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Understanding bladder & bowel comorbidities in children & young people with additional needs – the importance of assessment

Children and young people with disabilities (both physical and learning) may present with delayed or altered bladder and bowel function and subsequently, sphincter control. This group of children represents a particularly challenging type of patient. In these affected individuals, bladder and bowel management need to be tailored as a consequence of several associated systemic and localised comorbidities, including psychological issues.

Currently the published literature on this subject is limited. Many groups have not focused on these aspects of bladder and bowel care. However there is evidence both from available literature and clinical experience those often underlying comorbidities influencing bladder and bowel function, are either missed or go unrecognised.

This is a potentially dangerous situation particularly as untreated problems can cause potential long term damage, such as renal failure or unresponsive chronic and intractable constipation ("Obstipation")

The scale of the problem

A number of studies have identified a higher incidence of lower urinary tract symptoms (LUTS) in children with physical and learning difficulties (Duel et al 2003, Handel et al 2003, Hicks et al 2007, Van Laecke et al 2001, and Roijen et al 2001). A study by de Waal et al (2009), identified individuals with moderate to severe learning difficulties have risk factors for developing

post void residuals (PVR) and a third of children with cerebral palsy in a study by Ersoz et al 2009, were also shown to have significant PVR. It appears however those children with Down's syndrome are particularly at risk of having an increased prevalence of both upper and lower urinary tract anomalies. The issue for this group of children is the sometimes familial and social low expectation of their ability to be toilet trained and achieve full continence. As a result, wetting problems are often attributed to the inability of achieving normal milestones rather than as a symptom of an underlying pathology.

Down's syndrome represents the most common group of individuals with learning difficulties and they frequently present comorbidities influencing both bowel and bladder function (gastrointestinal comorbidities: Hirschsprung's disease, duodenal anomalies, renal anomalies, bladder outlet functional obstruction, cognitive impairment, etc). Down's syndrome is the most common chromosomal abnormality and individuals with Down's syndrome have increased risk of congenital conditions including cardiac and gastrointestinal defects, as well as metabolic and renal diseases. However, renal and urinary tract anomalies have received less attention than other congenital malformations in Down's syndrome.

In 1960, Berg et al first noted the coincidence of renal anomalies and Down's syndrome with 3.5% of autopsy cases having renal malformations and later studies

showing a higher incidence of up to 21% (Ariel et al 1991). Hypospadias and urethral abnormalities (such as posterior urethral valves – PUV's) have also been noted in this population. The incidence of hypospadias has been calculated as appearing in approximately 0.3% of all live-births, with the incidence in boys with Down's syndrome being approx 6.5% - almost 20 times increased risk (Lang et al 1987).

The overall prevalence of renal and urinary tract full anomalies (RUTAs) in the Down's syndrome population is 4-5 times higher than in the general population, including the increased risk of PUV's (Kupferman 2009). Dysfunctional Voiding (DV), a condition in which the sphincter does not relax, whilst the bladder tries to expel the urine by contracting, is at higher risk in Down's syndrome than in general population (please see reference from ICCS guidelines for the management of DV). The aeiology of this condition in Down's syndrome is not clear, but it may be partly related to overtraining of the pelvic floor in an attempt to encourage the individual to stay dry, leading to DV. Dysfunctional voiding leads to functional obstruction due to associated urinary retention with increased bladder pressures. Another contributing factor is the presence of abnormalities such as unrecognised posterior urethral valves (Seki & Shahab 2011).

Hicks et al (2007) carried out a study to verify the hypothesis that boys with Down's syndrome have

bladder outflow obstruction secondary to detrusor sphincter dyssynergia (DSD). They identified high potential for renal injury: 50% of boys studied required urinary diversion for dilated upper tracts following bladder outflow obstruction, 77% had bladder dysfunction and 68% had history of wetting. They concluded that the high risk is not fully appreciated and it was important that all children and young people with Down's syndrome, particularly those with wetting problems, have detailed history and a bladder scan. This problem has also been reported in adult patients and although most common in boys, it has been reported in a female with Down's syndrome (Culty et al, 2006, Kai et al 2007)

The issue for this group of individuals is that history taking may be difficult due to impaired cognitive function and that the existence of some problems may be masked or neglected resulting in a delay in diagnosis. Voiding impairment is a common problem in this group and could be a contributory factor to the development of a DV. As a result of the relatively high incidence of urinary problems in individuals with Down's syndrome, regular review and symptom investigation should be carried out to help facilitate early diagnosis and prompt treatment intervention in order to prevent upper urinary tract deterioration.

