CASE REPORT

From disability to ability: comprehensive rehabilitation providing a holistic functional improvement in a child with neglected neural tube defect

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SUMMARY

Neural Tube defects are one of the most common congenital disorders, presenting in a paediatric rehabilitation set-up. With its wide spectrum of clinical presentation and possible complications, the condition can significantly impact an individual's functional capacity and quality of life. The condition also affects the family of the child leaving them with a lifelong impairment to cope up with. Through this 16-year-old child, we shed light on the effects of providing rehabilitation, even at a later stage and its benefits. We also get a glimpse of difficulties in availing rehabilitation services in developing countries and the need to reach out many more neglected children like him with good functional abilities.

frequent than closed defects, such as tethered cord syndrome.¹

The condition has varied presentation ranging from being an incidental finding to significant neurological deficits, depending on the neurological level and the compromise on neural tissue elements. After the initial medical care and surgical excision, lifelong management of complications and a good rehabilitation becomes a primary requisite to lead a near normal functional life in these patients.

In developing countries, with limited resources, poor accessibility and low awareness, even patients with good rehabilitation potential may get missed and may not be able to avail the required facilities. This leads to poor quality of life not only for the patient but for the family as well; affecting the overall psycho-socioeconomic dimensions in particular.

The following case is a glimpse of such a scenario in these countries. Even on providing rehabilitation at a later stage to the following child, the physical impairments and the psycho-social issues were lessened, thereby completely shifting the whole perspective of child's life. It further reduced the caregiver burden, improving the family dynamics as a whole.

BACKGROUND

Neural tube defects are a group of congenital abnormalities arising from incomplete closure of brain or spinal cord during the first trimester of pregnancy. They are one of the most common birth defects, with incidence of nearly 300 000 every year. It is categorised as open or closed based on neural tissues being exposed or covered with skin, respectively. Open tube defects include spina bifida, anencephaly and encephalocele and are more

Figure 1 ICF model for spina bifida prerehabilitation.

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Figure 2 X-ray pelvis with pseudo articular hip joint.

CASE PRESENTATION

A 16-year-old male child was brought by mother on a stretcher in a bed-bound and completely dependent state with concerns of weakness and sensory loss of bilateral lower limbs and bladder–bowel incontinence since birth. His history revealed the presence of meningocele noticed at birth for which surgical excision was done on eighth day of postnatal life. Rest of the birth history was not significant. Child had mild delay in the gross motor milestone and was able to stand with support by 2–3 years. However, due to weakness of limbs, child was unable to ambulate and spent most of the time in sitting posture. Child developed recurrent urinary tract infections and recurrent pressure ulcers due to continuous sitting and bladder–bowel incontinence. Hence, mother was advised to place child in the prone position. In order to avoid recurrent ulcers, mother preferred to keep the child in the prone posture and any attempts of sitting led to dizziness and swelling of limbs, making him bed-bound. The family issues of father leaving his family and mother being the sole breadwinner with a younger sibling, the burden on mother was high. This led her to seek help a last time. His systemic examination showed normal higher mental functions, normal cranial nerves, motor: upper limb: bulk normal bilaterally, tone normal, power: grade 5 bilaterally, hand grip: good bilaterally, no involuntary movement, lower limb: bulk: wasting present, tone: flaccid bilaterally, power: grade 0 hip, grade 1 in right quadriceps and 0 in left quadriceps, hamstring: 0 bilaterally, grade 0 bilaterally at ankle, grade 0 spine extensors, poor trunk and pelvic control, sensation: normal till T10 dermatome, decreased in T11/T12 dermatome, absent below T12, reflexes: deep tendon: biceps/triceps/supinator: 2+ bilaterally, knee/ankle jerk: absent bilaterally, superficial: plantars: mute bilaterally,

Figure 3 X-ray lumbosacral spine.

Figure 4 MRI brain.

abdominal: preserved. No cerebellar signs. No meningeal signs, gait: bed bound, no ataxia. Musculoskeletal examination showed full range in upper limbs, lower limbs: flexed deformity at hip of 80° and at knee of 70° with limb in external rotation and abducted attitude. Spine examination showed lumbar scoliosis. Neurological level was T10 complete. The International Classification of Functioning, Disability and Health (ICF) model (figure 1) sums up related factors and impact of the condition.

INVESTIGATIONS

On admission for institutional rehabilitation, routine blood investigations showed raised alkaline phosphatase levels. X-ray of pelvis (figure 2) with both hip joints was suggestive of pseudoarticular hip joints with dysplastic acetabulum with features of osteopaenia. X-ray lumbosacral spine (figure 3) showed scoliosis with apex at L2 level. MRI brain (figure 4) and spine (figure 5) were done which showed features of rhombencephalosynapsis with vertebral segmentation fusion anomaly in lumbar spine at L2 level with posterior dysraphic defects and associated tethered cord. Ultrasound abdomen showed features of neurogenic bladder and hydronephrosis.

