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Case Report

Peadiatrics Section

Incontinence in Intellectual Disability: An Under Recognized Cause

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ABSTRACT

Many children with Down syndrome may develop urinary incontinence during adolescence or nearing adulthood. Most often low mental ability, behavioural issues, urinary tract infection, hypothyroidism, atlanto-axial subluxation or sexual abuse may be suspected to be the reason. We report a case of Down syndrome with tethered cord syndrome (TCS) and Lipoma of Filum terminale with Cauda equina in normal position, as a cause of bowel and bladder incontinence. The need for operating with Cauda Equina in normal position is debated. But a conscious decision was taken to operate and the incontinence improved markedly which was documented by using a standardized questionnaire (King's questionnaire) and thereby making a difference in the child's life. A literature search did not result in any case of Down syndrome with tethered cord syndrome and secondary incontinence as presentation. Considering the possibility of TCS as a cause of incontinence, often neglected even in normal children, careful evaluation and correction of such problems will make a difference in the life of many intellectually disabled children. Incontinence should not be casually attributed to intellectual disability without ruling out other causes.

Keywords: Down syndrome, Urinary incontinence, Encopresis, Tethered cord syndrome, Lipomeningomyelocele

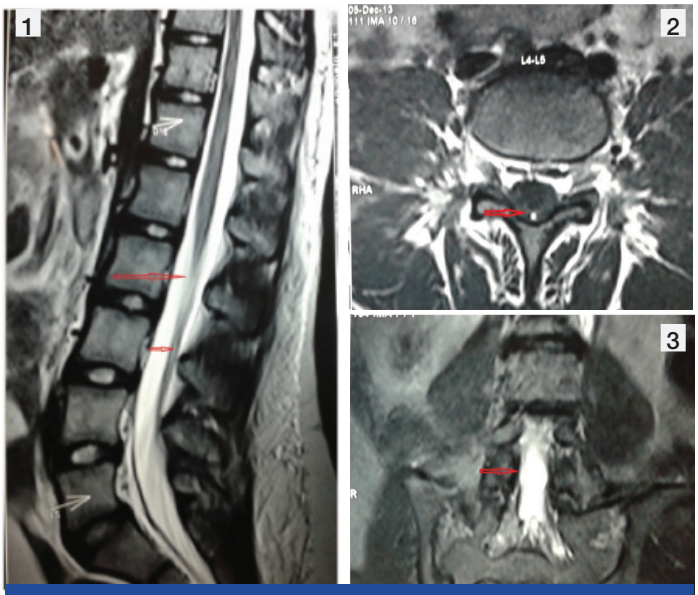
CASE REPORT

A 13-year-old girl child with Down syndrome, presented with loss of control of urine and stools for 1 month. She was the second child, born of a second degree consanguineous marriage with an uneventful antenatal and perinatal period. She was a low birth weight baby (2.17 Kg; <10thcentile for children with Down syndrome) born at term by normal vaginal delivery with a normal post natal period. Her immunization status was complete with mild delay in developmental mile stones when compared with neurotypical children of her age. She was diagnosed with hypothyroidism at the age of 10 years, on a screening following an episode of weight gain and was on regular adequate doses of L- thyroxin. She attained daytime continence by around 3 years of age and by 5 $^{1}/_{2}$ years was dry by night also. Bowel control was obtained by 3 years. But since 1month before presentation, she developed bladder incontinence both in day and night, without any history of polydipsia or symptoms of UTI. She was doing all her activities of daily living independently. She also noticed intermittent episodes of increasing bowel incontinence. She was staying with her biological parents and no physical or sexual abuse was noted. Of late, she was resisting going to school or even stepping out of home. On examination she had mild intellectual disability, but was aware of the incontinence and was feeling shy about it. She weighed 47 kg (75thcentile for children with Down syndrome) with a height of 132 cm (25thcentile for children with Down syndrome). Head to toe examination had all features of Down syndrome. Her systemic examination was unremarkable. She had little laxity in perianal sphincter with fecal soiling. She had no evidence of neurocutaneous markers, gait abnormalities or tone difference in lower limbs.

A diagnosis of Down syndrome with secondary enuresis and encopresis with hypothyroidism was made. Pelvis muscle strengthening exercises were advised for 3 weeks while awaiting investigations; but response was poor with a Clinical Global Impressions-Improvement scale (CGI-I) score 5 indicating minimally worse. King's health questionnaire on incontinence, developed independently by Linda Cardozo and Con Kelleher and recommended in NICE guidelines, 2013 [1], showed her general health perception score-75, incontinence impact score- 100, role limitation, physical limitation and sleep/energy score-83.3 each and emotions score-66.6, severity score=83.3. Her liver function tests, thyroid profile, complete blood count, electrolytes, RFT and urine culture were normal; Chest X-ray, X ray- Lumbosacral spine, USG- abdomen, USG-KUB and CT brain and spine were normal. Her urodynamic study showed nonfunctioning internal sphincter and underactive detrusor contractility. MRI spine showed lipoma of Filum terminale and tethering of cord [Table/Fig-1-3]. L3 laminoplasty with removal of lipoma and detethering of cord was done with significant recovery from incontinence within 3 weeks following surgery and got discharged in another week with full recovery by pelvic floor muscle strengthening exercises. She was also given psychological support to overcome from the trauma. CGI-I scale showed a score of 1 (very much improvement) indicating the significance of intervention. After detethering, King's health questionnaire was scored again (GHP score-0, incontinence impact score-33, role limitation-16, physical limitation-50 and sleep/energy scores-16.6, emotions score-11, severity score-0.25) showing marked improvement in various socio-emotional parameters. Till date the child has not reported any incidence of urinary or fecal incontinence.

