

## Dual Diagnosis Down Syndrome and Autistic Spectrum disorder

### Warning

The diagnosis of Autistic spectrum disorders (ASD) needs a trained team working within specific guidelines and should not be attempted by parents who have merely read this article. Many children with Down syndrome will have some autistic like behaviours, this does not mean they have ASD. Hearing loss (common in DS) can also produce such behaviours.

This article is intended to give very basic information and a little understanding of autistic spectrum disorders and perhaps start alarm bells ringing if your child needs assessment.

### Why should DS-ASD matter to all of us?

These two quotes highlight our failings in supporting families who have a child with DS-ASD and challenge us to do more.

“Today I know the isolation of being the mother of a child who is different from his peers with Down syndrome.

It has been four years since our lives changed dramatically due to Andy’s autism. I don’t want to live them again. They were lonely and full of despair. If you have a friend or know someone in your support group who is having similar struggles, don’t wait for them to ask you for help.... They aren’t coming to you or to the group meeting because it hurts or it is too hard to leave their child right now. If you want to help show up at their door and offer to clean, cook, do the laundry or sit and talk....

....The gift of your time without judgment, will mean the world to them. It *still* does to me. After all we’re far from done with this journey.”

*Joan E. Guthrie Medlan, parent (1)*

“Unfortunately, I have found that parents in this situation [with child with DS-ASD] almost universally withdraw from local Down syndrome support groups.... There are a variety of reasons for this including “the topics discussed don’t apply to my child, “It’s just too hard to see all those children doing so much more than my child,” and “I feel like people think I’m a bad parent because of my daughter’s behaviour.”

Ideally someone in the parent group would recognize this when it is happening and offer additional support instead of watching them withdraw.”

*George T. Capone, paediatrician .(1)*

### ASD, DS & DS-ASD what’s the difference?

When I searched for academic articles on Down syndrome and autistic spectrum disorder (ASD), I found loads comparing children with DS and those with ASD, but very few about children with the dual diagnosis.

It is only in the last few years that the possibility of a child having both conditions has been entertained, with literature starting around 1995 (2). Previously (and often today as parents may discover to their frustration) unusual patterns of development in language or behaviour were assumed to be related to severe or profound mental retardation.

To some extent Down syndrome may mask the diagnosis of autistic spectrum disorder, as some autistic characteristics are normal (although often to a lesser level) in a child with Down syndrome. The table opposite compares and contrasts the conditions.

Medical content has been checked by developmental paediatrician Dr Giles Bates.

Down syndrome (DS)	Autistic Spectrum Disorder (ASD)	Dual Diagnosis Down Syndrome and Autistic Spectrum disorder DS-ASD
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	A life-long condition causing delays in learning and development	Difficulty in making sense of the world. Developmental disorder of social and communication skills	A life-long condition causing major difficulties in social and communication skills and delays in other areas of learning and development.
<b>Occurrence</b>	1 in 1000, steady occurrence rate no link to external factors	between 1 in 1000 and 1 in 100 depending on definition  rate possibly increasing (may just be better diagnosis)  many possible links to external factors	around 5-7% of people with DS may have associated ASD (1,3)
<b>Diagnosis</b>	At birth  Objective diagnosis  By a simple blood test	As a toddler/child  Subjective diagnosis  after multiple investigations by a team of professionals	often not till child/adult (although earlier diagnosis is possible)  Subjective diagnosis  after multiple investigations by a team of professionals/specialist clinic
<b>Due to</b>	Extra chromosome 21 material	Unknown (although various suggestions/relationships described)	may be associated with family history of ASD, infantile spasms, hypothyroidism, brain injury (1,4,5)
<b>Developmental delay</b>	across all areas of development	very patchy distribution	often regression or slowing of development especially in language and behaviour
<b>cognitive delay</b> <i>(intelligence)</i>	always	often but wide spectrum of intelligence	often moderate delay (not the profound delay which itself causes autistic like behaviours without DS-ASD diagnosis)
<b>strengths</b>	people skills, social skills, imitation	motor skills, cognitive skills	visual learning
<b>weaknesses</b>	language, motor skills, cognitive skills	language, people skills, social skills, imitation	especially language (people skills less impaired than children with ASD only)
<b>play</b>	like a younger child  but with usual understanding of pretend play and how to use a toy	unusual play; may focus on parts of a toy, or line toys up  little pretend play	unusual play
<b>useful interventions/treatment</b>	medical treatment of heart, bowel, vision and hearing problems  speech language therapy especially signing and reading  patient repetition, using visual and social methods of learning with small incremental steps	speech language therapy especially picture exchange communication systems  psychological programmes eg. Applied Behavioural Analysis & developmental social programmes  sensory integration therapy  drug treatment for some behaviours	more intense therapy than is usual for children with DS (therapies as for ASD)

