Case Report

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Difficulties in prenatal diagnosis of tumour in the fetal sacrococcygeal area

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Abstract

Prenatal ultrasound at the 20th week of gestation revealed a 3-cm tumour in the sacrococcygeal area. Initially, a sacrococcygeal teratoma was suspected on the basis of fetal ultrasonography, which revealed normal heart anatomy and an increasing tumour mass. The diagnosis was then changed to fetus in fetu or teratoma. Prenatal magnetic resonance imaging at the 34th week of pregnancy confirmed the ultrasound diagnosis. No other anomalies were found. Elective caesarean section was performed at term. The care team included a paediatric surgeon, obstetricians, neonatologists, midwives, and an anesthesiologist. A female newborn was delivered in good condition. The tumour was resected in the operating room and mature teratoma was established by histopathological evaluation. Surprisingly, agenesis of the right forearm was revealed which had not been detected prenatally, despite many examinations (both in our hospital and earlier at a primary care obstetrician office).

Keywords

Fetus in fetu, teratoma, meningocele, forearm aplasia

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Background

Tumours of the sacrococcygeal area belong to rare retroperitoneal malformations diagnosed during intrauterine life. At present it is believed that the prevalence of these malformations fluctuates from 1:500,000 births with fetus in fetu (FIF) to 1:20,000 births with sacrococcygeal teratoma, the latter being to the most common tumours in newborns. The issue referring to the difficulties in the differential diagnosis has been rarely raised in recent literature.

Case report

A multiparous woman in her third pregnancy underwent an ultrasound scan at 12 weeks of gestation. The fetal nuchal translucency (NT) was found to be 1.7 mm. She underwent an amniocentesis following a combined

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Down screening test, which produced a risk estimate of 1 in 33. During this scan a cyst measuring 23×22 mm with an internal echogenic mass (15×14 mm) was detected. At the 20th week, an increase of the mass or tumour size to 32×28 mm was observed and the pregnant woman was referred to our tertiary centre with the suspicion of sacrococcygeal teratoma.

During the first US examination in our center, at the 24th week, the lesion was described as a mixed solid and cystic structure of 4.4 cm diameter (Figure 1) and the presence of a sacrococcygeal teratoma was confirmed. The fetal heart showed normal anatomy, and the echocardiographic functional assessment was normal. The cardiovascular profile score (CVPS) was 10 (normal). Three weeks later, at the 27th week of pregnancy, there was progression of the tumour size. Its diameter was over 7 cm and the appearance was atypical, resembling brain tissue (Figure 2). The cardiovascular profile, however, was normal. The differential diagnosis based on US findings and normal echocardiographic examination suggested FIF or teratoma, but probably not sacrococcygeal teratoma.

At 31 weeks and 6 days of gestation, the tumour size was 80×90 mm, with an amniotic fluid index of 15 cm (normal). The heart function was again assessed as normal on echocardiographic detailed examination. At 33 weeks of gestation, a fetal MRI showed the tumor size to be $120 \times 100 \times 85$ mm with two fluid spaces (36 mm and 54 mm). The differential MRI diagnosis was similar to that of the ultrasound: atypical teratoma, or due to existing fatty tissue, FIF.

Due to an increased tumor mass during gestation, our fetal surgeon made an attempt to directly approach the surface of the fetal tumor. He had hoped to obliterate at least some of the vessels, and decrease the size of the tumor. However, during fetoscopy it turned out that the tumor was covered by skin, and no major vessel was seen during the procedure. Longitudinal echocardiographic monitoring of the fetal heart in the prenatal cardiology department had (since the 24th week of pregnancy) always confirmed good circulatory function and normal fetal biometry (Figure 3). The course of diagnostics in the prenatal period in our institution is presented in Table 1 and the differential diagnostics in Table 2.

Delivery by caesarean section was planned at term, at the 38th week of gestation, involving a multidisciplinary team of anesthesiologists, neonatologists, obstetricians, and pediatric surgeons. The newborn was delivered in good general condition, with a birth weight of 3120 g and Apgar scores of 9 each at 1, 5, and 10 minutes. The tumour, weighing 430 g ($15 \times 10 \times 8$ cm), was resected immediately after birth (Figures 4 and 5). After resection the baby weighed 2690 g.

