetic disorder caused by the presence of an extra whole or partial chromosome 21. This condition is characterized by mild to moderate intellectual disability, developmental delays, and distinct facial features such as a flat nasal bridge, upward slanting eye folds, and a protruding tongue (Bull, 2020). The incidence of Down syndrome increases with advancing maternal age, with prevalence rates ranging from 1 in 319 to 1 in 1,000 live births (Mai et al, 2019). Although the frequency of fetuses with Down syndrome is relatively high at conception, approximately 50–75% are miscarried before reaching full term (Morris et al, 2002). Other autosomal trisomies, such as trisomy 13 and 18, may occur more frequently, but the postnatal survival rate of individuals with Down syndrome is significantly higher compared to other autosomal trisomies (Weijerman & de Winter, 2010).

To date, the exact cause of Down syndrome is not fully understood, although several risk factors have been identified. Trisomy 21 can occur due to errors in chromosome separation (nondisjunction) during meiosis or in the early mitotic divisions of the zygote. This process most commonly occurs in oocytes that are arrested in prophase of meiosis I until ovulation. Nondisjunction may be triggered by factors such as viral infections, radiation exposure, aging of the oocyte, and maternal age over 35 years (Hassold & Hunt, 2001). If an ovum carries two copies of chromosome 21 and is fertilized by a normal sperm, the resulting zygote will have trisomy 21. In addition to this, other forms such as translocation and mosaicism can also occur, although they are less common.

Prenatal diagnosis of Down syndrome can be performed using several methods. Ultrasound examination between 14 and 24 weeks of gestation is used to detect soft markers such as nuchal fold thickening, a small or absent nasal bone, and enlarged cerebral ventricles (Nicolaides et al, 2005). Nuchal translucency (NT) can also be measured via ultrasound between 11 and 14 weeks of gestation, and increased NT thickness may indicate a risk of Down syndrome or other chromosomal abnormalities (Kagan et al, 2008). Invasive procedures such as amniocentesis and chorionic villus sampling (CVS) are also used to confirm the diagnosis, although these procedures carry a miscarriage risk of approximately 0.5% to 1% (ACOG, 2020).

Children with Down syndrome face challenges in physical, cognitive, communicative, and adaptive behavioral development. Therefore, the role of the family particularly the parents is crucial in supporting the child’s overall growth. Parents are not only responsible for meeting the child’s basic needs but also serve as facilitators of therapy, educational companions, and managers of the child’s emotional and social well being (Van Gameren-Oosterom et al, 2011). Holistic care that incorporates medical, educational, social, and psychological approaches can significantly improve the quality of life for children with Down syndrome (Fidler et al, 2012). However, studies exploring the experiences of families in supporting children with Down syndrome from pregnancy through to education remain relatively limited, particularly within the cultural context and healthcare system of Indonesia.

Therefore, this study aims to describe the journey of a family in supporting a child with Down syndrome from pregnancy, birth, and daily care through to education. It is expected to provide an in-depth understanding of the dynamics of the family’s role in the holistic care of a child with Down syndrome and to contribute to the development of policies and services that are more responsive to the needs of both the child and the family.

METHODS

This study employed a qualitative approach using a case study design. The aim was to gain an in-depth understanding of the parental role in caring for a child with Down syndrome, from pregnancy through to primary school age. The subject of the study was a single family (parents and child) residing in Blora Regency, Central Java, who have a child diagnosed with Down syndrome. The child is currently seven years old, and data were collected regarding the family's experiences from pregnancy to the present. Additional data were obtained from medical records, therapy history, and the child's educational development. Observations were conducted periodically from July 2017 to May 2025.

Result

Subject Identity

To gain a comprehensive understanding of the case background, the following table presents the identity information of the subject who was the focus of observation in this study.

