Section 1
Assessed Syndromes
ASSESSSED SYNDROMES

People are always pushing her forward and letting her do things ... She is just another young girl full of life ... And that makes you want to stand up and cheer.

Parent of child with special educational needs, from ‘Something to Cheer About’, Tom Gresham in The Daily Progress, 7 May 2000

The SESS is aware that the range of assessed syndromes is wide, and while a selection are presented in this publication, it envisages that information pertaining to other specific syndromes will be presented in future issues of its newsletter, CABHAIR, which will be delivered to schools and will be accessible from the main website http://www.sess.ie/.
I’m not a ‘Down’s’. I am a person with Down syndrome ... Down syndrome is just something I have, not who I am.

*Share the Journey*, 2007 Calendar Down Syndrome Ireland

Down syndrome is a genetic condition caused by the presence of an extra chromosome. While students with Down syndrome may share certain physical traits, each student is an individual and the level of general learning disability will range from mild to profound. The student with Down syndrome may have problems such as heart defects, respiratory problems and eye defects, and may variously exhibit the following characteristics: auditory and visual impairment, delayed fine- and gross-motor skills, difficulties with thinking and reasoning and applying knowledge in new situations, limited concentration span, poor auditory memory, speech and language impairment and sequencing difficulties. Generally speaking the student with Down syndrome will be better able to understand language than to communicate it expressively. Consequently cognitive skills are often underestimated. Each student with Down syndrome should be treated as an individual whose education is based on an assessment of his/her strengths and needs. Some students with Down syndrome may have Atlanto Axial Instability (AAI), which is an abnormality in the vertebrae of the spinal column. Physical activities such as gymnastics and aquatics are particularly dangerous for those with AAI and may result in spinal damage. An X-ray is required to diagnose this condition and it is advisable that an AAI X-ray is requested in respect of students with Down syndrome who enrol in schools.
Students with Down syndrome have strong visual learning modalities. Teaching reading to students with Down syndrome should be characterised by a strong emphasis on visual learning. Visual demonstrations, pictures and illustrations can also be successfully used to assist in providing effective instruction in other subject areas of the curriculum.

The teaching of phonics and phonological awareness should not be neglected.

The use of manipulatives and activity learning can be beneficially employed in the development of number concepts. Numicon© visually-based mathematic materials have been developed with particular reference to the learning strengths and needs of the student with Down syndrome.

It is important that high expectations and predictability characterise all learning and teaching opportunities.

Students with Down syndrome generally demonstrate good social skills, which can be constructively utilised to increase learning and teaching opportunities.

Tactile demonstrations and activities also appeal to many students with Down syndrome.

Directly teach timetables, routines and school rules to students.

Speak directly to the student, using clear language and short sentences, and use appropriate and unambiguous facial expressions.

Allow adequate time for the student to process language and respond.

Structure learning and teaching opportunities to enable the student to engage in tasks with other students, who can act as appropriate role models.

Be aware that the student may become unnecessarily dependent through the availability of excessive one-to-one support.

Additional Resources/References

1. Down’s Syndrome Association (UK), website: http://www.downs-syndrome.org.uk/
2. Down’s Syndrome Scotland, website: http://www.dsscotland.org.uk/
4. Down Syndrome Ireland, website: http://www.downsyndrome.ie/
   — Including Children with Down Syndrome in Your School  
   — Including Teenagers with Down Syndrome in Your School  
   download from: http://www.downsyndrome.ie/about_us/booklets.html


Fragile X

They tell us that they want to be a firefighter and a tow truck driver; they want to have a family and a job; they understand that’s what people do. They have dreams and we will support them in anything they choose ... We love them for who they are, who they want to be and neither of us would have it any different.

The parents of twins Conor and Zach, who have fragile X, taken from http://fragilex.ca/sons.html

Fragile X is a genetic disorder, so called because of a fragile site on the tip of the long arm of the X chromosome where, although not quite separated, it looks as though the end is broken off. It is diagnosed by DNA testing on a sample of blood.

