REACH Journal of Special Needs Education in Ireland, Vol. 27 No. 2 (2014), 103-112.

The Educational Needs of Children with 22q Deletion (22qDel) Syndrome.

An increasing number of children with special educational needs have a known genetic cause for their difficulties. The genetic condition, 22q deletion syndrome (22qDel), has an associated profile of medical, cognitive and behavioural needs. Global cognitive functioning is below average and IQ scores straddle the boundary between borderline functioning and the mild range of intellectual disability. In academic areas, children with 22qDel tend to do better at word reading and spelling tasks compared with reading comprehension and mathematics. Children with 22qDel are at high risk of experiencing significant symptoms of ADHD. Symptoms of inattention and social skills difficulties are common in the syndrome. With regard to school- based supports there is a particular emphasis on the need for intensive early intervention in mathematics, strategies to support reading comprehension and strategies to support memory. Research suggests that there is both a lack of knowledge of the syndrome and a lack of recognition of the specific educational needs of affected children in educational settings.

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INTRODUCTION

The prevalence of 22q11.2 deletion syndrome (22qDel) is estimated to be one inm4000 (Goodship, Cross, Li Ling and Wren, 1998) occurring equally in both sexes and all races. Over time a number of names have been used to refer to the syndrome including velo-cardio-facial syndrome (VCFS). Individuals with the 22qDel are still sometimes referred to as having DiGeorge syndrome but the large majority of those with 22qDel do not have true DiGeorge sequences (Shprintzen, 2005). The specific genetic cause of 22qDel, diagnosed via blood testing, was found in 1992 when a microdeletion of chromosome 22 at band q11.2 was described (Scambler, Kelly, Lindsay, Williamson, Goldberg and Shprintzen,1992). Although it is possible for the condition to be inherited, approximately ninety percent of cases are new, with neither parent affected (Swillen, Devriendt, Vantrappen, Vogels, Rommel, Fryns, Eyskens, Gewillig and Dumooulin, 1998).

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Physical Characteristics and Medical Conditions Individuals with 22qDel have a characteristic but subtle facial appearance with increased vertical length of the face and increased nasal height common (Shprintzen, 2005). Although there are 180 possible clinical features major medical features include palate anomalies ("velo") and congenital cardiovascular defects ("cardio") (Robin and Sphrinzten, 2005). While none of the features occur with one hundred percent frequency, congenital heart disease is present in approximately seventy percent of cases (Shprintzen, 2008). Other characteristics noted by Shprintzen include:

• Early feeding a difficulty

• Immune disorders are often encountered in infancy

• Chronic upper and/or lower respiratory infections in late childhood (Shprintzen).

• Other increased risk of epileptic seizures

• Chronic leg pains

• Hearing loss

• Anomalies of the kidney.

Communication, Cognition and Behaviour

One of the earliest noticeable developmental difficulties in 22qDel is a difficulty with speech and language. Onset of spoken language is typically delayed with receptive language developing more rapidly than expressive language (Glaser, Mumme, Blasey, Morris, Dahoun, Antonarakis, Reiss and Eliez, 2002). There are often significant deficits in early vocabulary acquisition and speech sound production (Golding-Kushner, 2005). Inadequate velopharyngeal closure or velopharyngeal insufficiency causes speech to sound hypernasal (excessive airflow through the nose during speech). The most common treatment for this hypernasality is surgery which can result in its immediate elimination (Golding-Kushner). Average IQ scores in 22qDel fall within the borderline range (70-79) (Green, Gothelf, Glaser, Debbané, Frisch, Kotler, Weizman and Eliez, 2009) although levels of cognitive functioning can range from average to significant intellectual disability. IQ test scores on verbal tasks tend to be better than non-verbal tasks but the difference is relatively small (Campbell and Swillen, 2005). Regarding specific aspects of cognition a strength in the area of simple auditory memory has been noted in these children (Antshel, Freemont and Kates, 2008). However, a relative weakness in visual-spatial memory and auditory memory for more complex tasks has also been found and can include a working memory deficit (Swillen, Vandeputte, Cracco, Maes, Ghesquiere, Devriendt and Fryns, 1999).