stares at  
fingers

no eye  
contact

DOESN'T  
TALK

upset by  
change

food  
refusal

stares at  
aloneness

fascination  
with lights

REPETITIVE  
MOTOR  
BEHAVIOURS

seems  
deaf

flapping  
hands

## How does DS-ASD present?

Gleen Vatter (6) suggests four key behaviors that may point to the possibility of ASD in a child with Down syndrome:

### 1) Extreme Autistic Aloneness

- does not relate to people normally
- prefers to be left alone
- considers other persons as objects, not people
- will not join in group play
- does not want to be held.

### 2) Anxiously obsessive desire for the preservation of sameness

- differences in daily routines cause a large upset.

### 3) Lack of eye contact

- does not make eye contact but will look away or “right through” other people.

### 4) Shows repetitive, “Stereotypical” movement

- eg. sits for long periods of time just waving an object and looking at it.

George Capone (1) has followed 30 children with DS-ASD over 6 years, he separates presentation into two groups

#### Group one

1) May have **associated medical conditions** eg. Seizures, dysfunctional swallowing, nystagmus or severe hypotonia

#### 2) Atypical Behaviour occurs as infant/toddler eg.

- Repetitive motor behaviours (fingers in mouth, hand flapping)
- Fascination with and staring at lights, ceiling fans or fingers
- Extreme food refusal
- Receptive language problems (appears deaf)
- Repetitive or absent spoken language

#### Group two

1) Typical early development, then **onset age 3 to 7 years** (onset age 3+ means this is ‘atypical autism’)

2) **Dramatic loss (or plateauing) in acquisition and use of language and social-attending skills**

3) **May have irritability, anxiety, repetitive behaviours**

## Why diagnose DS-ASD?

It is now recognized that parents are often relieved to get a diagnosis explaining their child’s difference and the diagnosis means they can access appropriate intervention services.

## DS/ASD which matters most?

Advice from a parent of a child with DS-ASD;

“Consider the autism as the primary disability. Children may have Down syndrome written all over them, but it’s the autism that gets in the way of their learning and reacting in a way that you expect from a child with Down syndrome.”

## Improving

Speech language therapy for children with DS-ASD does not have the high focus on speech that is usual is most therapy. Instead the focus is on the process of communication in any form. This may include the use of signs, picture symbols or high tech augmentative communication devices as well as vocalization.

There must be agreement about communication methods between all members of a child’s intervention team and consistency in their use.

Communication furthers a child’s intellectual and social development and prevents the child being a passive participant who lacks control and learns to be helpless. Children who are unable to be understood may stop trying to communicate or become frustrated and resort to violent behaviours (hitting, kicking, biting) as their main form of communication.

## How is DS-ASD managed?

The key areas are

### 1) Supporting parents

In the midst of intensive therapy, it is vital that parents remember their primary role is that of a parent and that no one else can fulfill this role.

It is essential too that parents look after themselves and are able to ask for and get the support they need (see article on page 42 of this newsletter)

### 2) Language/communication (see below)

### 3) Behaviour management

### 4) Other forms of management

eg. Sensory integration therapy, drug therapy, dietary change

## Where can I learn more?

Disability Solutions is an on-line magazine for families and others interested in Down syndrome and related disabilities. Special issue, volume 3, issues 5 & 6 is a 40 page issue just about DS-ASD written in parent friendly language, and is an excellent resource.