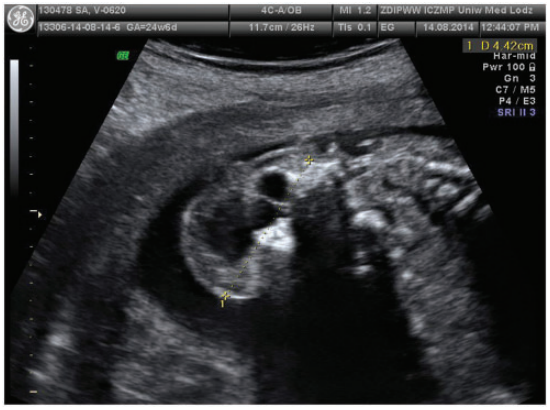

Figure 1. Ultrasound scan at 24th week of pregnancy showing a tumour in the fetal sacral region measuring 4.4 cm.

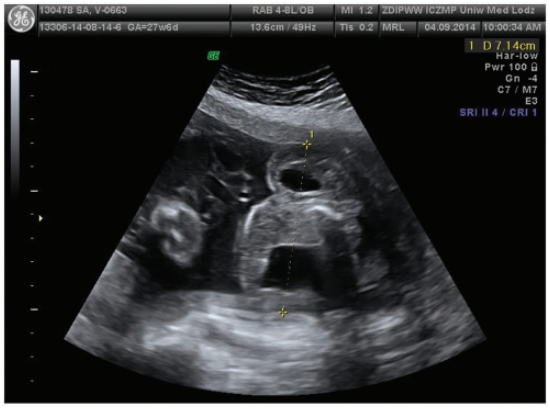

Figure 2. Ultrasound scan at 27th week of gestation showing a tumour diameter over 7 cm and tumour mass resembling brain tissue.

The tumour was partially covered with hairy skin and was solid and cystic. There was a bone in the central part of the tumour. Histopathological examination revealed a mature cyst (teratoma maturum) with the presence of all three germ layers. Hypoplasia of the right forearm was found and the right hand was missing (Figure 6).

The newborn was discharged, and allowed to go home in good clinical condition without complications after 16 days of hospitalization with the recommendation of further highly specialized care, including surgical and orthopedic rehabilitation.

Discussion

Teratomas are neoplasms consisting of the tissues of all three germ layers and may contain hair, teeth, bones, tissues, and organs. They are the most frequent types

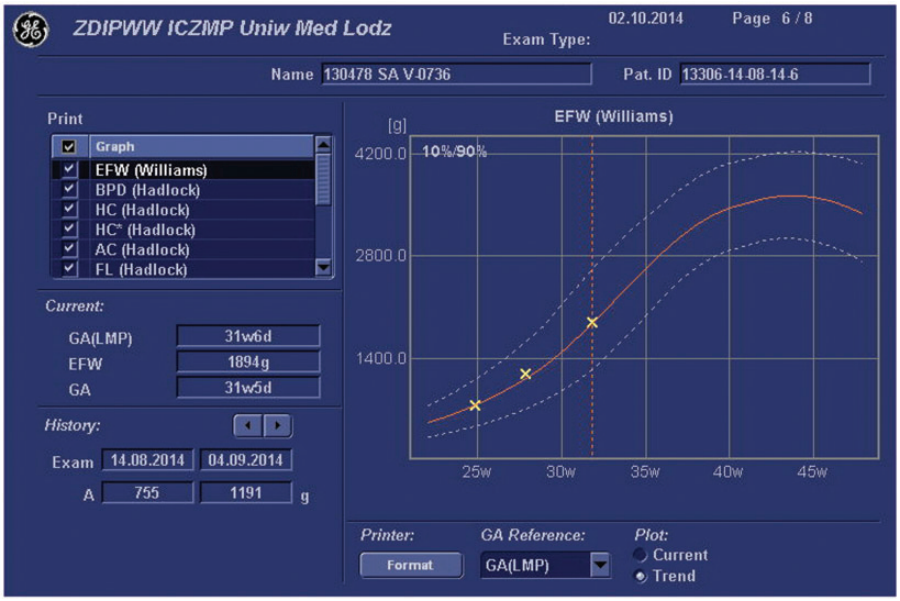

Figure 3. Normal fetal biometry from 25th week of gestation.