DISCUSSION

Children with Down syndrome often have intellectual disabilities, delayed development, lower passive tone of muscles and as highlighted by Powers Mk et al., are prone for developing incontinence of urine as they enter into adult life or even before, due to multiple and often compounding reasons [2]. It may be due to behavioural issues, functional incontinence, hypothyroidism, low IQ, spinal cord abnormalities –congenital or acquired or even atlanto axial subluxation and early Alzheimer's disease [3]. Functional incontinence, which is quite common in children, refers to cases of urinary incontinence in which no structural or neurologic abnormality can be identified; but disorders of both the storage and voiding phases of the bladder cycle may be seen [4]. In children with Down syndrome, often it may be attributed to low intelligence, behavioural issues, hypothyroidism etc and even shyness and reluctance to go to school may be attributed to similar reasons and correctable causes may be missed. A recent study on urinary incontinence in Down syndrome children had found that 45% of Down syndrome children had urinary incontinence, abnormal uroflometry and possibility of complicating renal diseases [5]. A recent study by Powers et al., also proved that incontinence is more in Down's syndrome children



[Table/Fig-1]: Saggital View: Conus ending above L2; but fatty tissue (white colour) in Filum terminale (red arrow). L5 and D12 are shown in White arrow
[Table/Fig-2]: T1 axial image showing fatty tissue-Lipoma (white spot intradurally-red arrow) with tethering **[Table/Fig-3]:** A STIR Coronal view n MRI showing the Lipoma and tethering

and also secondary enuresis rate is also significantly high in Down syndrome children [2]. Even otherwise, adolescents with Tethered Cord Syndrome (TCS) perhaps are the most neglected individuals in those with a neurosurgical disease. A literature search of reports of Down syndrome with tethered cord presenting with incontinence did not yield any result.

Lipoma of the Filum terminale, is mostly incidental and is of no clinical concern except when associated with signs and symptoms of TCS. TCS is suspected when a constellation of neurological, musculo-skeletal, urological, or gastrointestinal abnormalities are associated with congenital abnormalities of spinal cord like Lipoma, meningocele, meningomyelocele, Lipomeningomyelocele or even a dermal sinus tract, with a Conus positioned at or below L-2 level [6].

It may also follow acquired causes like post spinal surgery for spinal dysraphisms (retethering) or even arachnoiditis. For lipomeningocele causing tethering, slow progression of bladder symptoms occur first often by age 2 years and motor, sensory symptom may appear during their teens. Children with TCS may develop detrusor overactivity, decreased bladder compliance, detrusor sphincter dyssynergia, and decreased sensation. Inadequate management can result in fibrosis and loss of bladder compliance, leading to high bladder pressures which can be transmitted to the kidneys, resulting in vesicoureteral reflux, nephropathy and renal failure requiring renal transplant. Yen E et al., had reported a teenager with Down syndrome presenting with acute kidney injury with significantly dilated bladder and hydronephros; the urinary obstruction of which was relieved by Foleys catheterization [7]. USG-KUB usually reveals bladder wall thickness, presence of bladder trabeculation, hydronephrosis, renal parenchymal thickness and stones in such cases. In the present case, on the contrary, urinary incontinence might have given the protective advantage to the bladder and upper tracts, as leakage happens at lower pressures minimizing the risk of developing high detrusor leak point pressures and VUR. However it may affect the quality of life of the child entering adolescence.

Lipoma with tethering and thick filum terminale may exist without any external neurocutaneus evidence. For children, thickness of the filum terminale >2 mm is considered abnormal. When the filum is thickened (with or without fat), likelihood of associated low lying cord and cord tethering is more [8]. In 92% of term babies the conus should reach L2 level or above at birth but may be lower in preterm babies, which will ascend up [9]. There are no reports of increased incidence in children with Down syndrome. In present case, conus were at L2 level with tethering and Lipoma presenting as secondary bowel and bladder incontinence. Deciding on benefits of division of a fatty filum on those who have conus at L2 with some symptoms suggesting TCS is controversial; but careful consideration may be of help in those who have symptoms, as in present case. Results of surgery on incontinence may vary; delayed response after over 6 months have been reported [10], but in our case the response was dramatic to be seen in 3 weeks time. In fact early detethering gave good outcome in 83% of cases in one study with possibility of halting neurological progression [11].

CONCLUSION

Careful consideration should be given to children with intellectual disability/limited genetic potentials like in Down syndrome who present with enuresis or encopresis and should not be attributed to low intelligence or behavioural issues alone without appropriate evaluation.

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