

and understand. The draft booklets were also given to teachers to read to ensure that, particularly the booklet for schools, provided the appropriate information. Feedback from everyone was very positive and one parent even asked if she could send the draft booklets to her child's school as they were just about to have a review meeting and they were just what she was looking for!

We engaged a professional children's graphic artist to do all the artwork which meant the booklets were more attractive to read. The funding from The Platinum Trust also meant that the booklets could then be distributed free of charge to families as well as being able to be downloaded from the website.

Four booklets were produced, Understanding Hirschsprung's Disease and Understanding Ano Rectal Malformations set out to give a simple explanation of what these conditions are. Understanding Bowel training covers the common problems encountered, such as constipation, and includes a stepped approach to good bowel management with example templates of assessment tools and care plans for schools. The Understanding bowel problems in schools booklet aims to help school staff understand why this group of children may struggle with bowel control and what their duties and responsibilities are.

The web based resource for both families and clinicians was then set up on the request of the families who wanted a single point of information access. These web pages are intended to be dynamic and develop over time as more information is added. We would therefore encourage both families and professionals to suggest any additional content to the website that they feel is missing. The web pages and links to the booklets can be found by clicking the link below:

[http://www.disabledliving.co.uk/PromoCon/Arms-\(1\)](http://www.disabledliving.co.uk/PromoCon/Arms-(1))

We are currently disseminating information about the availability of the resources via a number of related organisations, journals and networks so that any new parents of children born with ARM's will have access to appropriate information and advice so that they never feel alone or 'abandoned' again.

We would like to thank all the families and clinicians for their involvement in completing the questionnaires and reviewing the developed resources with particular thanks to the Down's Syndrome Association and The Platinum Trust for their support in enabling this project to be completed.



Dual Diagnosis Autistic Spectrum Disorder (ASD)/ Down's Syndrome (DS)

Stuart Mills, Information Officer, Down's Syndrome Association

ASD is far from straightforward, even in cases when there is no other condition present. When the child also has DS, parents have often struggled to obtain a diagnosis, faced with professionals insisting that their child's behaviour was an inevitable consequence of the syndrome. Fortunately these problems are lessening as dual diagnosis is beginning to be more widely documented.

However, having obtained the diagnosis, what then? There is currently a shortfall in information about the needs of this group and their families and supporters. Much of the information available to us has so far focussed on diagnosis. It is often said that once ASD is identified, it becomes the 'primary diagnosis', at least as far as education is concerned. Parents tell us that they feel stranded, fitting in neither with the local DS group, nor with local ASD groups, and that the DS information we provide does not apply to their child.

In 2009 we began to look at ways in which DSA might begin to address some of these issues. Our first step was to set up an email forum for parents and those with a professional interest in dual diagnosis ASD/DS. The forum has evolved into a strong source of support and information and now has 150 members. Parents use the forum to discuss their need for opportunities to meet and share experiences and to have contact with professionals with an understanding of and interest in, ASD/DS. In recognition of this we have organised a series of meetings for parents at the DSA's National Office.

• 5 October 2013

Gillian Bird (Gillian provides training, assessment and advice services for the DSA. Gillian has 30 years of experience supporting people with DS, their families and teachers.) Gillian will be talking about practical behaviour management strategies and current thinking around ASD/DS

• 5 April 2014

Professor Jeremy Turk (Consultant Child and Adolescent Psychiatrist in the South London and Maudsley NHS Trust's Behavioural Phenotype Learning Disabilities Service, and a Professor of Developmental Psychiatry at the Institute of Psychiatry.) Professor

Turk will be talking about the use of medication in managing behaviour.

• 4 October 2014

Dr Georgina Warner (Institute of Psychiatry, Kings College, London.) Dr Warner will be reporting back to the group about the results of a research project some members of the forum participated in. The research attempted to identify the range of ability and numbers of those children with DS who also have atypical behaviour.

Dr Jeremy Parr (Clinical Senior Lecturer and Consultant in paediatric neurodisability at the Institute of Neuroscience, University of Newcastle) Professor Parr will be talking about his research into prevalence and clinical and behavioural differences between DS and ASD/DS. In collaboration with the Children with DS Study (<http://www.cdss.org.uk>), and funded and supported by the DSA, Professor Parr is looking at whether screening between the ages of three and five can indicate ASD in children with DS.