It contains articles by parent Joan Medlen, doctors George Capone & Bonnie Patterson and speech language therapist Julie Bierman Gee from which I have quoted.

<http://www.disabilitysolutions.org/3-5.htm>

The Riverbend parent support website and Len Leshin's medical site also contain much useful information.

<http://www.altonweb.com/cs/downsyndrome/index.htm> look under medical series autism (6)

<http://www.ds-health.com/>

Autism New Zealand have a wide range of resources and hold regular training programmes. They would welcome membership from families with a child with DS-ASD. Their website is [www.autismnz.org.nz](http://www.autismnz.org.nz) and phone 0800 AUTISM.

*Angela Harper  
January 2004*

### References

- (1) Disability Solutions volume 3, issues 5 & 6
- (2) Dev Med Child Neurol. 1995 May;37(5):406-14. The recognition of autism in children with Down syndrome—implications for intervention and some speculations about pathology. Howlin P, Wing L, Gould J.
- (3) Dev Med Child Neurol. 1999 Mar;41(3):153-8. Comorbidity of autistic spectrum disorders in children with Down syndrome. Kent L, Evans J, Paul M, Sharp M.
- (4) J Intellect Disabil Res. 2000 Oct;44 ( Pt 5):562-6. Autism in Down's syndrome: a family history study. Ghaziuddin M.
- (5) Dev Med Child Neurol. 2001 Nov;43(11):750-4. Autistic disorders in Down syndrome: background factors and clinical correlates. Rasmussen P, Borjesson O, Wentz E, Gillberg C.
- (6) <http://www.altonweb.com/cs/downsyndrome/index.htm> look under medical series autism

## Language and Communication in Children with DS-ASD

Useful early techniques include;

### Following the Child's Lead

Children with DS-ASD may move frequently from one activity to the next. A natural tendency is to 'force' the child to stay with one activity but research has shown it's more effective to follow the child's shifting focus (assuming it's safe!) and find ways to interact with the child about the current interest. (For example the child starts pulling everything out of a cupboard and the caregiver signs and speaks the name of each item – "cup out, plate out, spoon out etc")

### 'Sabotaging' Situations

Deliberately engineering situations where a child has to seek out someone for help and signal their need. (For example a favourite toy is in clear view but out of reach on a high shelf or inside a plastic container or the child is all ready for a bath, but there is no bath water.)

### Using Portion Control

Setting up a situation where a child needs to request more. (For example giving very small snacks, then modeling how to sign 'more', pausing a video and modelling a signal to restart.)

### Building in Choices

Making choices throughout the day helps children realize that they can make things happen in their lives and is an important step in communication. (For example at snack time show food packaging from two favourite foods, or have photos or pictures of foods attached to the refrigerator so the child can indicate which one is wanted.)

*From Julie Bierman Gee (1) see also [www.hanen.org](http://www.hanen.org)*



encourage communication by choice of food cards on fridge



and 'sabotaging' play by putting toys in containers



# James's story

**If there was a group of similar objects on the ground he had to run his fingers through them and scatter the objects everywhere. It became an obsession and the only way to stop it was to put away out of sight anything he might "shuffle"**

James was born 8 ½ years ago in Toronto, Canada. He arrived on July 1, which is Canada Day so he is our Canadian boy! Not only was the Down syndrome not expected but also he had swallowed meconium and it got into his lungs. Hence, he stayed in hospital, on oxygen, while the meconium cleared. The doctors did the usual medical checks, he had no heart problems, and they kept an eye on him for seizures. He did not have any so after 2 weeks he was sent home.

For the first year he was fine and made good progress. After putting him onto solids and formula the health problems started, with lots of infections and diarrhoea. At the age of 3 he was tested for Coeliac disease and was positive. I put him on a gluten free diet and his health dramatically improved.

From about the age of 2, James began to "shuffle". Also he tried hard to talk and occasionally a word would pop out but instead he developed a babble of lalalala, which he repeated over and over.

