Caring for a child with Down’s syndrome in the acute care setting

Geraldine Kyle discusses the common health issues associated with Down’s syndrome and the nurse’s role in supporting the child and his or her family.

Abstract

This article discusses Down’s syndrome and its effect on the child. Some common medical conditions that may necessitate a child with Down’s syndrome attending hospital either as an inpatient or an outpatient are identified. These include cardiac disease, atlantoaxial instability, growth, thyroid function and eye, ear, nose and throat issues. The nurse’s role in minimising the stress involved for the child and the family is discussed. Practical suggestions are offered for ways of enhancing practice and improving outcomes, specifically in the area of communicating with the child and family.

Keywords

Chromosomal disorders, communication, Down’s syndrome, learning disability

MANY CHILDREN’S nurses will encounter children and young people with Down’s syndrome particularly in acute care. Down’s syndrome derives its name from John Langton Down, who first described the syndrome in 1866. It is also referred to as Down syndrome. Both terms refer to the same syndrome. It is a common chromosomal disorder that causes a range of development delays and varying degrees of intellectual disability. Although having Down’s syndrome does not automatically guarantee ill health in other areas, it is useful for all children’s nurses to understand the key health issues associated with it, so they can provide comprehensive and up-to-date care for children with Down’s syndrome and their families.

What is Down’s syndrome?

Chromosomes contain the genetic material that controls the growth and function of cells in the body. Most people have 46 chromosomes, 23 of which are inherited from the mother, and 23 from the father. They influence factors as diverse as eye and hair colour, and the probability of contracting certain diseases in adulthood. Down’s syndrome occurs as a result of a chromosomal abnormality affecting the 21st chromosome. Instead of having two copies of chromosome 21, individuals with Down’s syndrome possess three copies. The majority of children with Down’s syndrome have trisomy 21, which means that there are three copies of chromosome 21 in every cell in the body (Down’s Syndrome Ireland 2011a).

There are two less common forms of Down’s syndrome known respectively as mosaicism and translocation. In mosaicism, some body cells have trisomy 21 but others do not (Leshin 2000a). Individuals with mosaicism may display some or all of the characteristics of trisomy 21, depending on how many body cells are affected. Translocation occurs when a piece of chromosome 21 is located on another chromosome. This individual has 46 chromosomes but possesses the genetic material of 47 (Paul and Paul 2008). The person with Translocation Trisomy 21 will exhibit all the same characteristics of a person with standard trisomy 21 (Paul and Paul 2008).

Down’s syndrome is not an inherited chromosomal anomaly in most cases. It occurs during cell division at conception or soon afterwards (Hartway 2009). In some cases of translocation, one of the parents is a carrier. This is because the parent has only 45 chromosomes but carries the chromosomal material for chromosome 21 on one of the other chromosomes. The carrier is unaffected by this but it can affect the chances of their children having Down’s syndrome. When a child is born and Down’s syndrome is suspected, it is common
practice to have a chromosomal analysis performed. This identifies what type of Down’s syndrome the child has. For those affected by translocation, it affords the opportunity for parents to be offered genetic counselling.

In the UK, approximately 1 in every 1,000 children will be born with Down’s syndrome (Morris and Alberman 2009). A maternal age of more than 35 is a known risk factor for conceiving a child with Down’s syndrome. When analysing the trends in live births and antenatal diagnoses in England and Wales from 1989-2008, the results of the National Down’s Syndrome Cytogenetic Register show that birth rates have remained similar over the years. However, the incidence of antenatal diagnoses rose by 71 per cent. Morris and Alberman (2009) conclude that the incidence of children being conceived with Down’s syndrome has increased, perhaps partly due to parents waiting until they are older to start families. However, this increase is offset by the uptake of prenatal screening and subsequent termination of pregnancies.

The incidence of children born with Down’s syndrome in other countries varies. For example, the incidence in Ireland is approximately 1 in 600 (Down Syndrome Ireland 2011b). This may be in part due to differences in antenatal screening options and access to facilities to terminate pregnancies.