We will try to ensure that there is time at each meeting for questions and for parents to have the chance to share experiences and information. If you would like to attend the meeting in October 2013, please contact Stuart Mills (Information Officer) Email stuart.mills@downs-syndrome.org.uk or Tel: 0333 1212 300. If you have any specific questions that you would like Gillian to answer, please let Stuart know.

To subscribe to the email forum send an email to: LISTSERV@listserv.down-syndrome.net with SUBSCRIBE DS-AUTISM-UK in body of message or go to: <http://listserv.down-syndrome.net/SCRIPTS/WA-DOWNS.EXE?A0=DS-AUTISM-UK> and follow instructions.

Autistic Disorder and Down's Syndrome

A Dual Diagnosis

By Dr Jennifer Dennis. Down Syndrome Medical Interest Group

Note: In this article the terms ASD, autistic disorder and autistic have been used interchangeably. In addition, in order to comply with editorial policy, the word 'Down's' has been substituted for 'Down' throughout.

In 1943 Leo Kanner, an eminent child psychiatrist working in the USA described 11 children who had a unique set of very distinctive behaviours. These included an intense need for aloneness, a lack of empathy, social communication skills and pretend play, stereotyped repetitive behaviours, and profound resistance to change. He considered this a highly recognisable behavioural syndrome and coined the phrase 'autistic' – from the Greek word 'autos' (self) – to describe these children.

Nearly 80 years earlier, in 1866, Dr John Langdon Down, a distinguished London physician had also recognised among his patients with learning disability a group with very distinctive, but in this case physical characteristics who were easily recognisable and who, because of their facial features, he referred to as 'of the Mongolian type'. His descriptions of this group are as pertinent now as they were 140 years ago and they are now referred to as having Down's syndrome. The hospital which he founded at Normansfield is now the headquarters of the Down's Syndrome Association.

It was however a further 70 years before Jerome Lejeune, a geneticist working in Paris, identified the extra chromosome 21 which is present in all those with the physical features of Down's syndrome. Hence today a positive diagnosis of Down's syndrome can be made. You either have it or you don't. This however is not the case for autism which remains a behavioural diagnosis. There is no gold standard diagnostic test. What is more the definition of the syndrome has changed markedly since Kanner's descriptions.

Today children with classic 'autism' as described by Kanner, together with those with Asperger syndrome and other children with some but not all features of either syndrome are classified as having 'Autistic Spectrum Disorder' (ASD). Despite huge efforts over decades and a plethora of diagnostic tools in the shape of questionnaires, interview schedules, observational schedules etc there still remain for some children, in the absence of a diagnostic test, uncertainty and differences of opinion as to whether he or she is or is not autistic.

There is evidence that ASD is biological in origin. That is, there is some underlying difference in some aspect of brain development which is present from birth.

Twin and family studies show that genetic and other factors are important. Among families of those with the syndrome there are often more people than expected with ASD, milder ASD behaviours or traits and other similar psychiatric disorders. Rarely a condition with features largely indistinguishable from ASD can be seen in children who have suffered very early physical or emotional abuse or neglect, for instance some of the children 'rescued' from Romanian orphanages in 1990. It is essential that these lookalike children are recognised because appropriate management strategies are different.

There is no doubt that autistic disorder is more prevalent (around 5 per 100) among those with Down's syndrome than in the general population (around 1 per hundred) but diagnosis presents problems particularly for professionals with little prior experience of the very distinctive behavioural and developmental patterns associated with the syndrome.

As already mentioned the hallmarks of the disorder are impairments in social interaction, social communication (including speech and language) and imagination. Repetitive behaviour patterns may occur as may resistance to change in routine. Bearing in mind that it is normal for children with Down's syndrome to be delayed in developing speech and language and pretend play, that many like doing the same things over and over again, and that both oppositional and opting out behaviours are common it can be quite difficult to disentangle

what is normal development for a child with the syndrome and what is abnormal.

This is where the opinion of someone with in-depth knowledge of the syndrome is important in establishing a diagnosis of ASD.

Langdon Down recognised some of these behavioural characteristics and the following extract from his writings (1887) will be recognisable today by many parents and carers.

Another feature is their great obstinacy – they can only be guided by consummate tact. No amount of coercion will induce them to do that which they have made up their minds not to do. Sometimes they initiate a struggle for mastery, and the day previous will determine what they will or will not do the next day. Often they will talk to themselves, and they may be heard rehearsing the disputes which they think will be the feature of the following day ... Whether it be the question of going to church, to school, or for a walk, discretion will often be the better part of valour, by not giving orders which will run counter to the intended disobedience, and thus maintaining the appearance of authority while being virtually beaten.

