

22q11 deletion syndrome: Parents' and children's experiences of educational and healthcare provision in the UK

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Abstract

Objective: 22q11DS is a genetic syndrome, prevalence around 1:4000-1:6000 livebirths, with a complex array of associated features, impacting on healthcare and educational support. This study reports the perceptions of families and individuals with 22q11DS in relation to these needs.

Design: Individuals and families of those with 22q11DS were approached through two national charities – the Max Appeal and 22Crew. An initial observational survey design was used to gather views via questions probing access to healthcare and educational experiences.

Results: 34 responses were received and the data subjected to descriptive analysis. Over half of respondents were diagnosed before the age of 1. 91% reported ongoing difficulties with learning at school, compounded by school attendance being compromised as a result of medical interventions. Individuals reported engaging heavily with educational support and a high number of health professions (mean 9.5; mode 10).

Conclusions: Age of diagnosis of 22q11Ds ranged from birth to 9 years. Families had ongoing concerns about aspects of education and healthcare services, and lack of knowledge and awareness of the difficulties faced by individuals with 22q11DS was raised. Healthcare and education providers should be aware of the range of services individuals required on a regular basis so as to provide a more holistic approach to care.

Keywords

22q11 deletion syndrome, education, healthcare

Introduction

Syndromes involving the soft palate known as velo-cardio-facial, DiGeorge, Schprintzen syndromes and others are now known to be due to a deletion on chromosome 22 (Cutler-Landsman, 2013) and are now all classified as 22q11 deletion syndrome (22q11DS), reflecting the common genetic basis, with prevalence estimated between 1:4000 and 1:6000 (Botto, May, Fernhoff et al., 2003). Genetic testing is now routinely offered to parents of referred children to make or confirm diagnosis (Bassett, McDonald-McGinn, Devriendt et al., 2011).

Due to the varied clinical presentation of 22q11DS, population-based estimates of the incidence and prevalence can differ (Kobrynski and Sullivan, 2007). The most widely cited UK reported incidence is that of Wilson et al. (1993) 1:4000 livebirths, and the syndrome is thought to be one of the most frequently occurring genetic syndromes (Devriendt et al, 1998). In this paper, we provide an outline of the range of difficulties that children with 22q11DSD may present with and report on a UK based survey of parent and child perspectives on the range of educational and healthcare supports provided to them.

Background

Children with 22q11DS may present with some or any of the following: cardiac problems, growth and immunological concerns, craniofacial manifestations, psychiatric illness and learning difficulties, with symptoms varying widely. An abundance of research describes the physiological and neurodevelopmental sequelae associated with the syndrome (e.g. Swillen, Vogels, Devriendt et al., 2000; Max Appeal 22q11DS Consensus Document, 2014), and it is

not the purpose of this paper to repeat information that has been provided in excellent detail elsewhere. Genetic testing is now routinely offered to parents of referred children to make or confirm diagnosis (Bassett, McDonald-McGinn, Devriendt et al., 2011).

However, following genetic diagnosis parents can be overwhelmed with this information and its relevance to their child's lifelong development (Hercher and Bruenner, 2008). The types of neurodevelopmental difficulties that children with 22q11DS deletion might face include learning and educational problems relating to attention difficulties, problems with maths, reading and social relationships as they mature, or autism (Campbell and Swillen, 2005). Children with 22q11DS are also at increased risk of developing neuropsychiatric illness in later life, including schizophrenia and bipolar disorders (Prasad, Howley and Murphy, 2008; Schneider, Debbané, Bassett, et al., 2014; Vorstman, Breetvelt, Duijff, et al., 2015).

Whilst every child will show an individual pattern of strengths and difficulties, meeting individual need is likely to involve health, social and education services over a child's lifespan, and often children will require additional educational support (Cutler-Landsman, 2013). At present no published study has been retrieved specifically of 22q11DS families' access to, use of, or satisfaction with support throughout their child's school or adult life, particularly in relation to the neurodevelopmental symptoms associated with the syndrome, although there is a developing understanding that there are discrete differences in the educational impact of various congenital conditions. For example, Reilly, Senior and Murtagh (2015) found differences in awareness of neurogenetic syndromes including 22q11DS between parents and teachers. Teachers, sometimes incorrectly, made assumptions that children with 22q11DS have similar educational needs as children with learning disability, while parents were more aware of their own child's specific needs.

