Myelomeningocele, Chiari & Co

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Abstract

Object: Before 1960 the survival rate of neural tube defects (NTD) was barely 10%. Due to continuous improvements in treatment regime, the 1-year survival rate has been raised to 87%. Now a days about 78% of the patients with spina bifida irrespective of its severity reach the age of 17. Improved understanding of the pathophysiology of the malformation syndrome and its associated comorbidities (“double hit theory”) finally led to changes in diagnostic and therapeutic procedures (center-based pre- and postnatally surgery), and consequently to a higher life expectancy and better quality of life.

Methods: In order to assess Myelomeningocele (MMC) associated comorbidities we analyzed retrospectively 49 newborns with MMC, who had been surgically treated in our institution between January 2007 and January 2016. Children, who were previously operated in utero were excluded.

Results: The 49 children had a nearly equal gender distribution. 18% went undetected during pregnancy. The MMC was repaired on the day of delivery in 90%, and there were no initial infection of the cerebrospinal fluid (CSF) observed. The follow-up period range from 5 to 8 months 11/12 years without death over the entire period. 88% of the patients suffered from hydrocephalus (HC) necessitating shunt insertion, while shunt revision was required in 33 patients (67%). Seven times the closure of MMC and the implantation of the ventriculoperitoneal shunt were carried out sequentially during the same anesthesia with no further complications. In 37 children we found significant Chiari malformation type II (CMII), syringomyelia was seen in 24 children. Additional typical magnetic resonance imaging (MRI) findings: beaked tectum (n=36), enlarged mass a intermedia (n=37), dysplasia of the corpus callosum (n=33), polymicrogyria/stenogyria (n=20), and heterotopias (n=9). Based on the results of neuro-urological diagnostics, early clear intermittent catheterization (CIC) was the most common management (85.7%). A Gardner decompression was necessary in two, myelolysis due to secondary tethered cord syndrome (sTCS) in five children.

Conclusion: Chiari II malformation and hydrocephalus are the most common diseases associated with MMC. Nevertheless we found numerous CMII features in our patients, mostly without clinical relevance. In order to prevent or delay renal failure, which is known to be a significant complication in the course of the life of MMC patients, we highly recommend continuous close attention to the diagnostics and treatment of neurogenic bladder disorder. In our own series of postnatally repaired myelomeningoceles no death occurred, the rate of foramen magnum decompression and myelolysis due to sTCS was low, and the rate of shunted HC is consistent with the literature. The rationale of prenatal MMC repair is the prevention of second hit conditions. Taking the low rate of secondary comorbidities of our series into consideration, the advantages of any prenatal management need to be discussed controversially.

Keywords: Neural tube defect; Hydrocephalus; Chiari malformation; Neurogenic bladder disorder

Introduction

Prior to 1960 survival rate was only 10% in patients with spina bifida aperta. This was in part due to poor medical care in general as well as to a variety of accompanying diseases. In previous centuries MMC was declared an untreatable disease “that was best left alone” [1-5]. Nevertheless throughout the centuries occasional attempts were made to treat spina bifida children. The treatment options consisted in injecting the sac with sclerosing solutions (iodine, potassium iodine or “Morton’s iodo-glycerine solution), applying taps and ligation and in compression or removal of the sac [5-7].
Due to continuous improvements in treatment, the 1-year survival rate has been raised to 87% and about 78% of the patients with spina bifida of any severity reach the age of 17 [2,8]. One of the surgical improvements was the introduction of multilayer closure of the spinal defect to avoid cerebrospinal fluid fistulas by Harvey Cushing [9]. Furthermore, in contrast to previous times MMC has to be considered a far more complex malformation syndrome as a simple incomplete neural tube closure during the period of primary neurulation, which ends in the 4th week of gestation with closure of the caudal neuropore. It is generally accepted that the first part of the “double hit” theory can be prevented in 70% of the cases by a consistent periconceptional intake of folic acid (400 µg/d, for recurrence prevention and in a high risk situation the ten-fold dose is recommended). The second “hit” takes place when the developing spinal cord lies open and is exposed to amniotic fluid and/or trauma during movements [10,4,11-13]. In MMC patients a number of partially life-threatening changes can be observed in other regions of the body. These include skeletal, gastrointestinal, pulmonary, craniofacial, cardiovascular and genitourinary anomalies [14]. In addition intellectual limitations are common in MMC patients, and shunted children with MMC have an average intelligence quotient of 80 [3,4].

