Original article

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Presacral masses in children: presentation, aetiology and risk of malignancy

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Abstract

Aim The risk of malignant changes in presacral tumours in children was investigated in relation to age at diagnosis, type of presentation and origin of the tumour.

Method A retrospective review was carried out in 17 patients surgically treated for congenital presacral masses over a 22-year period.

Results Constipation was the main symptom in 14 (82%) of 17 patients. The lesions were evident on digital examination in 14 patients. Mature teratoma ($n = 9$, 64%) was the most common lesion, including three malignancies. Currarino syndrome was diagnosed in 10 (71%) patients. Two unclassified variant HLXB9 gene mutations were found in five (29%) patients who underwent genetic testing.

Conclusion Congenital presacral tumours in children were mostly mature teratomas, either as sacrococcygeal teratomas or as part of the Currarino syndrome. The risk of malignancy in patients older than 1 year necessitates early surgical resection.

Keywords Presacral tumour, Currarino syndrome, teratoma, sacrococcygeal teratoma

Introduction

The presacral region is an area of embryologic fusion of the hindgut, proctodeum, neural elements and bone. It contains tissues derived from all germinal layers that can lead to a variety of neoplasms. These tumours are rare, with an estimated adult incidence of 1/40 000 admissions and 1.4–6.3 patients per year in a major referral centre [1,2]. Presacral tumours generally have been categorized into congenital and acquired. With a prevalence of 50–77%, congenital tumours account for the majority [2]. A classification of congenital tumours can be found in Table 1. In children, presacral masses are either sacrococcygeal teratomas (Altman types III and IV) or tumours as part of the Currarino syndrome, which is a rare syndrome presenting with a typical bony sacral defect, often in combination with a presacral mass with or without an anorectal malformation. The bony sacral defect consists of a partial agenesis of the sacrum typically involving vertebrae S2–S5, showing on X-ray the so-called scimitar sign [3]. The anorectal malformation is usually an anorectal stenosis, but other types of lesions such as atresia also occur [4]. Different types of tumours occur in this syndrome: most frequently teratoma, anterior meningocele, dermoid or duplication cyst, or a combination of these [5,6]. The syndrome has a clear familial association and mutations of the HLXB9 homeobox gene on chromosome 7q36 have been identified [4,7]. The exact risk of malignancy remains unclear and estimates differ for specific types of masses.

In this study, a series of presacral masses in children is reported with regard to the mode of presentation, origin and type of the tumour and incidence of malignancy.

Method

From January 1987 to January 2009, patients with a congenital presacral mass treated by a paediatric surgeon were included in the study. The medical records were reviewed to determine patient demographics, medical history, symptoms at presentation and physical findings. Radiological reports of X-rays, ultrasound and magnetic resonance imaging of the pelvic region were reviewed for

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Table 1 Classification of congenital presacral tumours.

Benign	Malignant
Mature teratoma	Chordoma
Anterior meningocele	Malignant teratoma
Developmental cysts	Yolk sac tumour
Epidermoid cyst	
Dermoid cyst (mature cystic teratoma)	
Tailgut cyst (retrorectal cystic hamartoma)	

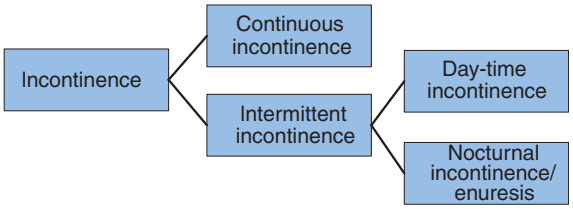

Figure 2 Subdivision of urinary incontinence in children (Adapted from Neveus *et al.* [9]).

a presacral mass and any sacral bony defect. Genetic testing reports were checked for the presence of the HLXB9 mutation. Follow-up was based on outpatient clinical records. Outcome measures included symptoms at presentation, histopathology, survival and surgical outcomes. Gastrointestinal, urological and neurological functions were assessed. Postoperative bowel motility function was determined using the three categories of the Krickenbeck conference classification (Fig. 1) [8]. Urological function at outpatient follow-up was categorized as continent, intermittently incontinent or continuously incontinent, according to the Report from the Standardisation Committee of the International Children's Continence Society (Fig. 2) [9]. Patients were contacted and interviewed in cases where data were missing.

Data are presented as median values with ranges, unless otherwise specified. Categorical data are presented as frequencies or percentages. Statistical analysis was carried out using the SPSS v.16.0.2.1 package (SPSS, Chicago, Illinois, USA).