TREATMENT

Comprehensive rehabilitation was planned with goals of independent sitting and wheelchair mobility, maximising independence for activities of daily living and bladder–bowel rehabilitation.

Orthopaedic opinion was taken regarding the pseudoarticular hip joints and in view of atrophic lower limbs and no plan

Figure 5 MRI spine.

of weight bearing; they advised no surgical intervention. Due to severe osteopaenia and financial constraints, he was started on calcium and vitamin D supplements and empirically on bisphosphonates.

He was placed on indwelling catheter and was started on tab tolterodine to reduce bladder pressure due to hydronephrosis. A trial of intermittent catheterisation, done subsequently, by nursing staff. There was no urine output on catheterisation with three trials and hence, a sinus or fistula was suspected as catheter could not be negotiated further across the firm resistance feel at the catheter tip. The indwelling catheter was placed back and he was referred to urology for cystoscopic evaluation and urodynamic study. However, none of these

Figure 6 Bed mobility: coming up to sit.

Figure 7 High sitting.

evaluations could be performed since they were not available at the institute. Monetary issues prevented referring to other costlier labs. He was taught timed pass and bowel regime for bowel management.

In therapy, he was oriented to vertical in a tilt table and initiated for bed mobility, that is, rolling and graded sitting ([figure 6](#)). The limb oedema hence, developed was evaluated and with no specific findings, was opined to be physiological oedema. A range of movement exercises (passive and active assisted) for lower limb joints and strengthening exercises including muscle stimulation was given for upper limbs. High sitting ([figure 7](#)) followed by long legged sitting ([figure 8](#)) was taught. Sitting endurance was worked over. Wheelchair transfers at the same level plinth and chair were taught ([figure 9](#)). Wheelchair manoeuvring and propulsion was subsequently started ([figures 10 and 11](#)). Tasks to maximise independence in activities of daily living were given. Hand functions including fine motor and co-ordination exercises were taught.

He was introduced to writing skills and basic numerical and alphabetical knowledge ([figure 12](#)). The ability to sit up, the recreational therapy sessions of playing throw ball ([figure 13](#)) and provision of a forum to interact with other patients who were differently abled enhanced his confidence level. He soon became a motivation to other patients and participated in numerous recreational tasks. The ICF model ([figure 14](#)) further explains the changes in activity limitations and participation restriction observed postrehabilitation.

Figure 8 Long legged sitting.

OUTCOME AND FOLLOW-UP

At the time of discharge, he was able to propel wheelchair independently with an endurance of 500 m with independent transfers. He was independent in most of his activities of daily living under set-up. He required moderate assistance for bathing and toileting.

This came as great relief on his caregiver, the mother who could now focus on other essential household and livelihood tasks. Prior to discharge, the home modifications and plans for educational and vocational opportunities were discussed.

DISCUSSION

Rhombencephalosynapsis is a rare cerebellar malformation characterised by partial or complete loss of cerebellar vermis with midline fusion of cerebellar hemispheres. It can occur in isolation or in association with other central nervous system and extracranial nervous system malformations like Gomez-Lopez-Hernandez syndrome and VACTERL syndrome. The aetiology is unknown with several observations suggestive of a genetic basis. The incidence is about 0.13%. There are only few reported cases for association of anomaly with isolated spina bifida.^{2,3} Literature shows that ataxia, abnormal eye movements, head stereotypies, behavioural impairments like attention deficits and hyperactivity, impaired cognitive functions including intellectual deficits and poor executive functions have been found to be present in these patients.^{3,4} However, at times, it may be not associated with characteristic group of neurological symptoms or any cognitive deficits or disorders preventing normal

Figure 9 Same level transfers.

everyday functioning and thus it maybe incidentally diagnosed, as was the situation in this patient.³ Child had been known to have a spina bifida through CT scan done at infancy. Hence, neuroimaging was repeated to know the status of cord at this admission at institute which showed features of this rare cerebellar malformation.

Spina bifida arises due to incomplete closure of caudal part of neural tube in a fetus and forms the second most common cause of disability in children after cerebral palsy. Both environmental factors and genetic factors play an important role in its causation.

Figure 10 Wheelchair propulsion.

Figure 11 Pressure relieving technique.

Figure 12 Writing practice.