Research studies into the syndrome suggest rates of autism spectrum disorder (ASD) of between fourteen and forty-five percent (Gothelf, Frisch, Michaelovsky, Weizman and Shprintzen, 2009) (although 'full blown' autism is rarer) and rates of attention deficit hyperactivity disorder (ADHD) of between thirty and fifty-five percent (Niklasson, Rasmussen, Óskarsdóttir and Gillberg, 2002; Gothelf, Presburger, Levy, Nahmani, Burg, Berant, Blieden, Frisch, Apter and Weizman, 2004; Niklasson, Rasmussen, Óskarsdóttir and Gillberg, 2005; Antshel, Fremont, Roizen, Sphrintzen, Higgins, Dhamoon and Kates, 2006; Niklasson, Rasmussen, Óskarsdóttir and Gillberg, 2009). However, rates of diagnoses of both conditions lag behind research estimates. In a recent study of seventy-six children in the UK and Ireland only seven percent had been diagnosed with ASD and ADHD respectively (Reilly, 2012). Difficulties with inattention predominate in respect to symptoms of ADHD. With regard to social skills, children with 22qDel can struggle to understand social situations and may not pick up on social cues. Green et al. (2009) reported high rates of anxiety disorders in children with 22qDel with an increased rate of depression (Feinstein, Eliez, Blasey and Reiss, 2002). Murphy, Jones and Owen (1999) found that around thirty percent of adolescents with 22qDel were at high risk of developing psychosis.

It is not clear why children with 22qDel have such a low rate of professional diagnosis of developmental disorders and mental health difficulties. This under- recognition (Reilly, 2012) may have consequences for provision in schools, leading to lack of understanding and support for children's emotional development and lack of resources to meet their needs. The increased genetic risk for mental health problems in the syndrome can be accentuated by environmental factors such as school related stress.

Academic Achievement

Numerical and mathematical impairments have been consistently reported in individuals with 22qDel (Campbell and Swillen, 2005). DeSmedt, Swillen, Devriendt, Fryns, Verschaffel and Ghesquiere (2007) reported that compared with controls, children with 22qDel show difficulties in number comparison, the execution of a calculation strategy, word problem solving and addition and subtraction problems. According to Kok and Solman (1995), these weaknesses in mathematics seem particularly pronounced in the areas of abstract reasoning, converting language into mathematical expressions, telling time, using money and problem solving.

Creative writing, grammar and spelling are relative strengths for many children with 22qDel according to Cutler-Landsman (2007).

Many of these school-aged children perform close to, or in the average range on phonological tasks such as sentence repetition, reading and phonological awareness (Glaser et al., 2002; DeSmedt, Swillen, Devriendt, Fryns, Verschaffel and Ghesquiere, 2003).

However, reading comprehension has been identified as a significant weakness when compared with children with other genetic syndromes (Reilly, 2012). Cutler-Landsman concludes that these children seem to be adept at 'learning to read' but have more difficulty with comprehension or 'reading to learn'.

EDUCATIONAL PROVISION

Most children with 22qDel will require some type of special education support (Cutler-Landsman, Simon and Kates, 2007). Given the wide variability within the syndrome there is likely to be substantial variation with regard to the nature of this support. Furthermore, the noted lack of professional recognition of neurodevelopmental and mental health needs as outlined earlier may result in the children's needs not being formally recognised or adequately addressed particularly early in the school years. The behavioural profile and level of cognitive functioning associated with the syndrome could mean that many children in the Irish education system may not be deemed to have a low incidence disability, resulting in lack of access to the intensive one-to-one support ideally suited to their learning needs.

The majority of children with 22qDel are likely to attend mainstream schools particularly at primary level (Reilly, 2012). However, more parents of children with 22qDel would like a specialised placement (Reilly). These children can often function in a mainstream school but will need resource support and many will require special needs assistant (SNA) support to help them maintain focus and to clarify steps and directions. Difficulties may become particularly noticeable in the upper primary years as the curriculum becomes more abstract in mathematics and literacy and there is a requirement to work independently following complex oral instructions (Cutler-Landsman, 2007).

Research suggests that knowledge of 22qDel among school staff is low compared with other genetic syndromes. Fewer teachers of these children report being knowledgeable about the condition compared with teachers of children with Prader-Willi syndrome or Fragile X syndrome (Reilly, 2012). Reilly found that only three percent of teachers of children with 22qDel had training input on the syndrome in comparison with twenty-nine percent of teachers of children with Fragile X syndrome. When compared with three other genetic syndromes more UK/Irish parents of children with 22qDel were dissatisfied with current provision. They felt that the system had not met their needs and that their children were not progressing as well as they could (Reilly).