In the UK, child welfare policies (Scottish Government, 2012, 2014, UK Government, 2014) aim to develop co-ordinated services for children with multiple needs. Scotland provides each child with neuro-developmental difficulties with a named individual who coordinates all support services needed. In England, those children with special educational needs who receive a co-ordinated education, health and care (EHC) plan will continue to access services such as occupational therapy, physiotherapy and speech and language therapy to support their educational development. Schools with children who do not receive such a plan act to secure additional services, to prevent fragmentation and inequality often through using a 'provision map'. This allows schools to ensure that they are able to plan for and provide support for all children and staff by documenting the range of additional support and staffing a school needs in order to secure resourcing. Schools may identify a single person to coordinate additional education and healthcare services for children with special educational needs.

It was timely therefore to explore families' and young people's perceptions of experiences in securing community based services, and how their views of service provision could be enhanced. This will offer a base-line against which policy-driven changes can be measured, and provide an indication of the range and type of services that may be required to meet each child's needs. This information might also identify specific trigger points during an individuals' school and adult lifespan where specific support services might be required, or identify services that families would have liked to access, but did not. Specifically, we were interested in finding out the views of families with children with 22q11DS of their child's educational and healthcare experiences along with those of individuals over the age of 16 with 22q11DS.

Method

The exploratory nature of the research questions above require that an initial observational survey design is used. A survey was generated using Qualtrics software, Version 1.201s of Qualtrics Copyright © 2015 Qualtrics (<http://www.qualtrics.com>), and was designed to explore the views of adults aged 16 years and over with a confirmed genetic diagnosis of 22q11DS and the views of parents/carers of individuals of any age with such a diagnosis.

Survey design

The survey questions (please see supplementary figure) explored individuals' and/or parents'/ guardians' views on education, whether individuals had additional support for learning at school; the extent to which they experience or experienced difficulties at school and their current involvement with medical services, both in tertiary and community settings. Age of the person with 22q11DS at the time of completing the survey and at the time of diagnosis was gathered, followed by five questions exploring educational provision, support and problems using a tick-box format; one likert rating scale and one free text comment; to allow respondents to describe their experiences in more detail. There followed two questions in a tick-box format to identify the health professionals that had been involved in person's care or whom participants would have liked to see, along with a further free text comment section.

<insert **supplementary figure** here>

Survey distribution

The survey was nationally distributed via family support networks. This provided a convenience sample, targeting those already involved with 22q11DS organisations, affecting

the survey's coverage. The survey's 'url' link was distributed through the two national UK based charities supporting those affected with 22q11DS: the Max Appeal and 22Crew via their website and social media. Respondents were targeted via information displayed when the url link was opened. The survey was open to respondents for a four- month period.

Participants

Thirty-four responses were received and analysed. Only one 16 year old responded, reporting on their personal experiences. Twenty-five respondents were parents and eight were carers. Descriptive information gathered via the survey was combined to represent the overall views of all respondents. 58% of respondents were informed of the study by the Max Appeal Charity and 42% from 22Crew.

Ethical permissions

Ethical permission was granted by the University Ethics Committee. Participant information and consent to complete the survey were embedded within the survey.

Data analysis

The data gathered from all respondents was combined to allow for descriptive analysis under the following subheadings which related to the survey questions:

- a. Educational support available;
- b. Educational difficulties reported by respondents;
- c. Involvement of healthcare professionals.

Free text responses were analysed qualitatively using content analysis to summarise the opinions and views offered by the respondents. These are reported to supplement the descriptive information gathered under the survey subheadings

Results and discussion

Age at diagnosis

43% were diagnosed as neonates, 12% before 12 months, 24% between 1 and 5 years, 12% between 6 and 10 years and 9% over the age of 10. The majority of cases of 22q11DS were therefore diagnosed in infancy, but others much later. This may be related to associated congenital heart disease which is found in approximately 71% of cases of 22q11DS (Shprintzen and Golding-Kushner, 2008) being diagnosed early in life. Since there is no routine antenatal screen for 22q11DS, children without diagnosed congenital heart disease may not be identified with the syndrome. This may prevent early parental counselling.