The student with fragile X will often have an unusual facial appearance characterised by large protruding ears, a long nose and a high forehead. Many may have flat feet and generally exhibit a ‘floppy’ gait owing to poor muscle tone. Learning needs can range from mild to severe, with girls usually less affected. The student with fragile X is often described as impulsive, acting before thinking, wanting everything straight away, having impaired concentration and being dependent on following a consistent routine. The student may appear uncooperative at times and exhibit oppositional type behaviours. Other behavioural habits include hand-biting/flapping, rocking and gaze avoidance. However, those with fragile X are interested in others and enjoy social contact on their own terms in a ‘safe’ environment. Inappropriate behaviours are triggered by an inability to select and organise sensory information from the environment, which causes the student to become overwhelmed by the mass of sensory input. Typically, more males are severely affected than females. It is important to remember, however, that while students with fragile X may share common traits, each student has individual and unique characteristics that need to be considered when devising learning and teaching programmes.
Structure the environment to reduce excessive sensory stimulation to a level that the student can cope with. The student with fragile X will respond positively to a calm, quiet classroom environment.

Provide the student with some freedom to move about the room and have times off-task when necessary.

Avoid, if possible, placement in a class of students with behaviour difficulties or emotional disturbance and/or behavioural problems.

Seat the student away from others in his/her own personal space.

Establish and maintain a consistent routine and ensure that the student knows what to expect and what is required during the day.

Appropriate coping and self-regulation strategies may be directly taught for the student to use when he/she feels himself/herself becoming over-aroused.

Be aware of the particular antecedent events that trigger inappropriate behaviours and plan to avoid them or to offer special help to cope if upsets are unavoidable.

The student may need a high level of individual assistance to complete tasks. The adult assisting should be quiet, calm and as unobtrusive as possible, avoiding touch and eye contact to decrease distraction levels for the student.

Avoid direct pressures (e.g. time limits, questions in front of others, eye contact or insistence on collaboration) on the student as these can be counterproductive.

Utilise the student’s preference for practical tasks, physical activity and visual learning within the learning situation.

The student may find writing difficult, so alternative recording methods may need to be explored (e.g. computer/specifically differentiated worksheets where less writing is required).

Modelling and imitation should be utilised for both behavioural and communication skills.

Mathematics can be presented in a visual and tactile manner with manipulatives, and experiential learning contexts can be used that are related to real-life experiences.

Board games and computers may be utilised for turn taking, communication, social interaction and the development of fine-motor skills.

Where possible verbal instructions should be accompanied by visuals or a practical demonstration.
Prader-Willi Syndrome

Daisy has always loved school. She participates in all areas of the curriculum with varying levels of support. She has always loved all of her teachers and had a good relationship with them. Each one ... [has] given her time and listened to her ... Daisy has a great big and fantastic personality.

Educating my daughter Daisy, Sharon Cliff, mother and teacher, taken from http://pwsa.co.uk/main.php

Caused by an abnormality on chromosome 15, this syndrome is congenital and is generally physically characterised by obesity, narrow-shaped eyes, small stature, and small hands and feet. Symptoms such as decreased muscle tone and delayed motor development are also associated with the syndrome. It ought to be remembered that each student with Prader-Willi syndrome is an individual and any one student with the syndrome will have different strengths and needs from another. Most students have some degree of general learning disability, usually in the mild range. A minority of students have been noted to have an IQ (Intelligence Quotient) of 100 or above on standardised IQ tests. Students’ reading and writing skills are often better than number skills and abstract thinking. Short-term auditory memory can be weak. However, long-term memory is considerably better. The student will often excel at sedentary activities such as computer work and colouring. Difficulties can present emotionally and socially. It is important to understand that the student’s behaviour is part of the syndrome and not usually intentional. Poor co-ordination and balance may also be observed. Affected students have an intense craving for food and may go to extreme lengths to get it. This results in uncontrollable weight gain.

Atypical Prader-Willi Syndrome: some students may have a diagnosis of Prader-Willi syndrome but will not have the typical physical features.

