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Transition from Paediatric to Adult Services for Patients with 22q11.2 Deletion Syndrome

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Abstract

22q11.2 Deletion Syndrome (22q11.2DS) is among the commonest microdeletion syndromes. Characterised by diverse clinical symptoms of different severity, it may remain unrecognised until later in life. The syndrome caries high risk for neurodevelopmental and psychiatric comorbidities. It is expected that people with 22q11.2DS will require care throughout their lifespan and will transition between services. Planning for comprehensive care is difficult due to the high variability of clinical presentations. Time of transition is particularly vulnerable. Additional focus on the transition will improve outcomes for the individuals with 22q11.2DS and their families, and will have a long-term positive impact on utilisation of services.

Introduction to 22q11.2 Deletion Syndrome

22q11.2 Deletion Syndrome (22q11.2DS), caused by a deletion of approximately 3-megabase on the long arm of chromosome 22, is one of the most common chromosomal microdeletion syndromes, with prevalence range of 1:2000 to 1:4000 life births[1]. In Ireland this equates to approximately 15 to 30 babies with 22q11.2DS being born per year. Whilst there is no register of diagnosed cases, the number of recognised cases is considered to be lower. It is expected that even fewer of them have accessed services. This is in part due to the variation in clinical manifestation ranging from mild facial dysmorphic features to severe multi-organ congenital abnormalities. Individuals with 22q11.2 DS frequently present with congenital heart defects, immunodeficiency, palatal abnormalities, hypoparathyroidism/hypocalcaemia, renal and gastrointestinal abnormalities[1]. Additionally, individuals have a complex neurodevelopmental profile with overall intelligence typically within the borderline (IQ, 70-84) or mild (IQ 55-69) intellectual disability range[2]. Developmental delay in the areas of gross and fine motor skills,

speech and language, and social skills are commonly seen[3]. Individuals with 22q11.2DS carry a very high risk of both neurodevelopmental and psychiatry disorders. Rates of autism and ADHD are present in approximately 30-40% of the younger 22q11.2DS population. Anxiety and mood disorders across all age groups occur in about 40% and 20% respectively. The risk of developing psychosis is 25 times higher than the general population[4]. 22q11.2DS is also associated with some late onset conditions such as Parkinson's disease[5]. The complex and varying nature of the clinical manifestations of 22q11.2DS brings up significant challenges for individuals with 22q11.2DS and their carers/families. It is not surprising that carer perceived burden is high. Parents report feeling frustrated, overwhelmed and strained by the 22q11.2DS, with little or no time for themselves and other family members[6,7]. Chronic multisystemic conditions, the presence of intellectual disability or learning difficulties, together with possible behavioural difficulties and psychiatric illness mean that input from many professionals and services would be required throughout the lifespan. Starting from early childhood to adolescence, and adulthood patients with 22q11.2DS need ongoing comprehensive care.

Transitional care

Youth with special medical and psychosocial needs are particularly vulnerable at times of transition when their care is moved from one healthcare setting to another, for instance, neonatal to paediatric, child to adult and on into services for older individuals. Research shows that patients often experience gap in care at the time of transition between paediatric and adult services[8]. There is also a conceptual difference between paediatric and adult care. Paediatric services by definition are focused on the young child's development and functioning. Decisions are made with parental/ carer's involvement. By contrast, adult medicine is designed towards patient autonomy, relying on the person themselves to outline, present and manage their own care[9]. Adult physicians feel less confident and willing to manage complex childhood onset conditions, and work with patients with intellectual disability or autism[10]. Parents and caregivers of adults with 22q11.2DS remain highly involved in the care of the grown-up individual, reporting feelings of being overburdened by their roles of advocate for services. They report significant dissatisfaction with the transition between paediatric to adult services both medical and social[7]. Adult patients also recognise their own limitations and wish for parental involvement in their health needs, along with a desire for adult physicians to be better informed. Adults with 22q11.2DS and caregivers find themselves in a position to be educators in various medical and social settings [6,7]. The diverse clinical presentation of 22q11.2DS makes it less recognisable to clinicians and might also account for the lack of awareness.

The period of transition is important not only in the context of healthcare, but also psychosocially. Moving through adolescence to adulthood creates different roles and expectations for the person as an individual, as part of a family, socially and occupationally. At an individual level, the most important factors for independent living are intellectual abilities and overall functioning, including communication and social skills[11]. In the context of 22q11.2DS, neuro-developmental, psychiatric and or behavioural issues are most frequently reported as a major challenge during transition[7]. The family plays a central role in supporting the process of transition and in a context of limited

resources it is an important contributor to success[12]. Finding balance between paternalism and encouraging independence may lead to personal conflicts for caregivers. A respectful and appropriate fear for parents is their own ageing and the need for future planning[7].

In addition to transitional health care needs, having access to educational and social support for employment/housing becomes an essential stepping stone towards successful transition[11]. Educational and social support is widely recognised to be inadequate[13]. The presence of intellectual disability and psychotic illness appear to be the factors with the greatest impact on functional outcomes and in need of the greatest transitional support[14]. Adaptive functioning and the ability to manage day to day tasks are found to be better predictors of employability than cognitive abilities and should be the focus of additional support during this time[15].

One of the challenges planning comprehensive care for people with 22q11.2DS is the diverse nature of the clinical presentation, cognitive and functional abilities, and high prevalence of psychiatric disorders. The HSE Model of Care for Transition in rare diseases highlights the holistic, multidisciplinary approach to patient's care evolving planning and individual approach[16]. In Ireland, a 22q11.2DS clinic has been established in one of the paediatric hospitals. The clinic is led by a general paediatrician. 195 children and adolescents are currently registered with the clinic. Clinicians from different disciplines including cardiology, immunology, plastic surgery, psychiatry contribute to the clinic. A position of a clinic coordinator was funded for a two-year period. However, integrated coordinated access to full multidisciplinary team including physiotherapy, speech and language therapy, psychology, social work to meet these patients' needs in a holistic and individualised manner is not yet established. Patients requiring such support need to be referred to services in the community - primary care, disability network teams, Child and Adolescent Mental Health Teams, many of which are under resourced, and operate with long waiting lists. A number of young people with 22q11.2DS have transitioned to a newly established adult clinic with a consultant lead. Transition guidelines for optimal transition will be developed considering challenges and strengths within the individual, the family system, along with the individual's medical and environment needs ("IF ME"). Further evaluation and resourcing of the transition process is required.

Improving transitions at different ages and between specialists/ services for individuals with 22q11.2DS who have complex needs has significant long-term benefits for these individuals and their families. Additionally, such co-ordinated services should lead to improved access to and optimal utilisation of specialty services.

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