Table: 1 Subject Identity

Variable	Description
Name	MHS
Gender	Male
Date of Birth	July 25, 2017
Mother's Name	SS
Mother's Current Age	41 years
Occupation	Teacher
Marital Status	Married

Antenatal History and Physical Health

The initial antenatal care began at 7 weeks of gestation, with routine assessments such as HCG, hemoglobin, and standard ultrasound. Although the pregnancy appeared normal, by 36 weeks the fundal height measured only 24 cm, suggesting suboptimal intrauterine growth. No specific screening for chromosomal anomalies was conducted, due in part to limited awareness and the absence of comprehensive prenatal counseling. Notably, the mother had been exposed to agricultural chemicals during early gestation while assisting her husband with pesticide application. After birth, the child weighed 2200 grams and measured 47 cm at 36–37 weeks of gestation. There were no immediate concerns communicated by health professionals. However, by five months, the child exhibited significant developmental delays. A diagnosis of Down syndrome was made based on hallmark phenotypic features, including hypotonia, flat facial profile, and widely spaced eyes. A congenital oropharyngeal adhesion, confirmed by a pediatrician, further complicated feeding and nutrition. The child struggled with complementary feeding, had persistent failure to thrive, and showed delays in gross motor milestones such as rolling over and holding up his head. In response, parents supported by local health services and community nutrition programs ensured consistent nutritional monitoring and sought medical guidance. By age seven, the child demonstrates some autonomy, including the ability to play, sing, and speak, although with articulation difficulties and challenges in emotional self-regulation.

Emotional Adaptation and Family Acceptance

The initial emotional response from the parents following diagnosis was marked by shock, grief, and guilt reactions frequently seen among families of children with disabilities. Despite limited resources, the family gradually transitioned from denial to acceptance. They sought medical and developmental information independently and maintained a proactive stance in caregiving. The emotional burden was further complicated by a lack of structured psychological support, but over time, the parents developed resilience. They embraced their child's condition with compassion, offering consistent emotional support while maintaining hope for his functional progress. Their acceptance not only enhanced their caregiving capacity but also created a stable emotional environment for the child’s development.

Social Engagement and Community Inclusion

Despite early emotional hurdles and societal prejudice, the parents have made a deliberate effort to integrate the child into the social fabric of the community. They frequently involve him in local early childhood programs and communal activities to build social confidence and reduce isolation. While stigma remains a challenge, especially regarding public perceptions of children with disabilities, the family has cultivated a supportive micro-environment through extended family, neighbors, and school staff. This social exposure has fostered improvements in the child’s interpersonal skills and responsiveness to unfamiliar environments.

Education and Stimulation

Formal developmental therapy was not accessible due to financial limitations; however, the parents, especially the mother, consistently engaged the child in informal learning activities. By enrolling him in kindergarten programs, they emphasized structured routines and encouraged social and cognitive stimulation. These early learning environments were used strategically to nurture basic competencies in communication, motor skills, and independence. The child has shown progress in adaptive behaviors despite professional therapy not being available. Plans are underway to enroll him in a special education primary school, which the parents hope will offer more individualized support and accommodate his specific learning needs

Summary of Challenges

This case illustrates the multifaceted challenges faced by families caring for a child with Down syndrome, including delayed diagnosis, limited access to therapy, economic constraints, and social stigma. However, it also highlights the vital, multi-dimensional role parents play not only in fulfilling physical and health needs, but also in offering emotional support, promoting social engagement, and facilitating educational growth. Through resilience and continuous adaptation, the family has developed a functional care system that addresses the child’s holistic development, despite minimal external resources

DISCUSSION

This case illustrates how limited information during pregnancy greatly contributed to delayed detection and early intervention for Down syndrome. Although antenatal care began at seven weeks’ gestation, no genetic screening such as a nuchal-translucency ultrasound, triple test, or quadruple test, all of which should be part of standard protocols in high-risk pregnancies was performed. The shortfall stemmed not only from health-service constraints but also from the family’s low reproductive-health literacy. Consequently, the parents did not take an active role in seeking information or making decisions about prenatal screening. Yet previous studies show that parental engagement during the antenatal period including understanding test results and participating in healthcare decision-making is a critical foundation for holistic, early care (Susilowati, Realita, & ’Aisy, 2023)

The suspicion of Down syndrome emerged when the child reached five months of age. Prior to that, the family had not received any information from medical personnel about the possibility of a genetic disorder at birth. Once physical features and developmental delays became noticeable, a diagnosis was made at the community health center (Puskesmas) based on typical clinical signs. Initially, the parents experienced shock, sadness, and profound guilt—emotional reactions commonly found among families of children with special needs. However, over time, these emotions transformed into an adaptive response. The parents began actively seeking information, consulting a pediatric specialist, and following medical guidance, despite financial limitations that prevented a karyotype test from being performed. This highlights the crucial role of parents as the primary agents in accepting a diagnosis and initiating appropriate care for their child (Fidler et al, 2012).