Results

There were 17 (three men) patients included in the study. Patient characteristics, symptoms and histological diagnoses are presented in Table 2. Age at presentation ranged from newborn to 19 years (median age 0 weeks). One patient (6%) was already known to have Currarino syndrome during childhood because of family screening, but she presented with pain at the age of 19. The main symptom at presentation was constipation (14/17) varying from moderate constipation to neonatal bowel obstruction because of anal stenosis or anal atresia. In 14 (83%) patients, a mass or anorectal malformation was evident on rectal digital examination. Eleven (65%) patients were suspected to have Currarino syndrome based on clinical findings, of whom 10 had a sacral defect and 1 had a lumbar spine defect. An anorectal malformation was present in eight of those patients. Three of those had had a tethered cord at delivery and five had the mutation or deletion of the HLXB9 gene on chromosome 7q36, typical of Currarino syndrome. Two patients had an unclassified mutation and in three, a mutation could not be located.

1. Voluntary bowel movements	Yes/no
Feeling of urge	
Capacity to verbalize	
Hold the bowel movement	
2. Soiling	Yes/no
Grade 1 Occasionally (once or twice per week)	
Grade 2 Every day, no social problem	
Grade 3 Constant, social problem	
3. Constipation	Yes/no
Grade 1 Manageable by changes in diet	
Grade 2 Requires laxatives	
Grade 3 Resistant to laxatives and diet	

Figure 1 International classification (Krickenbeck) of outcome (Adapted from Holschneider *et al.*[8]).

Three malignant tumours were diagnosed, all in patients over 1 year of age. These included one patient with a metastasizing yolk sac tumour treated by several debulking procedures and brachytherapy. The second patient had a mixed malignant germ cell tumour containing yolk sac tumour and malignant teratoma treated by adjuvant chemotherapy. The third patient was diagnosed with a chordoma for which she underwent adjuvant proton therapy. In two other patients with a non-malignant tumour, the lesion recurred after incomplete resection.

Surgery

All 17 patients underwent surgery, in which the presacral mass was extirpated, explored or debulked. A posterior sagittal anorectoplasty (PSARP) with subsequent anal dilatations was performed in seven (41%) patients. Two (12%) underwent a procedure to untether the spinal cord. Several underwent more than one procedure.

Follow-up

The minimal period of follow-up after surgery was 44 days, with a median of 33 (1,5-216) months. One patient (#12) died at the age of 4 years because of a metastasizing yolk sac tumour 5 months after the last debulking procedure.

Table 2 Clinical data in 17 patients aged 0 weeks–19 years.

Case	Sex	Age at first presentation	Histopathology presacral mass	Symptoms	Sacral defect	ARM	Tethered cord	Genetics
1	F	Prenatal	Mature cystic teratoma, noninvasive	Prenatal screening ultrasound	–	None	–	Not assessed
2	M	Prenatal	Mature teratoma, non radical resection	Prenatal screening ultrasound	–	None	–	Not assessed
3	F	0 weeks	Lipoma	Constipation	–	Anal stenosis	–	No VATER mutation no 22q11 deletion
4	F	0 weeks	Mature cystic teratoma, non invasive	Urological problems, constipation	+	Anal stenosis	+	7q36 deletion not located
5	F	0 weeks	Mature teratoma, non invasive	Urological problems, constipation	+	Anal stenosis	–	Not assessed
6	M	0 weeks	Mature teratoma, noninvasive	Neonatal bowel obstruction	+	Anal stenosis	–	Mosaic trisomy chromosome 13
7	F	0 weeks	Benign teratoid tumour & anterior meningocele	Constipation	+	Anal atresia, RP fistula	–	Unclassified variant mutation HLXB9 gene
8	F	0 weeks	Mature cystic teratoma & intradural teratoma	Constipation	+	Anal stenosis	–	Not assessed
9	F	0 weeks	Ruptured dermoid cyst, no malignancy, nonradical resection	Constipation, sacral pain	–	None	–	Not assessed
10	F	3 weeks	Mature teratoma, adherent to spinal cord	Constipation	+	None	+	No HLXB9 mutation
11	M	8 months	Lipoma	Constipation	+	Anal stenosis, RP fistula	+	No HLXB9 mutation
12	F	20 months	Yolk sac tumour, invasive, non radical resection	Retarded motorical development, sacral dimple, urinal retention, constipation	+	Unknown	–	Not assessed
13	F	21 months	Mixed malignant germ cell tumour (YST in malignant teratoma)	Abdominal & sacral pain, swelling and haematoma, constipation	–	None	–	Not assessed
14	F	3 years	Mature teratoma, non radical resection	Sacral pain, swelling and haematoma, sacral dimple	–	None	–	Not assessed
15*	F	4 years	Hamartoma, non invasive	Constipation	+	Anal stenosis	–	Unclassified variant mutation HLXB9 gene
16	F	4 years	Chordoma, invasive, non radical resection	Constipation, sacral pain, swelling & haematoma	–	None	–	Not assessed
17*	F	19 years	Mature teratoma & anterior meningocele	Constipation, dysmenorrhea, dyspareunia, sacral pain	+	None	–	Not assessed

ARM, anorectal malformation; RP fistula, rectoperineal fistula; YST, yolk sac tumour. *Patients 15 and 17 are first cousins.