Table 1. Prenatal findings in the presented case

Week of pregnancy	15	20	24	27	31	34 (MRI)	In vivo
Tumour size (mm)	23 × 22	32 × 28	44 × 31	72 × 50 × 43	80 × 90	120 × 100 × 85	150 × 100 × 80
Fetus mass (g)	48	284	510	820	1510	2370	2690
AFI (cm)[a]	N[d]	N	16	15	14	14	–
Heart size (mm)	–	–	23	27	32	–	–
TeiRV[b]	–	–	0.35	0.35	0.36	–	–
Tei LV[b]	–	–	0.36	0.36	0.38	–	–
CVPS[c]	–	–	10	10	10	–	–

[a]AFI—amniotic fluid index—to determine the AFI we used a four-quadrant technique.
[b]Tei ratios—It is an index that incorporates both systolic and diastolic time intervals in expressing global systolic and diastolic ventricular function (RV—right ventricle, LV—left ventricle)—norm 0.39 +/− 0.05 (calculated by Tei Index = a−b/b = (IVCT + IVRT)/ET) IVCT—isovolumic contraction time, IVRT—isovolumic relaxation time, ET—ejection time.
[c]CVPS—cardiovascular profile score—from 1 to 10.
[d]N—normal.

of brain neoplasm in infants and may be potentially malignant (in about 12%).[1,2] The prevalence of sacrococcygeal teratoma is about 1:20,000 births and it is the most common tumour found in newborns. The prevalence of fetus in fetu (FIF) is 1:500,000 births. In the past, FIF was considered as a highly differentiated form of mature teratoma and now it is accepted as an abnormal embryonal development. Recent literature questions the previously binding definition of FIF, denying the necessity of the existence of axial skeleton (definition initiated by Willis in 1962) or individual bone parts as it has been thus far, until the establishment of the final and reliable diagnosis.[1,3] While there are several theories regarding the development of FIF, it is possible that FIF develops due to abnormal embryogenesis in a diamniotic monochorionic twin pregnancy as a result of the improper division of blastocystis.[4]

FIF and sacrococcygeal teratoma belong to rare retroperitoneal malformations diagnosed in

Table 2. Differential diagnosis

	Sacrococcygeal teratoma	Fetus in fetu	Myelomeningoceole
Differentiated tissues coming from three germs layers	+	+	–
Presence of organs, big parts, e.g. limbs, bones	+/–	+/–	–
Vertebrae and maintained bilateral symmetry	–	+/–	–
Vascularisation	Rich in solid tumours	Small (a few relatively big connecting vessels)	–
Growth	Fast	Slow	Slow
Cardiovascular efficiency	Possibility of decompensation	Maintained	Maintained

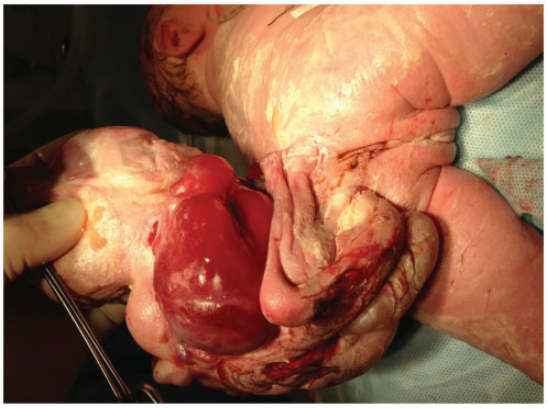

Figure 4. Immediately after delivery.

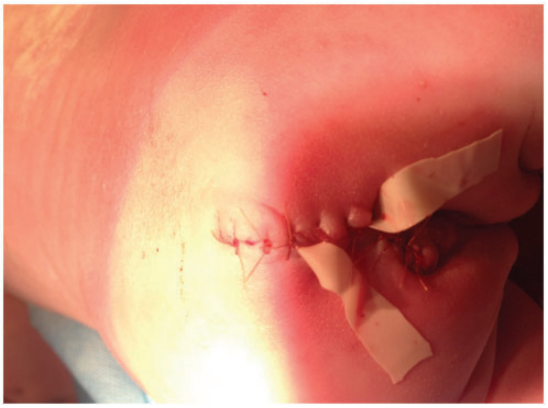

Figure 5. Neonatal surgery immediately after delivery.