Health issues that emerged during the early stages of growth, particularly during the introduction of complementary feeding (MPASI), further intensified the challenges faced by the family. The child experienced difficulty swallowing due to a throat adhesion, which negatively affected nutritional status and contributed to delayed growth. Significant developmental delays were also observed, such as the inability to roll over or support the head at six months of age. In this situation, the parents played a vital role in daily caregiving. They not only worked to meet the child’s nutritional needs but also consistently sought information and solutions through active communication with healthcare professionals. The parents’ role as daily caregivers and as intermediaries between the child and the healthcare system proved essential for sustaining the child’s care in a setting with limited resources (Van Gameren-Oosterom et al, 2011).

Financial limitations became one of the main obstacles for the family in accessing professional developmental therapy services. However, these constraints did not hinder the parents’ efforts to provide alternative stimulation within the home and community environment. The mother actively involved the child in early childhood education and kindergarten activities as a form of non-formal intervention. The child was introduced to various activities designed to stimulate fine motor skills, communication, and independence. Although not directly guided by professional therapists, the continuity of stimulation provided by the parents led to noticeable functional progress in the child. The child began to speak, sing, and interact, although emotional regulation remained a challenge. This highlights the critical role of parents as facilitators of learning and trainers of independence in the developmental journey of a child with Down syndrome within a family-based intervention context (Rohmadheny, 2016)

The family also faced psychosocial challenges, particularly the stigma and social pressure from their surrounding environment. The parents experienced emotional distress due to the negative perceptions directed toward their child with special needs. Despite this, they chose to continue involving the child in social and educational activities, including planning for enrollment in a Special Needs School. This decision reflects the family's psychological resilience and empowerment in dealing with external pressures. The role of parents as emotional protectors and supporters is crucial in fostering the child’s self-confidence and minimizing the adverse impact of social stigma. The choices made by the family across health, education, and social domains demonstrate that parents serve not only as technical caregivers but also as active agents shaping the holistic development of a child with Down syndrome (Rosantia, et al., 2025).

This case as a whole illustrates that even in the midst of limited information and resources, the family remains the central pillar in the care of a child with Down syndrome. Parents play a crucial role not only in overcoming medical and developmental challenges but also in establishing appropriate social and educational support systems for their child. These findings emphasize the importance of a family-centered approach in every intervention strategy and public policy aimed at children with special needs. Empowering parents with accurate information, adequate access to services, and emotional support will be key to improving the quality of life for children with conditions such as Down syndrome within the community.

CONCLUSION AND SUGGESTION

The findings from this case observation demonstrate that parental involvement is a key determinant in the care and developmental trajectory of children with Down syndrome, particularly in contexts where access to information, economic resources, and professional therapy services is limited. Although early detection was not achieved during pregnancy and the diagnosis was confirmed relatively late, the parents successfully adapted to their child's condition, both emotionally and functionally. Their active role was reflected in continuous efforts to seek information, ensure nutritional fulfillment, participate in basic health services, and initiate self-directed stimulation through non-formal educational activities.

This level of involvement suggests that holistic care should not be limited to medical interventions but must also encompass psychosocial, educational, and emotional dimensions. These elements are significantly shaped by the parents' resilience and awareness of their child's needs.

We must support families of children with Down syndrome more effectively, it is crucial to implement policies and programs that empower parents with access to accurate information, affordable therapy services, community-based support, and emotional guidance. Building a responsive support system that integrates healthcare, education, and social services will ensure better developmental outcomes and an improved quality of life for both the child and the family.

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