Details of postoperative bowel function were available for the other 16 patients. Voluntary bowel movement was present in 9 of 15 patients over the age of two. Soiling was present in 4 and constipation continued in 10 of 16 patients. The latter included seven with grade 2 and three with grade 3 constipation. Five patients over the age of two were completely continent. Urological dysfunction after the age of five was present in 2 of 11 patients. Both suffered from continuous incontinence of urine. A neuropathic bladder was present in 3 of 5 patients under the age of five. Neurological symptoms were present in one patient who had a gait disturbance caused by a tethered cord despite surgical untethering.

Discussion

This series of congenital presacral masses in children demonstrates differences in the form of presentation, origin of the mass and the presence of malignancy. Constipation has been reported to be main symptom of presacral tumours [6], and this is confirmed by our series. In a large series of presacral tumours, Jao *et al.* [10] found that the tumour was palpable on rectal examination in 97% of patients; in our series, this was 84%. This emphasizes the importance of digital examination in patients presenting with constipation. The female predominance is consistent with different series of patients with presacral tumours, including sacrococcygeal teratoma and Currarino syndrome [2,6,11,12].

In patients with a malignant tumour, symptoms started after the age of 1 year. In a study in the Netherlands over the period of 1970–2003, Derikx *et al.* showed that in all types of sacrococcygeal teratomas, the risk of malignancy increased with age. At birth, the risk was 1.5 per cent, but sacrococcygeal teratomas discovered after the age of 1 year had a risk up to 40% of being malignant [13]. Mature teratoma is a benign well-differentiated tumour with a minimal risk of malignant degeneration; nevertheless, some studies suggest that the risk of malignancy accompanying a mature teratoma increases with time and thus with age [13–15]. Although this theory is still a matter for discussion, treatment, i.e. surgical exploration, should be carried out as early as convenient. Studies showed that removal of the coccyx is essential in minimizing the chance of tumour recurrence. This is relevant, as the chance of malignant degeneration of a teratoma increases when the tumour returns after a nonradical resection [16]. Malignancy in a tumour as part of the Currarino syndrome has so far been reported in three children and four adults [4,17–20]. The age of diagnosis in Currarino syndrome ranged from 1 day to 89 years. Theoretically, an adult Currarino patient with a partial phenotype may present with a presacral mass and a history of constipation, but it should be kept in mind that the diagnosis was made before the age of 12 in 81% of patients with this rare syndrome [12]. In adult patients, a presacral tumour was seldom part of a syndrome and was most often a benign developmental cyst, although malignancies do occur [1].

Different theories exist regarding the aetiology of the different types of presacral tumours. Currarino *et al.* suggested a model of the 'split notochord syndrome', in which persistent abnormal adhesions between the endoderm (future gut) and the neural ectoderm occurred before the appearance of the notochord. This caused the notochord to split or to be displaced, preventing anterior fusion of the vertebrae. A fistula could form between the gut and spinal canal, giving possible rise to a meningocele or enteric cyst. The presacral teratoma would be formed by local enteric and neuroectodermal elements combined with mesodermal elements [3]. More recently, Gegg *et al.* proposed a theory in which the pluripotent caudal eminence, precursor of all caudal embryo tissues, is affected. By errors in secondary neurolation or migration, it would incorporate adjacent endoderm, which not only results in a trilaminar teratomatous mass but also affects the caudal spinal cord, anorectal complex, genitourinary tract and neural crest derivatives, giving rise to Currarino syndrome features [4,21]. The origin of sacrococcygeal teratomas is widely believed to be the consequence of aberrant patterns of migration from primordial germ cells; the exact oncogenetic mechanisms are still being studied [22].

In conclusion, in this series of congenital presacral tumours, most of the children presented with constipation. Most tumours were mature teratomas: either sacrococcygeal teratomas or tumours as part of the Currarino syndrome. There is a risk of malignancy in patients older than 1 year, which makes early surgical resection obligatory.

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