Antenatal screening can involve tests that merely assess risk, such as blood tests to measure certain hormones and proteins, as well as a nuchal translucency scan. Mothers who are deemed to be at high risk can then opt for more invasive tests such as amniocentesis or chorionic villus sampling. These tests will provide more definitive answers but as they are invasive, they carry a one per cent risk of miscarriage (Macnair and Hicks 2010).

Background
Many children with Down’s syndrome have no health problems and live fit and healthy lives. However, there are a number of health issues commonly associated with Down’s syndrome. Internationally, health professional groups with specific interest in Down’s syndrome have conducted research and literature reviews to identify the most prevalent conditions associated with the syndrome, as well as recommendations and guidelines to assist in detecting and treating issues that arise. Some findings are presented in this article, with recommendations for the children’s nurse caring for the child with Down’s syndrome in the acute care setting.

Even a child with Down’s syndrome who is in good health should be under the care of a paediatrician who will be responsible for ensuring that all appropriate screening is carried out. This may be a community-based paediatrician who is linked to an early intervention team, or it may be a GP in a hospital setting who has a particular interest in Down’s syndrome.

Cardiac disease
Between 40 and 60 per cent of babies with Down’s syndrome will be born with some form of cardiac abnormality. The most common associated defect is atrioventricular septal defect, which will always necessitate cardiac surgery (Down’s Syndrome Ireland 2011b). It is recommended that all babies born with Down’s syndrome should have a series of investigations (Box 1).

The nurse’s role
For many parents, the diagnosis of Down’s syndrome comes as an emotional upheaval, even if it was diagnosed antenatally. Parents experience feelings often associated with the grieving process such as denial, anger, bargaining, depression and acceptance (Kübler-Ross 1969). In effect, they are grieving the loss of the baby they thought they were having and coming to terms with the arrival of a child with many perceived challenges and unknowns (Skallerup 2008). In addition, they may then learn that there is a significant risk of other health issues.

When caring for families awaiting a diagnosis of a cardiac condition, it is important for the nurse to assist them in finding a support network. There are many Down’s syndrome-specific support organisations internationally, many of which have national and regional offices. Some organisations employ nurses with a specialist interest in Down’s

Box 1  Recommended investigations to detect congenital cardiac abnormalities commonly found in babies with Down’s syndrome
- Clinical examination.
- Chest X-ray.
- An electrocardiogram and an echocardiogram should be carried out between birth and six weeks. These will assist in identifying or ruling out cardiac anomalies such as an atrioventricular septal defect.
- Children should be reviewed again if they display any signs or symptoms of cardiac disease
(Hoey and Murphy 2001, Van Cleve and Cohen 2006, Down’s Syndrome Medical Interest Group 2008)
syndrome who may be able to visit or telephone the family. In some cases it is possible to arrange a visit from a parent of an older child with Down’s syndrome to offer peer support to the new family. It is important to check whether the appropriate community nursing service has contacted the family. If a baby is not discharged from hospital for some weeks because of ongoing investigations or surgery, it is possible for parents to miss out on valuable support and advice from the community and early intervention services in their area.

Children with Down’s syndrome who undergo cardiac surgery have the same pre-operative and post-operative care needs as any other child requiring the same surgery. Graham et al (2009) discussed the importance of a rigorous screening process and the input of a multidisciplinary team that has expertise in managing children with special needs. Their study found that with these in place, good clinical outcomes are likely. Nakazawa et al (2003) discussed the fact that children with Down’s syndrome are prone to airway difficulties because of a variety of structural issues. This necessitates the careful planning of anaesthetic induction methods, as well as the possibility of delayed extubation post-operatively.

The nurse is in an opportune position to explain to parents that the surgical journey may be lengthier than normal for their child, both in preparation and in recovery, but that this does not mean that the outcome of the surgery will be compromised. Additional considerations for some children may involve additional physiotherapy input, assisted feeding methods, such as tube feeding and careful positioning, taking note of pressure areas and limb alignment.