With recent radiological advances, it is feasible to diagnose the condition in early fetal stages. Lack of early diagnosis leads to secondary motor neuron findings, sac on back with skin changes and associated vertebral deformities in the infant. The level of lesion determines the functional status of patient. The disorder impacts child development in various domains including physical, psychological and social. Physical restriction of activities, limited mobility, chronic pain and easy fatigability are seen. There is poor emotional functioning, psychological issues like depression, anxiety, lower self-esteem, limited social interaction with peer group and consequent, deficits in social skills with social rejection worsening the scenario. Moreover, these patients also experience various complications ranging from neurological, orthopaedic to renal and urogenital complications, needing lifelong healthcare provisions.⁵

With increasing age, a decline in all physical aspects is seen especially with thoracic lesions such as poor respiratory reserve, obesity, latex allergy, worsening kyphoscoliosis. About 20%–50% of children who walk become wheelchair dependent as adults. Hence, an intense preparation in childhood helps achieve a better chance of independence in adult life.⁶

The condition also affects family dynamics and functioning as the caregivers are catering to needs of the child and hence, experience an increased stress and burden in a long run.⁷

In addition, lower levels of parental satisfaction and worry for vulnerability of child has implication in teaching independence skills to these children. It has also been found that mothers from low socioeconomic strata have reduced social support and

the neural tube defects are common for the low socioeconomic strata.⁸ Hence, it is important to correctly educate the parents of these children regarding the requisite care and the functional abilities of their child. The resources/infrastructure providing greater social support at community and governmental level should be enhanced. This will facilitate proper management and rehabilitation of these children and also improve scope of community inclusion of these children and their families.

Figure 13 Recreational task: throw ball activity.

Figure 14 ICF model for spina bifida postrehabilitation. ICF,

The situation is worse in developing countries with poor socioeconomic status where not only the incidence of neural tube defects is higher, but also a major cause of health problems in surviving children.¹ The social and economic impact of this disease is not well documented. Nearly 75% of adult survivors may be dependent on parents or other providers. Even with the best support, <40% will have gainful employment.⁶

Therefore, rehabilitation in congenital neural tube defects should begin as early as in neonatal period and must continue lifelong. It is mainly aimed at maximising functional independence, prevention of complications and improving quality of life.⁵

It must be family centred and should be planned taking family scenario into consideration.⁵ Children must be encouraged to join in family events. Education, both academic and practical, must be imparted. Social skills as simple as crossing roads and shopping must be taught.

A multidisciplinary approach is considered a gold standard to manage the wide range of clinical spectrum.⁵ Research comparing different models of care in terms of outcomes in adults is, in fact, lacking. Even in developed countries like USA, majority of adults with spina bifida are not treated in multidisciplinary settings at all. There are no exclusive clinical set-ups providing a coordinated medical and rehabilitation facilities with a systematic approach to care both in adult and paediatric setting.⁹

Hence, even in an era of improved survivability and outcomes of patients with spina bifida due to advanced prenatal and neonatal management, the healthcare facilities to provide a sustained medical care and rehabilitation leading to improvement in quality of life are lacking.

In developing countries, the basic awareness and accessibility to such facilities is still lacking.

As seen in child discussed above, the mother was totally ignorant of child's abilities and the benefit she and her child can achieve of rehabilitation.

Regular counselling sessions with family and child regarding the disease, prognosis and possible outcomes helped mother and child to develop realistic goals in life. Not only did their understanding of the medical condition improve, they also learnt proper methods of managing the disease's complications. Their perspective shifted from the disability to the possible abilities of the child.

In spite of undergoing rehabilitation programme at a later age, the child showed significant functional improvement in terms of

mobility, activities of daily living, enhanced interest in education and social interactions and activities.

As the child became more self-reliant, the burden on caregiver significantly reduced; giving the mother ample opportunity to focus on other aspects of family.

The child is only one among large number of children who due to unawareness or inaccessibility or lack of comprehensive rehabilitation set-ups become victims of disability and handicap.

Even for children receiving rehabilitation at early stages, it is essential to provide a smooth and co-ordinated transition of medical care from a paediatric setting into an adult setting.⁹

Learning points

- ▶ As a whole, the case is a reminder for the need of early comprehensive multidisciplinary rehabilitation in children with congenital neural tube defects.
- ▶ The rehabilitation should be based on realistic goals and felt needs of the child and family.
- ▶ Inclusion of community-based organisations providing outreach efforts and greater social support can help connect families from low socioeconomic background and hence, improve parental resources.
- ▶ A large-scale educational effort to provide the necessary information about medical issues typically faced by children and young adults with spina bifida should be provided.
- ▶ Development of outreach clinics and programme providing care above and beyond what is delivered in an institutional set-up should be planned, especially in developing countries. Newer alternative approaches such as mobile health and telerehabilitation can be tried to enhance rehabilitation care to undeserved patients.⁸

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