CLASSROOM STRATEGIES AND SUPPORTS

As difficulties with memory and attention are likely to be the most common issues faced by children with 22qDel in classrooms the following are some strategies for supporting them:

Table 1: Supporting children with 22qDel who have memory or attention difficulties

Memory

- Word Banks1
- Additional opportunities for repetition and practice1
- Visual step-by-step charts1
- Schedules of activities and transitions1
- Using a tape recorder while reading to remember previous parts of a book1
- Use of memory techniques (e.g. chunking, mnemonics)2
- Use of handouts of class notes2
- Time accommodations for examinations/assignments2

Attention

- Short targeted activities3
- Clear instructions - 3
- Use of visual resources - 3
- Hands-on and practical activities - 3
- One-to-one attention from supporting adult - 3
- Positive feedback and continuous reinforcement - 3
- Consistency - 3

1 Cutler-Landsman (2007); 2 Max Appeal! (2011); 3 The 22Crew (2012)

Mathematics has been identified as the greatest area of need in the curriculum by parents and teachers of children with 22qDel (Reilly, 2012). To help children in this area Cutler-Landsman (2007) suggests that it is important to begin remediation as early as possible. It is vital that children understand what numbers mean, as without a fundamental understanding of number, difficulties with mathematics will persist. The children may

appear to comprehend concepts but will only have a ‘rote understanding’ unless significant interventions are made.

According to Cutler-Landsman direct teaching will yield more success than a discovery approach particularly in terms of developing mathematical language.

Cutler – Landsman claims that early learning should emphasise larger more obvious differences in quantities and suggests the ‘min strategy’ for addition whereby children recognise it is easier to begin with the larger number when adding 7 + 2. He suggests that these children will benefit from assistance in generalising problems to other situations using role-play, real-life examples and computer assisted learning. According to Cutler-Landsman (2007) children with 22qDel will most likely require intensive support in the area of reading comprehension. With regard to social skill development, he suggests they may have deficits and need support from an early age. With adults monitoring social interactions and helping children understand the language needed for play and friendship he notes that role-playing, practising social scripts, direct instruction of appropriate behaviour and social stories can help enhance social skills as the child gets older. These children maybe over-sensitive to criticism/feedback and, as a result, may need to be taught when they should tell adults and, when they should try and deal with the situation themselves (The 22Crew, 2012).

In relation to environmental accommodations in the school setting, Cutler- Landsman (2007) advocates giving the child preferential seating near the teacher.

She contends that the classroom environment should be uncluttered, as a cluttered environment with many distracting pictures and signs will make it difficult for the child to locate materials and focus on tasks. Accordingly he claims these children will do better when pages are uncluttered and print is enlarged. The 22Crew (2012) suggests that equipment/material lists could be attached to the back of daily timetables and subjects or lessons colour coded on a timetable to support understanding. As well as these accommodations, additional time will be needed toaccomplish tasks and process information and a summary page of the key points of lessons may be useful (Cutler-Landsman).

These children may be good at copying others and while they may appear to understand concepts it will be important to question them regarding their understanding, so as not to overestimate their ability.

Children themselves may not be able to articulate their lack of understanding (The22Crew). The 22Crew also argue that having all the steps in a task on one sheet of paper gives the child the ‘big picture’ as children will have difficulty processing verbal instructions. Learning by doing is preferable with very little of what is said in a large group setting being retained and some will need one-on-one or small- group experiences to grasp concepts (Cutler-Landsman).

While children with 22qDel can often function in a mainstream education classroom with withdrawal for some subject support, many will require permanent adult classroom support keeping them focused and clarifying steps and directions (Cutler Landsman, 2007). Most problematic behaviours tend to be related to mental health issues which may be most effectively dealt with by psychological and/or psychiatric assessment and treatment. If issues do arise, teachers and parents should use modelling, verbal cues, rewards and foreshadowing of mtransitions to help with behaviour (Cutler-Landsman). The 22Crew (2012) indicates that children with 22qDel often answer questions with an irrelevant response and may need reminding that their contribution must be relevant.