It has been said that gastrointestinal abnormalities, both structural and functional, affect up to 77% of all individuals with Down's syndrome (Moore 2008).

Hirschsprung's disease (Congenital Megacolon) and ano-rectal malformations, including imperforate anus are more common in Down's syndrome than in the general populations and if these are not well managed and treated appropriately, early in childhood they may lead to chronic problems in adulthood.

An audit of adults with Down's syndrome attending a hospital clinic identified a wide range of problems (Wallace 2007). These included celiac disease which was likely in 12%, constipation in 19% and unexplained diarrhoea in 19%. They recommended that specially designed protocols should be developed to help identify and manage these problems appropriately.

Constipation may also be a particular problem in those with learning and physical difficulties for a number of reasons, including poor mobility and altered muscle tone. As the onset of constipation can be quite insidious it may be difficult to detect not be recognised by those individuals who have reduced ability to perceive and report their symptoms. Often the first sign that constipation may be present is that the child or young person starts to soil due to underlying faecal impaction. It is important not to presume therefore, that the development of faecal soiling is due to the person developing a behavioral issue or 'incontinence' and to ensure that they are fully investigated for the presence of any underlying constipation.

Specialist Advice – the following points should be considered:

- Try to communicate with the child or young person, no matter how difficult it may be. Disabled children and young people can communicate much more than is generally thought. Should speech be impaired, other means of communication such as drawing and using a computer may help. This will help both dealing with pain and with emotional compromise.

- In case of intractable constipation or abnormal urine output (reduced/increased frequency, 'dribbling' or with a weak flow, or as a continuous leak) rather than a normal flow: investigate for faecal impaction and have an ultrasound of the urinary system with a full bladder done and a post-void bladder scan to evaluate the post-void residual.

- If any abnormality of the urinary system is detected, early paediatric urological evaluation is required. A routine renal function blood work-up and urine analysis and culture are advised annually, in suspicious cases.

- Any required interventions including intake of medications, clean intermittent catheterisation (CIC), bowel management with oral laxatives and enemas/rectal wash-out will need to be

performed regularly and with particular care to non-recognised pain and associated risk of injury (perforation) of rectum and urethra (particularly in infants and small children).

- Sometimes, although less invasive options such as rectal washouts would normally be considered first, clinicians may tend to indicate (M)ACE or Mitrofanoff's continent stomas in this group of patients, to facilitate home-care and to drastically obviate the difficulties in communication between child and parent/carer. It is a very delicate problem. In fact, management of a stoma and post-operative complications and risks, can be also very complex in cases affected by disabilities and their comorbidities.

- The involvement of an Occupational Therapist to help facilitate correction of functional aspects that affect appropriate toileting is crucial.

- Intensive Physiotherapy should be offered to all cases with mobility limitation starting as early as possible. In fact, autonomy is mostly connected with ambulation and limb usage. This will help to prevent late onset of spastic contraction and severe limitation to clean self-intermittent catheterisation (CSIC) and bowel management.

- School toilet and home toilet accessibility and cleanliness are crucial. Teachers' level of

knowledge and understanding of the problem are of paramount importance.

Last but not least, we need to remember the following order of priority, with all aspect of bladder and bowel management in children affected by disabilities and comorbidities influencing continence and bowel function:

1) Renal Function has to be monitored and protected.
2) Bowel emptying has to be once a day at least and with soft stools.
3) Urinary infections and enterocolitis must be diagnosed early (risk of renal damage and sepsis)
4) Procedures in these patients should always be non-invasive or minimally invasive first.
5) The use of Bowel-Diary and Bladder-Diary are even more important in this particular group of patients than in other groups, to replace the reduced ability to communicate and due to the fact that different individuals will take care of this patients throughout the day.
6) A Multidisciplinary Team (MDT) approach is essential.