Autistic disorder should be considered, but is not necessarily the cause, in any preschool child with Down's syndrome who is failing to make expected progress in speech, language, communication and social skills and in any older child who shows regression of these skills.

I still think that one of the best articles about how parents may recognise the possibility of autistic disorder in their child is that written for the summer 2001

issue of the Down Syndrome Newsletter by Dr George Capone, from Johns Hopkins Hospital, Baltimore – *Down Syndrome and Autistic Spectrum Disorder: A Look At What We Know*. This can be accessed on the Down Syndrome Medical Interest Group website at www.dsmig.org.uk/library/articles/capone-autistic-spectrum-disorder.pdf.

One point stressed by Dr Capone is that in Down's syndrome autistic disorder may be first recognised at a later age (between 3 and 7 years old or sometimes even in the teenage years) than is usual in other children (usually by age 2-3 years).

In these later onset children with Down's syndrome the hallmarks of the disorder are loss of speech, social withdrawal, lack of initiative and a general air of not being interested in anything and just not wanting to be bothered about anything or by anyone – the intense need for 'aloneness' described by Kanner.

It is critically important that autistic disorder when present in a child with Down's syndrome should be recognised as early as possible because the dual diagnosis has implications for education and management which will carry through into adult life.

Often a diagnosis of autism may be more important as far as educational placement is concerned than the fact of a learning disability. Unfortunately because of a lack of training and knowledge of the syndrome by some healthcare and educational professionals to whom a child may be referred it is

not unusual for the diagnosis to be missed.

It is only 15 years or so ago since a 10 year old child with Down's syndrome was referred to me with severe and unmistakable autistic disorder but whose paediatrician had written:

I gather that the school are now querying whether he has autistic traits, but I explained to his mother that there is an overlap between autism and mental retardation and I do not think the label is a particularly useful or helpful one

I hope that this could not happen today but I believe that there continues to be among some professionals a certain reluctance to move down the path of investigating the possibility of a dual diagnosis of Down's syndrome and ASD. Hence it remains important that parents and carers should be resolute in seeking an informed opinion.

So how is a diagnosis of autism achieved? Usually it will start by a parent expressing concern about their child's development which they perceive as different from that of their peers with Down's syndrome. Sometimes suspicion may first be voiced by a healthcare, educational, or social care professional who has knowledge both of children with Down's syndrome and of autistic disorder.

Once suspicion has been raised the child is usually referred to a specialist multidisciplinary team who will take a detailed history from the parents, observe the child in a clinic setting and often at home with family members and in

school or nursery or some other group situation with which the child is familiar.

A variety of special diagnostic tools – questionnaires, interviews and observational schedules – will usually be used in parallel with history taking and clinical observation but these tools are never used in isolation to establish a diagnosis. They do not in themselves provide a definitive answer to the question 'Does my child have autism?' A specialist will take all the information together to advise you of their opinion.

Parents of children and even adults with a dual diagnosis of Down's syndrome and ASD nearly always say that they wish this could have been recognised at an earlier age. With this in mind a number of research teams are setting out to raise awareness of the dual diagnosis of Down's syndrome and autistic disorder and the need of making an early ASD diagnosis. In particular they want to find out how ASD can be identified in very young children with Down's syndrome.

The research teams have had a positive response from parents of young and older children with and without ASD and through consultation, parents are influencing the direction of research. The researchers are investigating whether screening questionnaires which are already known to be useful for other young children can also be used effectively for those with Down's syndrome. This is a much needed initiative and we look forward to its continuing development.

Contribute your experiences to the DSA Journal

Is your child using a communication aid?

If your child uses any type of assistive communication technology, this may be an app on a tablet/smartphone or a program on a laptop/desktop PC, please contact us. We would like our readers to review these for the journal.

This technology allows people to talk by accessing a large vocabulary of symbols or text. It may be used at home or when going out and about. Additionally it may be used in speech therapy.

There are a range of apps and programs available and we are keen to share your experiences with our readers.

Your stories

We are always interested in receiving general articles and personal stories on any aspect of life with Down's syndrome. This may cover any stage of life from birth to adulthood. These articles could include your experiences around education, health, sport, using new technology or work. If you have an interesting story we would like to hear from you.

If you would like to write something for us please contact: Ian Jones-Healey, DSA Journal Editor ian.jones-healey@downs-syndrome.org.uk