By this time we were living in the United States and at the age of 3 James started a preschool disability programme at a primary school in the district. James was bussed there for 4 mornings a week. On site were all the therapists, also a psychologist, and lots of teacher aides as well as the special needs teacher. In spite of speech therapy for approximately 20 minutes twice a week he said very little and only occasionally said a word. Sign language did not work because he showed no inclination to initiate signs and was not good at imitating. His occupational therapist was lovely and spent 20 minutes working one on one with him twice a week, and this helped his learning more than anything else.

At the age of 4 ½ we returned to Christchurch. James was not able to get straight into the Champion Centre so he went to the special unit at a local kindergarten. After he had been there for a while the teacher remarked on his autistic tendencies and a little light went on in my head. By this time it was obvious his speech was delayed even by Down syndrome standards. He did not make eye contact with people other than the closest around him, did not try and engage his parents' interest in something he found interesting, did not initiate communication and his pointing at objects was poor. We were getting nowhere with the toilet training but he could watch lengthy videos for hour after hour! I was worn out looking after my very active and bright 2 year old as well as trying to toilet train James. So the teacher's comments about autism got filed away for future reference!

At age 5 ½ James started at the local school at the end of January. James did mornings only and as he tired easily we kept him at home in the afternoons. However, he enjoyed school and going on the bus with his brother to school but had difficulty staying focused and was not very cooperative. Later on, a parent who has a child with autism and also a child with Down syndrome and who helped James in the classroom commented on James's autism. By this time I had more energy and took his comments on board. I took James to see Dr. Austin, the developmental paediatrician in Christchurch. He confirmed the diagnosis of autism which I was able to add to Coeliac disease and Down syndrome. This additional label was a relief because now I understood where James was coming from, and a lot of other things that he did made sense.

I joined up with Autism New Zealand Inc as well as the Autlink Foundation. The people at the Autlink Foundation were the most helpful because they came to the school as well as our house.

**"Understanding The Autism Spectrum" is a book I am going to use as a guide to explain a little about autism and what part of James's behaviour is autism and not Down syndrome.**

**Autism means "self interest" and a person's thinking is guided by desires and effects rather than goal directed. It is a triad of impairments:**

- 1) Impairment of communication**
- 2) Impairment of social interaction**
- 3) Impairment of creative/imaginative thinking.**

**These impairments go hand in hand with rituals, obsessions, compulsions and a dread of change/ anxiety along with sensory sensitivities.**

## **Obsessions and repetitive behaviours**

In this area James is not too bad, but he does blow repetitively and flap his hands when he is excited and jumping up and down.

He does not like an unexpected change in routine and he does have to have the lights on to eat his meals and watch videos regardless of how bright it is outside.

He seldom "shuffles" these days but will still do it if he is particularly tired or unwell.

One thing I have learnt is that if I try and stop a repetitive behaviour then another will take its place!

## Communication - James;

- Will indicate needs by using an adult's hand to point, or pulls on it to show the adult something.
- Uses lots of single words eg. come, go, bus, Dad.
- Uses phrases over and over eg. "You're OK", "It's alright", "What's that", "You dumb dork"
- Poor and infrequent use of 2 words together eg. "Dad go", "Mum come".
- Is not able to say a sentence
- Eye contact is difficult for him but he is getting better at it.
- Does read facial expressions
- Does understand the meaning of gestures or tone of voice

## Sensory Sensitivities

People with autism can have a wide range of sensitivities to light, sound, people touching them, tastes, smells and so on.

**Light;** James is sensitive to light so sunhats, sunglasses, pulling curtains all help. People with autism may also see the wrong amount of light, i.e. too much blue, red or yellow light. At the moment James is trialling tinted red glasses with a magnification in the lens as he is also slightly longsighted. These glasses are definitely helping him to focus on small printed words in his books at school.