Acquired Prader-Willi Syndrome: symptoms associated with Prader-Willi syndrome can be ‘acquired’ by damage to, or a dysfunction of, the hypothalamus (the hypothalamus is the part of the brain that controls body temperature, cellular metabolism, and functions such as eating and sleeping). In these cases, the student will not have any of the genetic abnormalities and few of the physical characteristics of the syndrome; however, the behavioural and appetite problems that are associated with the syndrome may present. Learning and teaching techniques effective with students with true Prader-Willi syndrome can also be helpful when working with students with acquired Prader-Willi syndrome.
Tips for Learning and Teaching

- Overlearning and repetition will be necessary if the student has a poor short-term memory.
- If the student is a literal thinker, he/she may find tasks involving the use of imagination challenging. Consider incorporating real-life experiences and concrete examples into lessons.
- Many students with Prader-Willi syndrome tire easily. This needs to be considered throughout the school day and new concepts and materials are best introduced early in the school day.
- Social skills may need to be explicitly taught.
- The student with Prader-Willi syndrome may have poor auditory processing skills, which will have an impact when following instructions. Visual cues may need to be considered in this area to assist the student.
- The student may have difficulties with gross-motor skills, which may necessitate modifications to lessons such as Physical Education.
- Concrete objects and experiences should be utilised in the teaching of Mathematics.
- If the student appears frustrated when completing a task, consider breaking it up into sections for the student to complete.
- In dealing with inappropriate behaviour, note that many students with Prader-Willi syndrome respond well to a positive-behaviour reward system.
- Discuss the approach to dietary control to be adopted by the school with the parent/carer. Consult also as to how to best deal with food-taking tendencies in the school situation.
- Some students find it difficult to work in groups: consider this when adapting the teaching process.
- Consistency of routine is very important, so if change in a routine is about to occur ensure the student is warned in advance.
- Within the teaching of Physical Education the student may need extra praise and encouragement to engage in tasks.
- Be aware that students with Prader-Willi syndrome can have a high pain threshold.
- Computer work appeals to many students and this should be considered as a means for the student to engage with new material and as an alternative way to demonstrate learning.
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Additional Resources/References


2. Irish Prader-Willi Syndrome Association, website: http://www.pwsai.ie/


5. Prader-Willi Syndrome Association (UK), website: http://pwsa.co.uk/main.php


Rett/Rhett Syndrome

Living with Rett syndrome is kind of like being on a roller coaster that never stops. It has its ups and downs, the smooth but bumpy course, the twists and turns that somehow seem to come when you least expect them, and knock you back into the reality that your child is different from others, but that doesn’t mean she can’t learn and can’t lead a full and impact-filled life.

Living with Rett Syndrome, Connie Coughlin, mother whose daughter has Rett/Rhett syndrome, taken from http://members.tripod.com

Rett/Rhett syndrome affects females almost exclusively and is a neurodevelopmental condition characterised by 'normal' early development followed by a period of regression where skills are lost. As with any syndrome there is huge variation in those with Rett/Rhett syndrome and deterioration, in varying degrees, may be noticed in expressive language use, gait, general learning ability, purposeful use of hands, distinctive head movements and head growth. Rett/Rhett syndrome is classified by the Diagnostic and Statistical Manual of Mental Disorders, (DSM-IV) (4th edn) as a developmental disorder of childhood. The symptoms are generally described in terms of four stages:

**Stage 1: Early Onset** (6–18 months of age): the slowing of development is generally subtle at this stage and consequently symptoms are often overlooked.

**Stage 2: Rapid Deterioration/Destruction** (1–4 years of age): this stage may be gradual or rapid. Typically, purposeful hand use and spoken language are lost at this stage. Gait patterns are laboured and unsteady, and periods of trembling or shaking may be evident. Slowing of head growth may be noticeable. Behaviours such as hand-wringing, hand-clapping or grinding of teeth may present.

**Stage 3: Plateau/Pseudo-stationary** (2–10 years of age): apraxia (difficulties controlling voluntary muscular movements in spite of the fact that there is no actual muscle weakness), motor difficulties and seizures may present at this stage. Alertness, attention span and communication skills may improve as may a general interest in the individual’s surroundings.