Educational support available

Respondents reported they had attended a range of educational settings from preschool to secondary levels. Support was variously in the form of a classroom assistant (part or full time); language unit attendance (part or full time), or support with some school subjects. 56% reported support of some kind during preschool; 85% at primary level, and 88% at secondary level, thus educational support increased from preschool onwards. Most respondents reported that the level of the support they received was very good or good as illustrated in Figure 1 below.

<insert **Figure 1.** here>

Educational difficulties reported by respondents.

Despite many receiving educational support, 91% of our respondents reported difficulties with learning at school. These include both curricular difficulties and general difficulties as shown in Figure 2. Curricular engagement was problematic for our participants, and it has been reported elsewhere that children with 22q11DS can have problems with aspects of numeracy (De Smedt, Swillen, Verschaffel et al., 2009); literacy (Swillen, Vogels, Devriendt et al., 2000; Antshel, Hier, Fremont et al., 2014), and attention (Marion, Scambler and Shprintzen 1993; Niklasson, Rasmussen, Óskarsdóttir et al., 2001). This can be compounded by school attendance being reduced as a result of some of the medical conditions that affect children with 22q11DS (Muenke, Kruszka, Sable et al., 2015). There is also increasing awareness of the potential consequences associated with the development of psychological problems associated with 22q11DS (Niklasson, Rasmussen, Óskarsdóttir et al., 2001).

<insert **Figure 2.** here>

While all but 2 of our respondents reported access to formalised support systems (i.e. a key worker, regular meetings with school staff, individualised education plans or additional support plans), content analysis of free text responses illuminated the kinds of issues that caused concern in relation to educational and healthcare support. The broad themes

present in these responses are summarised in Figure 3 below with the *n* indicating how frequently each theme was raised.

<insert **Figure 3.** here>

Several comments were made relating to the level of knowledge and understanding of 22q11DS in schools, both in terms of what children are capable of and what difficulties they may have. The following extracts illustrate this: *“His condition is not understood and therefore appropriate teaching strategies are not in place. Some professionals see him as naughty, stupid and disruptive”, “Feel little is known about di George so schools feel it's not relevant or the help isn't needed....”* and *“We have struggled to have people understand that the condition is a spectrum - expectations were low and we have had to fight against that...”*

Some families also reported a constant drive to ensure support was provided at the level suitable for the child: *“I feel if we weren't frequently on the case it would be a poorer education system”*.

This was further compounded by challenges relating to transitioning through different stages of the education services: *“Our child had two very different experiences in these two environments”* and *“My son had fantastic support in primary school but not very good at secondary school”*

Involvement of health care professionals

The experience of our respondents reflects involvement with a range of different professionals (as shown in Figure 4) all of whom provide specialist support. Not all cases of 22q11DS will see each and every profession, and some would like to have had access to other professions not on our list, including podiatry, psychology and social work. It was not

uncommon for families to engage with several health professions, as would be expected for families caring for children with a range of specific needs (Pelchat, Lefebvre and Perreault, 2003). The mean number of health professions consulted was 9.5 with a modal value of 10. One respondent had engaged with only one profession, with the others ranging from 6-13. The extent to which health and education professionals engage collaboratively was outwith the remit of this exploratory study. It would be interesting to explore professionals' awareness of the range of people supporting children with 22q11DS. Such a study might investigate the 'web of relationships' around families supporting their child's development, as this web is central to successful management (Dickinson, Smythe and Spence, 2006).

<insert **Figure 4.** here>

Implications of the Results

The respondents to this survey presented with a wide range of individual experiences, and it would be unwise to draw general conclusions on the basis of the small dataset presented. Notwithstanding this, there are some sensible suggestions from the participants which services might find helpful to consider.

While many children are diagnosed with 22q11DS in infancy, not all are. For some late diagnosis can result in difficulties in accessing the right support: *"Late diagnosis meant battling with the education/health authorities to try and make them understand the complex nature of 22q11.2 deletion syndrome and the associated problems that go hand-in-hand with the condition."* Families of children in this situation might benefit from some specific support to help them understand the range of difficulties that might arise and navigate the various services that can provide them with support.