The term “Arnold-Chiari-Spina bifida complex” (Table 1) was used for accompanying intracranial changes [15]. Better knowledge of associated comorbidities and improved diagnostic and therapeutic procedures should lead to a further increase in life expectancy as well as to an improved quality of life.

Methods

The objective of this study is to analyze and report a single institute experience with postnatal repair of myelomeningocele with special consideration of comorbidities. In this retrospective study baseline data of all newborns were used, who had undergone repair of myelomeningocele between January 2007 and January 2016. Children who had previously been operated on in utero were excluded. The magnetic resonance imaging (MRI) scans of 46 cases were evaluated for intracranial anomalies, the position of the tonsils in the cervical spinal canal and the presence of syringomyelia. The type of neural tube defect, its location and all associated orthopedic and urological comorbidities, including diagnostic and therapeutic features were recorded.

Results

Between January 2007 and January 2016, a series of 49 newborns with myelomeningocele were treated neurosurgically in our department. We found a nearly equal gender distribution (25 males, 24 females). In nine children the defect remained undetected during gestation. The follow-up period ranges from 5 to 8 months 11/12 years (loss on follow up in 2 cases). In 90% (44 newborns) repair of the MMC was performed within the first 24 hours of life. In 61.2% an open defect was found, in 36.7% a thin arachnoid membrane. In one case the information was missing. After repair; local spinal complications comprised in 14.3% a subcutaneous cerebrospinal fluid (CSF) collection, in 6.1% a CSF fistula, and in eight cases (16.3%) abnormal skin healing. No subcutaneous CSF collection or fistula occurred in the children who underwent simultaneous repair of MMC and shunt insertion. Early revision surgery was necessary in 6 patients (12.2%), 4 cases (one of them 3 months after initial surgery) due to disturbance of wound healing, twice for CSF fistula. Only one local infection was seen. The child with the largest myelomeningocele (lumbosacral, 10 cm × 12 cm) also suffering from bowel and bladder incontinence, was supplied with urinary stoma and colostoma, and is mobile with orthoses. The smallest MMC was located sacrally, it was 1 cm × 2 cm in size. This child can walk without orthosis, has no hydrocephalus and no CMII. Due to parental incompliance and refusal to catheterize, bladder wall thickening with diverticula and a marked progressive vesicourethral reflux with damage to the renal pelvis developed (Figure 1).

The cranial extension of the bony spine defect was in the thoracic spine in 3, in the upper lumbar spine in 12 and in the lower
Figure 3: MRI T2 sag.: tonsillar herniation, diastematomyelia (arrowhead), vertebral malformation, dysplastic spinal cord.

lumbar spine in 20 patients. 11 children had a sacral defect. 88% of the children suffered from hydrocephalus, which necessitated shunt insertion. The closure of MMC and the shunt insertion were carried out sequentially during the same anesthesia with no further complications in 7 children. In all cases a non adjustable valve with integrated gravitational unit was implanted. Shunt revision was required in 33 patients (67%). In these patients the revision rate within the first year was 91% (Table 2). In group one the average period until the first shunt revision was 23 days. Failure of the central catheter was responsible for revision in 58% (occlusion [n=12], dislocation/malposition [n=6], CSF fistula [n=1]). In 18% a change of the valve and central catheter were required, total system replacement was necessary (dislocation of the central catheter and increased flow resistance in the valve and peripheral catheter) in one patient. In 2 cases only the valve was replaced. Other causes for revision were found in 15% (Table 3). In 37 children typical CMII features were found, these were hindbrain and tonsillar herniation, beaked tectum, enlarged massa intermedia, dysplasia of the corpus callosum (Table 4, Figure 2). The tonsils descended below the 2nd cervical vertebra in 26 children, beaked tectum, diastematomyelia (Fig. 3).