**Volume;** Too much sound really bothers James and we avoid firework displays, Santa parades, parties and rooms filled with people talking. These situations will send James ballistic whereby he covers his ears, cries, and runs as fast as he can to get out of the situation. It is possible that certain frequencies cause intense pain in his ears but as he can not communicate the problem to us we avoid these situations.

**Mouth;** James rarely tries new foods but when he does, it is put to his lips and then tossed aside.

**Taste;** Many people with autism are sensitive to many foods and prefer a bland diet. Also many have intolerances to gluten and lactose which can cause huge problems.

James does not eat any fruits or vegetables with the exception of fruit juice and a variety of vegetables on his pizza under a thick layer of cheese. He is *very* sensitive to sprays, colours and other agents. He will not eat biscuits or lollies, crisps or any other junk food with the exception of McDonald's French fries. James lives on a bland, organic, gluten free diet of rice milk, rice bread, gluten free cereal, pizza, juice, ice cream, meat and French fries. Too much ice cream will cause diarrhoea and a trace amount of gluten will trigger a horrific toilet accident! In spite of this boring and expensive diet James is very healthy and will probably live to a ripe old age.

I no longer get him to try new foods and always have on hand the foods he prefers in the fridge i.e. cooked gluten free sausages, slices of cooked gluten free pizza and his yeast free, gluten free rice bread.

## Social Interaction` - James;

- Will withdraw and not respond to others. (In the classroom he does not withdraw but in public with groups of people he will withdraw)
- Does enjoy physical contact such as tickling
- Does tolerate some affection from others
- Difficult to motivate if he does not want to do it
- Does not like to make mistakes
- Often relates best to people older or younger than himself
- Difficulty with waiting
- Needs space to retreat to (After school he has to watch videos and is difficult to motivate to do anything else)

## Creative/Imaginative Thinking -James;

- Tends not to use pretend play (bounces his soft toys on the ground or puts them in a toy car or in a toy house)
- Will focus on a part rather than the whole
- Rules of play are difficult so will not participate (Now he can play a simple board game with his teacher aide but seldom will play T-ball with his classmates outside)
- Good at imitating, especially from videos, and can follow rigidly and repetitively



## Summary

Life with James goes very smoothly and he is a very happy boy. We focus on his strengths and visual skills along with his interest in animals and water. We no longer focus on the things he cannot do or will not do.

This means at speech therapy we no longer use conventional speech therapy methods. We use PECS – Picture Exchange Communication System. With this system the emphasis is on communicating and not on speech. When James wants pizza he puts on his sentence strip (a strip of plastic with Velcro on it) the picture card for each of the following individual words: I, want, pizza, please. He gets his pizza in exchange for giving me the sentence strip regardless of whether he says anything. He has a folder full of picture cards for each of his food items, toys etc and which can be taken anywhere.

James is mainstreamed at the local school and has 2 teacher aides. One teacher aide looks after him in the morning and the other one in the afternoon so James has full coverage. We employ the morning teacher aide (have an employer number, deduct tax etc) and the ORRS funding pays for the afternoon teacher aide. Unless James has someone with him all the time he withdraws to the computer or to a corner and fiddles with things. The Correspondance School provides his preschool programme and is excellent. His social interaction at school is improving, the older children are rostered to look after him and play with him at morning break, and now he has a friend who encourages him to communicate and interact.

Outside school we encourage activities otherwise he watches too many videos because his skills at imaginative play are so poor. He has private swimming lessons, goes horse riding, goes to the respite carer to jump on their trampoline and play with their dogs, and loves going to our Down Syndrome Association socials even though he will not interact!

I believe that many of our children with Down syndrome have a little bit of autism in them but it is only when they are at school and their speech and social interaction is significantly behind the norm for Down Syndrome that we should explore autistic strategies and tools to help them.

We are encouraged by the way James continues to improve, is far more cooperative, interacts more, has more language, focuses more on things and is a very happy and healthy boy!

*Kaye Young  
Christchurch*

## Kevin's

Kevin is 11 years old and the youngest of our five children. Kevin is a happy and charming young boy. He has Down syndrome but Kevin is not like most children with Down syndrome that I know. Last year he was diagnosed with autism but getting to that point has been a long journey. Since this time we have discovered that the second diagnosis is even more important than the first in terms of helping Kevin to learn.