**Stage 4: Late Deterioration of Motor Skills** (typically 10 years onwards): this stage can last for a number of decades and is characterised by reduced mobility, muscle weakness, rigidity, spasticity and scoliosis. The individual may lose the ability to walk (if he/she managed to acquire this skill) at this stage. Repetitive hand movements and seizures may decrease and eye gaze may improve. It is important to note that there is no cognitive loss at this stage.
It is important to work closely with the speech and language therapist and parent/carer in selecting and reviewing the most appropriate communication system for the student and in deciding whether physical movements, picture communication systems or electronic devices are most appropriate.

Hand or elbow splints can be used to reduce repetitive hand movements and increase the student’s purposeful use of the dominant hand. This would have to be discussed with the student’s parent/carer and occupational therapist.

It is important that the student is involved socially with his/her peers. Appropriate social skills and strategies may have to be explicitly taught.


2. International Rett Syndrome Foundation (IRSF), website: http://www.rettsyndrome.org/


5. Rett Syndrome Association (UK), website: http://www.rettsyndrome.org.uk/
Tourette Syndrome

I Have Tourettes, but Tourettes Doesn’t Have Me

Title of HBO documentary aired 11 December 2005, listed on http://tsa-usa.org/

It is essential that multiple motor tics (twitches) and one or more vocal tics (noises) are present for at least one year for a definitive diagnosis of Tourette syndrome. These tics may appear simultaneously or at different times and will tend to fluctuate in occurrence and intensity. The onset of symptoms is usually before the age of twenty-one with the average age of onset being seven. Tourette syndrome is biochemically based and genetically transmitted. Motor and vocal tics may be simple or complex.

Simple motor tics include tics of the head and face such as eye blinking, squinting, eye rolling, nose twitches, mouth twitches, making faces, tongue protrusion, nodding or tilting of the head sideways, shoulder shrugging, arm jerking and extending, leg kicking and knee jerks, and abdominal contractions such as pulling in the tummy. Complex motor tics include the smelling and licking of objects, spitting, touching parts of one’s own body and touching others, and unusual gaits such as twirling, squatting, hopping, skipping and bending down.

Simple vocal tics include sounds such as repetitive sniffing, snorting, throat clearing, coughing and gulping, while more complex vocal tics include grunting, barking, yelping, quacking, whistling and belching.

Other characteristic features of the syndrome can include palipraxia (repetitive movements such as knotting a tie over and over again), echopraxia (copying or imitating what other people do), echolalia (repeating or imitating what people say), palilalia (repeating oneself over and over, or constantly repeating the last word or phrase uttered by oneself) and copraxia (involuntary inappropriate cursing and swearing).

Most students with Tourette syndrome test within average limits on standardised IQ (Intelligence Quotient) tests. The difficulties experienced by students with Tourette syndrome in the classroom are often related to the symptoms of the disorder themselves (such as when tics disrupt other classmates or interfere with handwriting or participation in class discussions). Some difficulties are caused by co-existing symptoms such as obsessive compulsive behaviour and attention deficit hyperactivity disorder (ADHD), while others are associated with learning and academic learning difficulties. Tics such as severe head shaking, neck stretching or eye rolling may cause the student to be unable to look directly at the teacher or read easily. Hand tics often interfere with legibility of handwriting and visual spatial deficits may result in the student having difficulty with copying from the board or elsewhere. Tics may also impede activities that have strict timing criteria, which may result in lowered test scores and associated inaccurate estimates of ability.
seat the student at the side/front of the classroom in order to provide special attention for instructions/directions. this will also enable the student to refocus when necessary.

- Be vigilant to prevent any teasing that may occur.
- Provide a quiet place for the student to complete activities and tasks.
- Consider providing the student with a headset to play instrumental music in order to block out distractions.
- Allow for freedom of movement in the classroom.
- Providing the student with a small squeeze-toy to fidget with during times when concentration is needed may be of assistance.
- Break down assignments into component parts and give shorter time frames for task completion.
- Make use of a computer to reduce the need for handwriting.
- Pair with a mentor if the tic causes an activity to be unsafe (e.g. a scientific experiment).
- Establish an agreed hand gesture/signal as a reminder to refocus during listening periods.
- Put a token-reward system in place to manage student’s impulsive behaviour.
- Do not rebuke the student for engaging in tic behaviour.
- Students often have a need for a strict routine.
- Directly teach organisational skills.