**Atlantoaxial instability**

Children with Down’s syndrome have generalised laxity of joint ligaments as well as hypotonia. This means that they are at greater risk of cervical spine instability of the atlantoaxial joint, which may become subluxated or out of line and unstable (Song and Maher 2007). Van Cleve and Cohen (2006) recommend that children should have a neurological assessment during routine visits to their paediatrician looking specifically for signs of spinal cord compression.

They also recommend cervical spine X-rays between the age of three and five years and before participating in activities such as the Special Olympics. If the X-ray reveals a gap of more than 4.5mm between the first and second vertebrae, Van Cleve and Cohen (2006) recommend placing a restriction on certain sports. There are surgical options for atlantoaxial instability, but they would not be considered in a person without symptoms, such as paralysis or pain. Leshin (2000b) reviewed some of the evidence on the topic and noted that there are differences of opinion on the value of cervical spine X-rays in a child who has no symptoms of spinal cord compression. Hoey and Murphy (2001) and Down’s Syndrome Medical Interest Group (DSMIG) (2008) stated that such X-rays do not have predictive validity for subsequent acute dislocations at the joint and they would not recommend restricting a child with Down’s syndrome from any sporting event in the absence of physical symptoms.

**The nurse’s role**

Regardless of the results of previous cervical spine X-rays, nurses should be aware of risk factors that can occur in hospital. In situations where a child is unconscious, their head and neck should be positioned carefully to prevent joint subluxation. Before a general anaesthetic, or if the child is involved in a road traffic accident, all relevant personnel should be reminded of the risk of cervical spine instability so that precautions can be taken to minimise the risk of injury (DSMIG 2008).

**Growth**

There are a number of specific issues to be aware of with regard to growth in individuals with Down’s syndrome. Short stature is one characteristic (Giordano 1992). Obesity is another issue for many older children and adults. DSMIG (2000) recommended the use of growth charts based on measurements of healthy children with Down’s syndrome in the UK and Republic of Ireland. Regular measurements using these charts and the body mass index for older children and adults can mean early detection of any problems relating to height or weight.

**The nurse’s role**

Infants with Down’s syndrome may have a number of feeding difficulties. These include decreased endurance for feeding, decreased arousal to feed, difficulty in sucking, swallowing and aspiration of feed. These difficulties can be associated with the hypotonia associated with Down’s syndrome or may be a result of other health issues (Spender et al 1996).

Some children will require feeding via an enteral tube and may have difficulty acclimatising to oral feeding. Older children may have difficulties with new textures and adapting to cups. Down’s Syndrome Ireland in association with Our Lady’s Children’s Hospital, Crumlin and Heart Children Ireland (2011) devised a booklet for parents entitled Supporting Feeding and Oral Development in Young
Children. This is also a helpful resource for nurses when supporting and advising parents on strategies to try. Liaising with speech and language therapists and play therapists may also be required when devising plans of action.

Thyroid function, ophthalmic problems and ear, nose and throat issues

Hypothyroidism is prevalent in individuals with Down’s syndrome. This prevalence increases with age. All newborns in Ireland and the UK have neonatal screening for hypothyroidism through the heel prick test at three to five days of age. It is further recommended that all individuals with Down’s syndrome should have screening for thyroid hormones at least every two years throughout their life. Possible signs of hypothyroidism that could be investigated include significant changes in (Hoey and Murphy 2001, Van Cleve and Cohen 2006, DSMIG 2008):

- Height.
- Weight.
- Activity level.
- Cognitive level.

There is a high incidence of eyesight disorders and more than 50 per cent of people with Down’s syndrome wear glasses. Careful screening is recommended in the first four years, followed by two-yearly assessments by an ophthalmologist or optometrist for life (Hoey and Murphy 2001, Van Cleve and Cohen 2006, DSMIG 2008).