CONCLUSION

Despite being one of the most common genetic conditions associated with SEN, 22qDel remains relatively unknown compared with other genetic syndromes. The absence of overt physical differences, often subtle cognitive difficulties and lack of overt behavioural needs means that the necessary supports for children with 22qDel may not be formally recognised in an expedient manner. In addition, they may miss out on vital supports due to not meeting criteria for established categories of SEN. Lack of recognition of their needs is likely to lead to a mismatch between what is expected of them and their actual capabilities, thus greatly adding to their anxiety and stress levels which are often underestimated unless formally assessed. Difficulties with reading comprehension and mathematics are particularly acute and require early recognition and intensive remediation. Social skill difficulties need to be addressed or the child may become isolated or the target of bullying behaviours. These children will struggle to learn in groups, but can make significant progress with one-to-one support. For some, there is the potential for the development of significant emotional difficulties and as a result, the child should be monitored closely with respect to mood and anxiety. It is likely that teaching resources for children with Asperger syndrome, ADHD and dyscalculia will benefit children with 22qDel. **However, all children with this syndrome need a comprehensive psychological evaluation and regular reviews to identify their needs and possible strategies for supporting them in school settings.**

REFERENCES

Antshel, K.M., Fremont, W., Roizen, N.J., Sphrintzen, R., Higgins, A.M., Dhamoon, A. and Kates, W.R. (2006) ADHD, Major Depressive Disorder,and Simple Phobias are Prevalent Psychiatric Conditions in Youth with Velocardiofacial Syndrome, Journal of the American Academy of Child and Adolescent Psychiatry, Vol. 45 (5), pp. 596-603.

Antshel, K.M., Freemont, W. and Kates, W.R. (2008) The Neurocognitive Phenotype in Velo-Cardio-Facial Syndrome: A Developmental Perspective, Developmental Disabilities Research Reviews, Vol. 14 (1), pp. 43-51.

Campbell, L. and Swillen, A. (2005) The Cognitive Spectrum in Velo-Cardio- Facial Syndrome. In Murphy, K.C. and Scambler, P.J. (Eds.), Velo-Cardio- Facial Syndrome- A Model for Understanding Microdeletion Disorders, Cambridge: Cambridge University Press, pp. 147-164.

Cutler-Landsman, D. (2007) Educational Interventions and Evaluation of Effective Practices. In Cutler-Landsman, D. (Ed.), Educating Children with Velo-Cardio- Facial Syndrome, San Diego, CA: Plural Publishing, pp. 113-215.

Cutler-Landsman, D., Simon, T.J. and Kates, W. (2007) Introduction to Education and the Neurocognitive Profile. In Cutler-Landsman, D. (Ed.), Educating Children with Velo-Cardio-Facial Syndrome, San Diego, CA: Plural Publishing, pp. 15-38.

DeSmedt, B., Swillen, A., Devriendt, K., Fryns, J.P., Verschaffel, L. and Ghesquiere, P. (2007) Mathematical Disabilities in Children with Velo- Cardio-Facial Syndrome, Neuropsychologia, Vol. 45 (5), pp. 885-895.

DeSmedt, B., Swillen, A., Ghesquière, P., Devriendt, K. and Fryns, J.P. (2003) Pre-Academic and Early Academic Achievement in Children with Velocardiofacial Syndrome (del22q11.2) of Borderline or Normal Intelligence, Genetic Counselling, Vol. 14, pp. 15-29.

Feinstein, C., Eliez, S., Blasey, C. and Reiss, A.L. (2002) Psychiatric Disorders and Behavioral Problems in Children with Velocardiofacial Syndrome:

Usefulness as Phenotypic Indicators of Schizophrenia Risk, Biological Psychiatry, Vol. 51 (4), pp. 312-318. Glaser, B., Mumme, D.L., Blasey, C., Morris, M.A., Dahoun, S.P., Antonarakis,

S.E., Reiss, A.L. and Eliez, S. (2002) Language Skills in Children with Velocardiofacial Syndrome (Deletion 22q11.2), Journal of Pediatrics, Vol. 140 (6), pp. 753-758.

Golding-Kushner, K.J. (2005) Speech and Language Disorders in Velo-Cardio- Facial Syndrome. In Murphy, K. and Scambler, P. (Eds.) Velo-Cardio-Facial Syndrome: A Model for Understanding Microdeletion Disorders, Cambridge, UK: Cambridge University Press, pp. 181-199.

Goodship, J., Cross, I., Li Ling, J. and Wren, C. (1998) A Population Study of Chromosome 22q11 Deletions in Infancy, Archives of Disease in Childhood, Vol. 79 (4), pp. 348-351.