7. Special Education Support Service (SESS), website: [http://www.sess.ie/](http://www.sess.ie/)

8. Tourette Syndrome Association of Ireland, website: [http://www.tsai.ie/](http://www.tsai.ie/)


I have always felt that I’m lucky to have Mellissa … but I feel in a way that she’s more special now. … Her teachers often comment on what a determined little girl she is. If she does find a task hard at school she does not let it worry her too much.

Taken from Cheryl and Mellissa’s Story, Aspects, June 2005 issue, the newsletter of the Turner Syndrome Support Society UK

Turner syndrome is a chromosomal condition that exclusively affects females. The syndrome occurs when one of the two X chromosomes normally found in females is missing or contains certain structural defects. Common characteristics associated with Turner syndrome may include short stature and a lack of ovarian development. Other physical characteristics such as a webbed neck, low set ears, low hairline, puffy hands and feet and/or arms that turn out slightly at the elbow may also be present. Some individuals with the syndrome may have scoliosis, dislocated hips, cardiovascular problems, kidney abnormalities and/or hearing difficulties. It is important to be aware that each individual presents with different combinations of symptoms. In the school situation the student with Turner syndrome may experience difficulties with mathematical and visual spatial problems. Social and/or emotional problems may also present. Research suggests that girls with Turner syndrome are weak in mathematical achievement relative to reading achievement and verbal performance.
Tips for Learning and Teaching

- Provide for opportunities within school to develop the self-esteem and sense of belonging for the student.
- Explicitly teach relaxation exercises.
- Organisational skills may need to be taught and the use of reminder signs and notes, and the use of colour-coded books and copies, may be of use in this regard.
- For some girls with Turner syndrome difficulties present when there is an unexpected change in routine. If the student is likely to encounter such difficulties, give her advance warning and explicit descriptions of what will happen. Having a ‘Buddy system’ may work well here.
- Do not overprotect the student. Apply the same rules, expectations and consequences, within reason.
- Bear in mind the short stature of the student and create modifications to the classroom, if necessary, to accommodate her needs.

Additional Resources/References

4. The Irish Turner Contact Group. Tel: 085 7042627.
   This has a list of publications and information pertaining to the syndrome from which items can be ordered. This includes information leaflets for teachers, website: http://www.tss.org.uk/info/TSSS%20Publications.pdf
Usher Syndrome

People with Ushers do not need people to think, ‘How terrible, this person is deaf and blind; there is nothing to do.’ What we need is for people to say, ‘This person is deaf and blind. What can I do to facilitate his achievement of his full potential?’

Lawrence W. Lee, 43 year old Afro-American with Usher syndrome.

Usher Syndrome is inherited and presents in the individual having difficulties with both hearing and vision. It is a major cause of Deafblindness (see page 144). Symptoms of Usher syndrome include hearing impairment and retinitis pigmentosa (an eye disorder that causes one’s vision to deteriorate over time).

There are three types of Usher syndrome:

Type I: the student is profoundly deaf at birth and will have difficulties with balance. Typically, develops vision problems by the age of ten.

Type II: the student is born with moderate to severe hearing loss and has normal balance characteristics. Most perform well in school and some may benefit from hearing aids. Visual problems progress at a slower rate than Type I and blind spots can begin to appear when the student enters adolescence at which time the student may experience stress and embarrassment.

Type III: the student is born with normal hearing and balance. However, hearing and vision deteriorate over time, at a rate varying between individuals. Typically, students develop notable hearing problems by adolescence. Blind spots appear from late adolescence onwards and by mid-adulthood the individual may have a total loss of sight.
Tips for Learning and Teaching

- The student may need specific orientation training in relation to the school buildings and grounds.
- Note that red, green and blue markers can be more difficult for the student to read when one is writing on the whiteboard.
- Lighting should be adequate but without glare.
- Windows should be behind the student, and the teacher should never teach from in front of the window.
- Contrast with background should be considered when utilising print.
- Consider using paper that is neither glossy nor glaring.
- Students may need extra time to complete work. If doing repetitive work assign them fewer questions/problems than their peers. If using a manual signing system, ensure signs are accurate and consistent.
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Additional Resources/References


4. Fighting Blindness: provides information on eye conditions including Usher syndrome, website: http://www.fightingblindness.ie/


Williams Syndrome
(Williams-Beuren Syndrome)

Meghan Finn is as charismatic as she is talented. ... [a] beautiful young lady who happens to have Williams syndrome ... that she lives and deals with courageously every day.