In our series we found one case with a diastematomyelia and deformity of the vertebral body (Figure 3). In 6% a cardiac anomaly existed. In case no.1 a patent foramen ovale (PFO) and a mild pulmonary stenosis (PST) with heart murmurs were found, both disappeared spontaneously. In the second child a ventricular septal defect (VSD) with pulmonary valve atresia and atrial septal defect were diagnosed echocardiographically - a cyanotic heart defect known as a severe form of Tetralogy of Fallot [16]. The boy underwent cardiac surgery at the age of three weeks, he performs now relatively well on cardiac stress tests and is able to walk.

The third child is a term male infant with hypoplastic heart syndrome. As the postnatal neurosurgical repair of the MMC as well as shunt insertion were of primary interest the palliation of the heart defect had to be postponed. Therefore, bilateral pulmonary banding and ductal stenting ("Gießen Approach") were performed at the age of 2 month. At the age of 6 month modified Glenn palliation was implemented successfully. Waardenburg syndrome was diagnosed (n=1), malrotation (n=1), duplex kidney (n=1) and undescended testicles (3♂).

Based on the results of neuro-urological diagnostics, early intermittent catheterization was the most common management (85.7%). Prophylactic measures were anticholinergics alone (n=18), in combination with cranberries (n=14) as well as antibiotics (n=2). A circumcision was performed in 16 out of 25 male patients. One child required a bladder stoma due to persistent incontinence, cystolithiasis and recurrent cystitis. No bladder augmentation was required. In summary a Gardner decompression was necessary in 2 children, myelolysis in 5 (one child underwent decompression of the foramen magnum and myelolysis and suffered additionally of diastematomyelia). Orthopedic surgery for scoliosis or kyphosis was performed in 4 children and for hip deformity in 3 children. Relevant deprivations of the feet were found in 26 patients necessitating surgical treatment in 8 cases. Only in one child an extension of the quadriceps tendon was required. In respect to mental development and behavior 10 children had documented limitations, particularly in language development, while above-average intelligence was seen in 4 children.

Discussion

Despite the introduction of folic acid supplementation and improvement of prenatal diagnosis, the disease myelomeningocele has not disappeared, and worldwide neural tube defects occur in 0.5–2 per 1000 pregnancies [17]. Myelomeningocele is not only a localized spinal problem. A number of associated cerebral abnormalities and congenital comorbidities with varying impact on development and survival rate are encountered. Chiari malformation type II, hydrocephalus and neurogenic bladder disorders (NBD) are the best known disorders associated with MMC. In a large retrospective series (502 patients with spinal dysraphism: MMC, caudal regression syndrome and spinal lipomas) neurogenic bladder was found in 97% of MMC cases and renal function impairment in 6.7% [18]. The latter was more often seen in patients with vesicouretal reflux and ureter widening. In our cohort 85.7% suffered from significant residual urine volume and underwent clean intermittent catheterization (CIC) with or without further prophylactic measures. This simple measure helped to avoid renal damage, an important life-limiting factor in MMC patients. Another major cause for morbidity in MMC patients is congenital heart disease in varying degrees, some requiring close attention to anticipate appropriate surgical intervention. The incidence specified ranges from 1.5 to 37% [19]. In their own series, the authors reported a rate of less than 3%, half as much as in our own series (6%). Hemodynamically irrelevant changes like small PFO and mild PST can disappear spontaneously, which is consistent with the natural course of physiological peripheral stenosis of the pulmonary artery [20]. A female predominance for cardiac disorders described in the literature this could not be confirmed in our series [19].

