Medical Research Archives

ESMED
European Society of Medicine

OPEN ACCESS

Published: May 31, 2023

Citation: Peters VJT and de Winter JP, 2023. The Complexities of Down Syndrome Healthcare: Medical Comorbidities, and Care Models, Medical Research Archives, [online] 11(5).
DOI: https://doi.org/10.18103/mra.v11i5.3956

ISSN: 2375-1924

RESEARCH ARTICLE

The Complexities of Down Syndrome Healthcare: Medical Comorbidities, and Care Models

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Main Text

Down syndrome (DS), often caused by trisomy 21, is the most common form of intellectual disability among newborn infants worldwide[1]. Differences in annual live births have been observed per continent: around 17,000 annual live births of children with DS have been estimated in Europe[2], around 5,100 annual live births of children with DS have been estimated in the US[3], and around 300 annual live births of children with DS have been estimated in Australia and New Zealand[4]. The presence of prenatal screening and elective terminations has negatively influenced the live birth rates of children with DS.

The availability and accessibility of prenatal testing (like non-invasive prenatal testing) and genetic counselling certainly have a current and future impact on the number of live births with DS[5]. In addition, there is a risk of the routinization of prenatal screening, where parents are no longer facilitated to make informed decisions based on their own moral and practical considerations, but family members, relatives, and friends alike, implicitly or explicitly expect that parents will choose prenatal screening, diagnostic testing, and, perhaps the termination of pregnancy[6,7]. Such potential societal pressure may increase the effect of the availability of prenatal screening on the live birth rate of children with DS. Other factors - such as religious beliefs, economics, the complexity of society, changing maternal ages, cultural beliefs, and social norms - likely play additional roles. The anticipated quality of life for a person with DS might also be an essential consideration in the decision-making for some expectant parents[6].

About live birth rates, the life expectancy of persons with DS has significantly increased in the last decades due to improvements in medical care, such as improvements in cardiac surgery, prevention of childhood infections, and broader access to standard care. This results in an expanding cohort of persons with DS who need medical care addressing their unique profile[8]. Given these developments, we aimed to provide a brief overview of progress made in the last few years regarding medical comorbidities and care models in the field of DS healthcare.

Persons with DS are known to have an intellectual disability and a variety of malformations like congenital heart defects, small ears, small mouths, and other physical findings[1], along with medical conditions like hip dislocation[9] and leukaemia[10]. Many medical conditions are more common in individuals with DS than the general population and affect health, development, and daily functioning. Therefore, secondary screening for comorbidity is an essential part of the care of persons with DS[11]. DS is associated with medical comorbidities and disorders that differ from population rates and impact organ systems throughout the body. As a result, persons with DS have an increased risk for conditions like obstructive sleep apnea[12], obesity[1], hearing problems[13], vision problems[14], congenital heart diseases[15], autism[16], regression disorder[17], and Alzheimer's disease[18,19], among others[1,20]. The healthcare professionals most frequently involved in treating these comorbidities are paediatricians, cardiologists, ophthalmologists, ENT physicians, dieticians, speech therapists, orthopaedic surgeons, physiotherapists, (paediatric) cardiologist, psychologist, and education generalist[11].

The best follow-up for persons with DS involves regular medical check-ups, developmental assessments, and social support. Medical check-ups should be scheduled every year or as recommended national guidelines to monitor any health concerns or conditions that may develop[10,21]. Developmental assessments help identify learning difficulties and other developmental delays, allowing for early intervention and tailored support. Social support is essential, including education and training for family members and caregivers on how to provide persons with DS with a supportive environment where they can thrive. This deserves attention from the moment the information about a suspected DS diagnosis, either before or after birth, is communicated with family members and caregivers[22]. Additionally, access to therapy and programs that promote social interaction, life skills, and independent living can help promote a high quality of life for persons with DS.