Taken from *Meghan Sings!*
http://www.meghansings.ws/

This is a rare congenital disorder of chromosome 7. It is diagnosed with a blood test and often can go undiagnosed for a long time. Characteristics of the syndrome are distinctive ‘elfin’ features, good auditory skills, love of music, an outgoing social nature and a gift for vocabulary.

Williams syndrome can affect the student cognitively, socially and behaviourally, while motor difficulties may also present. Motor difficulties may have an effect on handwriting, Physical Education and practical work. While some students have particular needs, it is important to note that not all of these difficulties are relevant to every student. However, they are typical of the syndrome and at least some will apply to each student. The students will probably show inconsistency in the level of their abilities across various domains.

Visual spatial needs will affect most school activities, especially practical work and Physical Education. Activities requiring spatial analysis skills, such as learning to distinguish between different letters and learning left and right, may prove a challenge. The students’ verbal abilities may be better than their cognitive abilities. Cognitively, students vary greatly from having above average abilities to moderate general learning disabilities. Difficulties with numeracy have been observed in some students as have delays in language acquisition.

Difficulties with inappropriate behaviour, such as talking out of turn in the classroom, can also present. Students may have a short attention span and become easily distracted, which can result in the student not following directions, getting out of their seat, etc. Despite the tendency for the student with Williams syndrome to have an overtly sociable nature, they can have difficulties establishing peer friendships and can be overanxious. The student may also dwell on ‘favourite’ conversational topics, showing a poor awareness of general conversational skills. The student may display difficulties in modulating emotions and may seem to over-react (e.g. tearfulness in response to what appears as mild distress).
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**Tips for Learning and Teaching**

- Incorporate frequent breaks into sustained periods when the student is required to remain on-task.
- Provide a differentiated curriculum that ensures a high degree of success for the student.
- Minimise visual and auditory environmental distractions. Be conscious of the fact that sounds such as the fire drill, school bell, etc. may result in the student becoming anxious and may also lead to behaviour problems. If possible, provide warnings prior to predictable noises. Elicit from the student the particular sounds in the school that are problematic for him/her.
- Implement a positive-behaviour reward system.
- Arrange opportunities for the students to work in small groups.
- Emotional and social skills may need to be explicitly discussed and taught, with an emphasis on the development of social skills. These skills can be taught on a whole-class basis (e.g. during Social, Personal and Health Education (SPHE)).
- Ensure consistency in all activities.
- Minimise unexpected changes in the student's routine.
- Provide a clear schedule for the student (picture-schedule perhaps for younger students, homework journal for the older student).
- Utilise role play, social stories™ and modelling to teach the student to manage anxiety-provoking situations.
- Consider a variety of friendship-building tasks such as a peer ‘Buddy system’, group work, etc.
- The student’s interest and strength in music can be capitalised on through the use of songs, instruments and recordings, wherever possible in the curriculum.
- Minimise pen/pencil and paper demands for the student who has difficulties with fine-motor skills.
- Utilise concrete objects in the teaching of Mathematics if visual-spatial skills are poor.
- Capitalise on the verbal and auditory ability of the student and incorporate these skills into differentiated lessons and activities.
- Use visual materials such as illustrations, videos, photographs, etc. as teaching aids to accompany verbal instruction. Often the student with Williams syndrome finds this approach to teaching very motivating.
- Ascertain topics that the student has a particular interest in as the student will approach a curriculum based on such topics with a high degree of motivation.
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Additional Resources/References


3. Williams Syndrome Association of Ireland, website: http://www.wsai.ie/


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Notes