A number of publications focus on CMII, which occurs almost exclusively in connection with open spinal dysraphism, and its
typical features [21-24]. Callosal abnormalities were seen in the series from Miller et al. [23] in 57% of the cases, gray matter heterotopias in 19%. In 72% they found cortical posterior medial xsternagria, changes of the adhesion interthalamica in 48.6%. In our analysis we found xsternagria and/or polymicrogyria nearly half as often (42%), mostly in the occipital region. On the other hand anomalies of the corpus callosum (CC) with varying severity we saw more often (71%), but not as often as described by Hannay HJ et al. [25] (96%). Two mechanisms are postulated to cause callosal pathologies. These are either developmental or destructive mechanisms. In the latter hydrocephalus plays a role, which is thought to lead to callosal hypoplasia [25]. In our records in 20.4% of the children retardation in mental and language development as well as in behavior were mentioned, perhaps these developmental deficiencies were associated with an anomaly of the corpus callosum [26,27].

In up to a third CMII malformation will be symptomatic (pathological breathing, dysphagia, changing the voice pitch, opisthotonus, cranial nerve deficit) and must be treated [2,28]. As an additional diagnostic tool polysomnography is available demonstrating central and obstructive respiratory disorders. Before performing an upper cervical bone decompression hydrocephalus must be considered. In case of an existing shunt a dysfunction must be corrected. The most frequent secondary neurosurgical intervention in CMII patients was shunt revision (76%) [21]. As risk factors for sudden death in young adults with NTD female gender, sleep apnea and midbrain length ≥15 mm (MRI) were identified in a retrospective series with 106 patients [29].

The shunt dependence in spina bifida patients ranges in the literature from 49.5 (prenatal surgery) to 91% (postnatal repair of MMC). It has a higher complication rate compared to other conditions requiring shunt insertion and represents a lifelong risk factor [2,30-33].

Neurological deterioration is observed in cases of secondary tethered cord syndrome, which should be treated by myelolysis. It was seen in 10% in our series. In a retrospective study (35 children) Khoshhal et al. [34] found in 22.8% a sTCS. Neuro-orthopedic conditions are not primarily life-threatening, but important for reaching social independence and mobility. These are responsible for secondary diseases like pressure ulcers, contractures and osteoporosis. A 38% incidence of scoliosis/kyphoscoliosis (109 cases out of total of 286 cases) was found in the series from Kumar et al. [35], these depended on the level of defect, increased with age and required surgical correction if progressive. Before corrective surgery re-tethering (one third of MMC patients) should be ruled out. If seen on MRI de-tethering should be first considered as it can lead to stabilization or even improvement of the scolioticdeformity [36,37].

Conclusion

MMC is a developmental non progressive disease, which needs lifelong follow-up due to the comorbidities and associated anomalies. Despite reducing the incidence of the disease in industrialized countries with folic acid supplementation and prenatal diagnostics with the possibility of abortion, it remains a significant disease with tremendous impact for the patient. Focus should be placed on improving surgical techniques, especially in fetal repair to avoid the “second hit”, a damage of the open lying spinal cord due to amnion fluid toxicity and so minimizing associated comorbidities (hydrocephalus, CMII malformation, sTCS), which can sometimes be life-threatening [38].

But both methods of fetal MMC repair (open, fetoscopic) have in addition to the positive effect (decline in shunt dependent hydrocephalus, hindbrain herniation, and benefit in motor function a number of severe complications (e.g. prematurity, amnion infection, pulmonary hypoplasia, oligohydramnios, infant respiratory distress syndrome, fetal death, and in open fetal repair maternal complications) [33,38-40]. In our series of postnatal repair of myelomeningocele, we had no severe complication, no death, a low rate of sTCS, a low rate of required decompression of the foramen magnum and no additional maternal risks. Accordingly the indication for prenatal management needs to be discussed controversially.

References

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