One respondent suggested that education staff could benefit from further information from healthcare services: *“Teachers failed to grasp how his conditions affected his ability to carry out school work until a [sic] occupational therapist met with his pr[imary] 2 teacher and explained how hypermobility especially affects him, feel there has been greater understanding from this teacher onwards that he has a lot of issues to deal with (he looks and behaves like any other child and we felt that they believed we were making more of it than there was). Believe a medical professional explaining how medical conditions affect the children would assist teachers with understanding what is going on with the child better and perhaps make allowances for their issues.”*

This kind of role might be appropriate for the person tasked with coordinating support across education and healthcare. The survey did not explicitly explore what support strategies are most helpful for children with 22q11DS.

Limitations

There are limitations in this study. It is acknowledged that the data presented is derived from only 34 web sourced responses, so the numbers are small and respondents already had access to charities concerned with 22q11DS, and may not represent a whole-population sample. A different set of findings might have been returned from a single geographical area, through targeted sampling of families known to services. Such an approach may be valuable in auditing how services might better work together to provide useful support for families, but it may not have provided what appears to be honest and frank reporting of some of the challenges these families face and illuminating the range of experiences that might arise across geographical areas.

As stated, participants were recruited via internet based advertising, which does increase the risk of self-selection bias in the experiences of the participants sampled. The range of positive and negative experiences sampled would however indicate that this was not necessarily the case, though replication of the survey is recommended. It would be useful to explore the issues raised in this study in more detail, in particular which support strategies prove to be most useful.

Conclusion and Recommendations

Neonatal/infancy diagnosis of 22q11DS is common as a result of assessment of co-occurring cardiac involvement, but for some families a diagnosis of 22q11DS comes later in life, after educational opportunities have already been taken up. Whatever the age of diagnosis, there are aspects of educational and healthcare experiences that are of concern to individuals and their families, and services need to be prepared to provide individualised support.

While families might be good at seeking educational support at various stages in the education process, some reported having to work very hard to get it. One of the biggest hurdles appears to be the level of knowledge and awareness of the potential difficulties faced by those with 22q11DS by both education and healthcare providers. This would suggest a need for increased general awareness of 22q11DS and the kinds of difficulties that might present. An educational perspective on this has been reported in a recent study of adults with rare medical diagnoses, with the overarching conclusion that educational providers need to have a greater awareness and understanding of the problems faced by children with syndromes such as 22q11DS (Jaeger, Røjvik and Berglund, 2015). While the families' experiences may not be distinctly different to those supporting children with other

complex neurological, developmental or genetic conditions, nearly half of our survey respondents reported diagnosis taking place over the age of 5 years, after entering into a formal educational system. This risks mis-diagnosis of developmental speech sound disorders or learning difficulties at a young age, and missing out on the specialist speech investigations and interventions required. Children with 22q11DS may thus be at a disadvantage compared to those with a known neuro-disability prior to entering education, although with increased early identification this might be avoided, particularly where parents might respond to differential diagnosis depending on the age at which diagnosis is confirmed (Hallberg et al 2010). As pointed out by Hallberg et al, where children are older at the time of diagnosis parents report a sense of relief and validation of there being an underlying explanation of their child's developmental progress. This is in contrast to parents of children diagnosed at a younger age where their expectations of their child's potential is altered.

With so many healthcare professionals being involved with children with 22q11DS, we propose that awareness-raising in an educational or healthcare context only is insufficient. Healthcare providers would also benefit from considering the educational and psychosocial impacts on their patients of 22q11DS so that a more holistic approach to long-term support can be offered. Given the need to secure appropriate multi-disciplinary services for individual children, co-ordinating intervention for children with 22q11DS could be complex. Specific guidance for professionals is needed and a starting point might be that the named coordinating professional carefully constructs a child centred network of the professionals involved.

This paper has provided only an initial survey of the experience of families of children with 22q11DS. Only one respondent gave views on their personal experiences, and further research hearing the views of those with 22q11DS is required. Similarly, personal indepth discussion with families and future research into their views could inform a holistic approach to offering support, as envisaged by the current UK policy framework. Throughout this study, respondents commented on the lack of knowledge and understanding they encountered, and a general unawareness of the genetic condition. Accessible public health information about 22q11DS is recommended.

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