Implications for practice:

Currently in the UK there are no specifically studied integrated care pathways (ICP's) for children and young people with disabilities and suffering of comorbidities, influencing bladder/bowel function, especially in regards to the progressive and adequate transition from paediatric to adult care.

In many cases, families and healthcare providers are not aware of the higher risk of underlying comorbidities affecting their bowel and bladder. Any resulting urinary incontinence or faecal soiling, are unfortunately often attributed to their disability. The consequence of this is that these children are not offered even basic continence assessments or treatments, increasing the risks of urinary infections, renal and intestinal compromise.

Specific groups, such as those with Down's syndrome present with a high incidence of dysfunctional voiding, polyuria and risk of dehydration. Dysfunctional voiding and the related detrusor sphincter dyscoordination/ dyssinergia (DSD) are causes of increased intravesical pressures and increased post-void residual. In cases like these, the ability of understanding the need for regular medication intake may be reduced as a consequence of communication issues, depression and autism. So management becomes difficult even under the best circumstances.

Clean intermittent catheterisation (CIC), bowel wash-out (MACE), rectal washouts (eg. Peristeen / Qufora / Aquaflush) or enemas, laxative and tablets intake, can become difficult or impossible due to postural issues (scoliosis, wheelchair dependency), dexterity (cerebral palsy, muscle/bone disease in MPS3, etc), depression, autism, aggressive behaviour, reduced communication ability both verbal and written.

Diagnosis of urinary and bowel issues is frequently delayed. Some of these children and young people do not actually present with wetting/soiling ie incontinence. They are clean/dry, toilet trained and it is not until they have a major issue such as UTI that underlying bladder problems are picked up - it is only when their history is taken that clear abnormal voiding patterns/bowel movements are identified.

Many of these children (particularly those with autism) *'hold on'* with very infrequent voiding and are therefore at high risk of developing dysfunctional voiding with large residuals. This group of children is particularly vulnerable as many cannot verbalise their symptoms.

However, it is also recognised that simple measures such as correcting fluid intake, regular toileting and introducing medication as necessary can make a huge difference in terms of improving bladder and bowel problems.

Comorbidities influencing bladder and bowel issues in this age group of patients with disabilities are:

- A late diagnosis due to difficult verbalisation and assumption that the urinary or faecal problems are simply part of the disability picture
- Reduced dexterity and posture issues
- Renal Failure
- Associated metabolic (eg Fanconi's disease) conditions and psychological conditions (depression, psychosis)
- Short Bowel Syndrome
- Feeding difficulties
- Swallowing difficulties affecting fluid intake and diet
- Dietary intolerances
- A paradoxical or allergic reaction to essential medication (anticholinergic, alpha-blockers, laxatives, etc).
- Forgetfulness regarding medication intake or deliberate avoidance
- Lack of clinical knowledge and experience with these conditions, at a primary care level.
- Lack of trained home care and school nursing

ALL children presenting with bladder and/or bowel problems should have the same access to a bladder & bowel (continence) services as any other child or young person regardless of any associated 'disability'. Presumptions should not be made as to the cause of the bladder/bowel problems and all children should undergo appropriate assessment to exclude any underlying pathology. The risk being that untreated

bladder and bowel problems, including incontinence, will persist into adulthood

A transition plan has to be put in place for those young people, with specific tailoring for their age and in consideration of their disability.

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Bladder Incontinence Symptom Checker:
http://symptoms.rightdiagnosis.com/cosymptoms/bladder-incontinence-all.htm

PromoCon, working as part of Disabled Living Manchester, provides impartial advice and information regarding wetting and soiling problems as well as products, such as bed protection and enuresis alarms. Also services and treatments for children and adults with bowel and/or bladder problems via our confidential helpline and website.

PromoCon has produced a range of resources for children with wetting and/or soiling problems, the majority of which are free to download from our website.

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This booklet was developed with the support of The Platinum Trust