More than half of the individuals with Down’s syndrome have a significant hearing impairment. All babies with Down’s syndrome should have a full audiological assessment between six and ten months of age. Regardless of the outcome, they should have a full assessment again at age 18 months, yearly assessments until age five and two-yearly thereafter. Depending on the outcome of these investigations, the child may have assessments more regularly and will require appropriate intervention if a problem is identified (Hoey and Murphy 2001, Van Cleve and Cohen 2006, DSMIG 2008).

The nurse has a key role in streamlining the process by arranging routine blood tests at the same time as other appointments or by ensuring that audiology tests are carried out in advance of ear, nose and throat consultations, thus saving the parents an additional trip to the hospital.

Finding glasses that are suitable for children with Down’s syndrome can be challenging because of the wide and shallow bridge of their nose. This is another area where access to peer support may prove invaluable to parents. Other families may have recommendations of particular frames or opticians that provide an extensive range of suitable options.

Typically children with Down’s syndrome can have narrow ear canals and small ears. If the child requires hearing aids, they may be resistant to keeping them in place. Both glasses and hearing aids have potential to become lost or damaged in hospital. The nurse should ensure that a suitable case is available to place aids and glasses in should they need to be removed. Parents should also be asked if there are any particular strategies or routines used to aid with compliance. Being consistent with routines from home can aid a child in adapting to a new environment (Trigg and Mohammed 2006).

Sign language and communication aids

Regardless of whether or not the child has impaired hearing, many children with Down’s syndrome have delays in their speech and language development (Almeida et al 2008). This is due largely to their intellectual disability but can be exacerbated by hearing deficits. Their receptive language is usually more advanced than their expressive language, which can be frustrating for the child. The use of a sign language modified for use by individuals with an intellectual disability can be an effective bridge for the child before developing spoken words (Toth 2009).

Makaton is a system that uses a combination of speech, signs and symbols to enable individuals with an intellectual disability to communicate (The Makaton Charity 2011). It is based on British Sign Language and also has a range of written symbols. Children are taught some core signs that are useful and motivating (for example, ‘more’, ‘drink’ and ‘biscuit’) and, as they become used to the system, they can expand their vocabulary of signs. Because the word is always said with the sign, the child learns the spoken word. Most children then learn to
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say the spoken word and will gradually stop using the signs.

Lámh is the system that is used in the Republic of Ireland (Lámh 2008). It is based on Irish Sign Language and the principles of use are the same as for Makaton but the signs are quite different. Across the world, just as with spoken language, there are many variations in sign language. It is important for health professionals to be aware if a child in their care uses a sign language system. For organisations that see a large number of children who use these systems, training should be provided so that staff can learn the core vocabulary. In areas where this is not warranted, it is useful for staff to learn some of the key signs that a child uses on a case-by-case basis.

Visual learning has been shown to be a strength in children with Down’s syndrome (Buckley 2003). Using pictures and written words in communicating can aid understanding and compliance. Some children may have picture communication books/boards with them in hospital. As previously stated, Makaton incorporates symbols and pictures as one aspect of aiding communication. There are other systems such as the Picture Exchange Communication System that are commonly used (Pyramid Educational Consultants 2007). Even where a child does not formally use such a method to aid communication, it is worthwhile noting the advantages of capitalising on the visual strengths of children with Down’s syndrome. This can involve showing actual hospital equipment or using picture books and digital cameras to capture images that will be meaningful to the child’s hospital experience.

Conclusion

This article has raised some issues that need to be considered by a nurse caring for a child with Down’s syndrome in the acute setting. The main considerations are medical conditions, such as cardiac anomalies, atlantoaxial instability, growth and feeding difficulties, thyroid function, ophthalmic problems, and ear nose and throat issues. The role of the nurse in communicating with the child and family was also discussed.

All children, regardless of any predisposing syndrome, will present with individual care needs and social circumstances. Nevertheless, it is helpful to be aware of commonly occurring conditions associated with Down’s syndrome, as well as evidence-based strategies for helping the child and family through their healthcare experience.

References