Gothelf, D., Frisch, A., Michaelovsky, E., Weizman, A. and Shprintzen, R.J. (2009) Velocardiofacial Syndrome, Journal of Mental Health Research in Intellectual Disabilities, Vol. 2 (2), pp. 149-167.

Gothelf, D., Presburger, G., Levy, D., Nahmani, A., Burg, M., Berant, M., Blieden, L.C., Frisch A., Apter, A. and Weizman, A. (2004) Genetic, Developmental and Physical Factors Associated with Attention Deficit Hyperactivity Disorder in Patients with Velocardiofacial Syndrome, American Journal of Medical Genetics, Vol. 126 (B), pp. 116-121.

Green, T., Gothelf, D., Glaser, B., Debbané, M., Frisch, A., Kotler, M., Weizman, A. and Eliez, S. (2009) Psychiatric Disorders and Intellectual Functioning Throughout Development in Velocardiofacial (22q11.2 Deletion)Syndrome, Journal of the American Academy of Child and Adolescent Psychiatry, Vol. 48 (11), pp. 1060-1068.

Kok, L.L. and Solman, R.T. (1995) Velocardiofacial Syndrome: Learning Difficulties and Intervention, Journal of Medical Genetics, Vol. (8), pp. 612- 618.

Max Appeal (2011) Max Appeal Website, http://www.maxappeal.org.uk (accessed October 1st 2012).

Murphy, K.C., Jones, L.A. and Owen M.J. (1999) High Rates of Schizophrenia in Adults with Velo-Cardio-Facial Syndrome, Archives of General Psychiatry, Vol. 56 (10), pp. 940-945.

Niklasson, L., Rasmussen, P., Óskarsdóttir, S. and Gillberg, C. (2002) Chromosome 22q.11 Deletion Syndrome (CATCH22): Neuropsychiatric and Neuropsychological Aspects, Developmental Medicine and Child Neurology, Vol. 44 (1), pp. 44-50.

Niklasson, L., Rasmussen, P., Óskarsdóttir, S. and Gillberg, C. (2005) Attention Deficits in Children with 22q.11 Deletion Syndrome, Developmental Medicine and Child Neurology, Vol. 47 (12), pp. 803-807.

Niklasson, L., Rasmussen, P., Óskarsdóttir, S. and Gillberg, C. (2009) Autism, ADHD, Mental Retardation and Behavior Problems in 100 Individuals with 22q11 Deletion Syndrome, Research in Developmental Disabilities, Vol. 30 (4), pp. 763-773. Reilly, C. (2012) Educational Provision for a Sample of Students with Genetic Syndromes with Associated Special Educational Needs, (PhD Thesis), Dublin: University College Dublin.

Robin, N.H. and Sphrinzten, R.J. (2005) Defining the Clinical Spectrum of Deletion 22q11.2, Disability Rehabilitation, Vol. 147 (1), pp. 90-96. Scambler, P.J., Kelly, D., Lindsay, E., Williamson, R., Goldberg, M.S. and Shprintzen, R. (1992) Velo-Cardio-Facial

Syndrome Associated with Chromosome 22q11 Deletions: Encompassing the DiGeorge Critical Locus, Lancet, Vol. 339 (8802), pp. 1138-1139.

Shprintzen, R.J. (2005) Velo-Cardio-Facial Syndrome. In Cassidy, S.B. and Allanson, J. (Eds.), Management of Genetic Syndromes (2nd ed.), New York: Wiley-Liss, pp. 615-632.

Shprintzen, R.J. (2008) Velo-Cardio-Facial Syndrome: 30 Years of Study,

Developmental Disabilities Research Reviews, Vol. 14 (1), pp. 3-10 Swillen, A., Devriendt, K., Vantrappen, G., Vogels, A., Rommel, N., Fryns, J.P., Eyskens, B., Gewillig, M. and Dumooulin, M. (1998) Familial Deletions of 22q11: The Leuven Experience, American Journal of Medical Genetics, Vol. 80 (5), pp. 531-532.

Swillen, A., Vandeputte, L. Cracco, J., Maes, B., Ghesquiere, P., Devriendt, K. and Fryns, J.P. (1999) Neuropsychological, Learning and Psychosocial

Profile of Primary School Aged Children with the Velo-Cardio-Facial Syndrome (22q11 Deletion): Evidence for a Nonverbal Learning Disability? Child Neuropsychology, Vol. 5 (4), pp. 230-241.

The 22Crew (2012) The 22Crew Website, http://www.22crew.org/education (accessed October 1st 2012).

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