With declining birth rates yet a higher prevalence of childhood and adult survivors with DS, it is important to identify the optimal model to deliver care for persons with DS in all life stages. Finding the optimal care model is especially relevant because although each separate clinical problem is often well known, the personal tailoring of the screening, prevention, and treatment in an individual with DS makes the organization and delivery of DS healthcare complex. Around the globe, there is significant variability in content, organization, provision, and access to care for persons with DS. They follow different models, serve different populations by age or location, and are organized by different medical specialties[23]. This diversity hampers the implementation of personalized care and leads to unmet care needs all around the globe. For example, in the US, specialty DS clinics exist where some see only the paediatric population, whereas others provide care

restricted to adults[24]. In Europe, multidisciplinary care for children with DS is organized in various forms: in the Netherlands, paediatric outpatient clinics organize multidisciplinary team appointments[20,25], and in Israel, multidisciplinary centers provide holistic care to persons with DS[26]. In Asia, integrated care for children with DS is realized by organizing multidisciplinary care with protocol-driven surveillance in Taiwan[27], and clinical guideline management by physicians in Singapore[28]. The importance of specialty clinics is also recognized in other countries like Oman[29].

Although various care models have been reported in the literature, there is no evidence that one model is more effective than others. Therefore, the variety in care models should make us pause and re-consider who should care for children with DS, and how that care should be delivered and organized. First steps are made in this regard by, for example, using a modular decomposition approach to provide insight into the underlying organizational structure of healthcare provision, which provides opportunities to offer persons with DS individualized healthcare based on their needs and requirements[25,30,31]. These first steps could eventually influence the quality of healthcare for persons with DS[32] and, consequently, the quality of life in persons with DS[33].

There are several areas of investigation that could be explored to understand the long-term outcomes of persons with DS. Some potential avenues of research might include 1) Medical follow-up: persons with DS are at increased risk for a variety of medical conditions, including heart defects, thyroid disease, and Alzheimer's disease. Studying long-term outcomes could involve tracking the incidence and progression of these conditions, as well as evaluating the effectiveness of different interventions or even medical treatments, 2) Intellectual development: long-term studies could explore the factors that contribute to intellectual development over time, as well as strategies for maximizing potential in different areas of functioning, and 3) Occupational and social outcomes: persons with DS often face employment discrimination and social isolation, despite the fact that many are capable of living independently and contributing to society.

Although DS is an incredibly complex, multi-system condition, the affected persons, their family members and caregivers, and involved healthcare professionals in both paediatric and adult care provision have an opportunity to collaborate and provide new insight into complex care delivery in the 21st century. One opportunity is actively involving persons with DS in health research, also known as inclusive health research[34]. However, historically persons with DS have been excluded from (clinical) research and family members and/or caregivers have served as a proxy[35]. This has proven helpful given that family members and caregivers often have more experience and expertise about their offspring, since they are in the lead of care content and organization. Hence, they face the life-long challenge of negotiating health and social service systems for these persons with DS who depend on others to understand and explain their needs. Although family members and caregivers are often used as proxies in paediatric care, differences between children and their proxies have been reported. Therefore, it might be worth exploring whether persons with DS, in addition to caregivers, could also serve as experts-by-experience in inclusive health research to capture the full potential of these persons.

Another opportunity arises concerning the transition from paediatric to adult care. This transition has many challenges, crossing all dimensions of life. Persons with DS and caregivers need to find adult healthcare providers, ensure insurance coverage, and, where possible, take ownership of their health maintenance. In parallel, transition in various other spheres, such as educational, vocational, financial, social, guardianship, and legal responsibilities[23]. Despite the attributed importance of this transition, only a few studies have been conducted on the transition of care for persons with DS[36], and no consensus has been reached on the organization of care transition for persons with DS.

Last, an increasing number of persons, besides persons with DS, live with complex care needs resulting from incredibly complex, multi-system conditions[37]. Due to lifelong care needs on multiple life domains, these persons present a challenging task for healthcare professionals and care systems to provide optimal personalized care, taking both the characteristics of the genetic disorder and the individual into account. The progress in the field of DS can be extrapolated to offer directions for dealing with complex care needs for persons with other rare genetic disorders like 22q11 deletion syndrome[38], Williams syndrome[39], and Phelan-McDermid syndrome[40].

The manifestations of DS are complex, warranting (ideally) expert and multidisciplinary care in all life stages. Due to lifelong care needs, persons with DS present a challenging task for health care providers and care systems all over the world to provide optimal care, taking both the characteristics of the genetic disorder as well as the individual needs into account. In the present paper,

we present an overview of progress made in the last few years regarding medical comorbidities and care models in the field of DS healthcare. We hope this overview is inspiring and leads to avenues for future research.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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