Kevin reached the early milestones above average for a child with Down syndrome. We invested time to teach him sign language from an early age and he was using signs well before he was two and speaking two words together. He could sing and sign and loved to interact with others. He was very competent at doing puzzles and still is. We discovered he had an interest in computers and from age two his Dad Trevor wrote software to help him and other children with special needs acquire language. Kevin progressed so well that we were invited to help establish a support group in Dunedin (later to become The Robbie White Charitable Trust) to help other children with language delay. With my teacher background, and Trevor being a software expert, we were motivated to help our young boy to develop his potential and share ideas with other parents.

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### **Kevin's idea of playing with his dinosaurs was to line them all up on the floor.**

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Looking back I think the changes began to appear by the time he was five. I wish we had received more guidance at the time but perhaps for all those working with Kevin his obvious ability made it more difficult to discern just how different he was becoming? He did not continue to progress in spoken language. As he got older he seemed to be losing language. He became more isolated and even insistent on being by himself, in fact he was stressed to be around groups of children. At school



# story

Last year he was diagnosed with autism but getting to that point has been a long journey

he would fall asleep (we now know this was a coping mechanism for sensory overload) and he became unable to cope with going into large halls with lots of noise and people. He began to be afraid of babies. He became *really* distressed when a baby was around particularly because he hated the sound of crying. He will happily allow dogs to lick him and play but small children totally intimidate him.

Kevin's idea of playing with his dinosaurs was to line them all up on the floor. He lost the ability to play creatively. He became more reluctant to make eye contact and also persisted in walking on his toes. Kevin became interested in unusual things. One of his favourites were crossword puzzles, not doing them just looking at them or collecting them. Another was junk mail because he loves the texture and he could happily spend hours rubbing a glossy piece of paper. Even the Statue of Liberty became a Kevin fascination. He could spot it anywhere and when we took him to see it in August 2003 he was one happy chap!

A therapist from the Champion Centre in Christchurch I respect was using our software and so I had kept in touch with her over the years, and she started us thinking seriously about the possibility of something further being wrong. Kevin has had the same paediatrician since birth and so we visited him about 18 months ago and this led to a referral to a psychiatrist who took very little time to make the diagnosis! It was really all too obvious.

Others' reaction to this have been interesting: from those who work with Kevin who said they had known for a long time (not sure why they never thought to tell us), to those who seemed to disbelieve the diagnosis or understate it's significance.

Ironically our language software had been proving useful for children who had autism and we had been perplexed by this since our son had Down syndrome and we were creating useful software for him. It all began to make sense.

What difference has the diagnosis made?

A huge difference. For a start we have rearranged Kevin's school day. He has always been mainstreamed in a totally supportive school but our reading about autism and the advice from the psychiatrist convinced us that we should change this. From the beginning of 2003 Kevin has the first hour of school at home. I teach him 3 mornings and Trevor 2. Fortunately we have flexible jobs that allow us to do this. As a result we can focus this time on language and use computer software to assist Kevin. It has given him a quiet, structured environment and his progress has been dramatic.

As a result of this intensive time he is a lot more confident, and he is once again gaining language, in fact his reading ability is well beyond his ability to speak. He knows he is succeeding, though progress is slow, and he is so much more motivated.

Our relationship with him has got much closer and he is spontaneously using language once more.

His teacher reports he is interacting with more confidence in the classroom and he does not want to withdraw so much at school. He has more energy. We are working to reduce the repetitive behaviours and hide the junk mail! We have joined an excellent toy library and give him a rich variety of toys.

I am just relieved we did not put off any longer gaining a formal diagnosis. It was much harder before, coping with his bewildering lack of progress. Given the dual diagnosis I have realised he is doing remarkably well and once more is making good progress.

*The Statue of Liberty became a Kevin fascination. He could spot it anywhere and when we took him to see it in August 2003 he was one happy chap!*