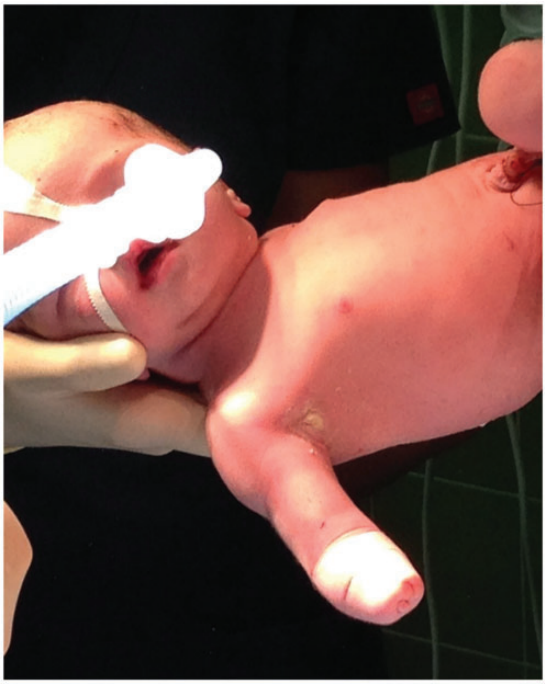

Figure 6. Right forearm aplasia and absence of hand (not detected before birth).

intrauterine life and they might be very similar at the early stage of pregnancy. The problem of difficulties in the differential diagnosis has been rarely raised in the recent literature and our case is the first to present the value of longitudinal assessment, including fetal echocardiography monitoring. According to the currently

developing hypothesis, at least one of the following conditions has to be fulfilled in order to diagnose FIF: 1. Content of mass in a separate sac; 2. Partially or fully covered with skin; 3. Possesses anatomical parts easily diagnosed; 4. Connected with the host through a few relatively big venous vessels; 5. Connected by the neural tube or alimentary tract.[3]

It should be noted that teratoma develops from non-physiological differentiation of the sex cell in the direction of all three germ layers and therefore its structure can be cystic, solid as well as mixed. In about 15% of patients, teratomas may be accompanied by congenital diseases such as imperforate anus, bicornuate vagina, spina bifida, or other malformations of the spine. Teratomas may grow at a very rapid rate and especially solid tumours may be highly vascular. Consequently, this may lead to heart failure, bleeding to the tumour, fetal anaemia, and finally to nonimmune hydrops fetalis.[5]

Location-based classification according to the Surgical Section of the American Academy of Paediatrics is:

Type I: 50% developing only outside the fetus (external location);
Type II: in 50%, extra-fetal with intra-pelvic presacral extension (internal location);
Type III: extra-fetal with abdomino-pelvic extension (mainly internal location);
Type IV: tumour developing in the fetal pelvis (completely internal location).

Another type of prognostic classification is prenatally limited, but initially the fetuses can be divided into three groups. Group A consists of tumours <10 cm. Group B tumours are the highest risk, and their diameter exceeds 10 cm. These tumours also are highly vascularized and have a rapid growth rate. Group C tumours are less than 10 cm with cyst structure; they have poor or no vascularization and slow growth.[6]

At present, according to the Spencer's theory, it is said that FIF and different teratomas are the result of abnormal monozygotic twin pregnancy. His hypothesis indicates that the spectrum of these anomalies extends between conjoined twins, acardiac twin to FIF and from the so called "fetaform" teratoma to well differentiated teratomas. Continuing Spencer's hypothesis—he believes that there is an association between FIF and teratoma because both forms are more prevalent in families with a history of twinning.[3] Furthermore, the possibility of coexistence of FIF and teratoma has been confirmed (our case and presented differential diagnosis also confirm this hypothesis). However, both these entities are similar in regards to radiological findings, but the lack of visibility of an axial spine in CT or X-ray does not exclude the existence of FIF.[4]

Attention should be paid to early diagnosis of the tumour in the fetus because then, the management of this condition will have greatest significance for both the fetus and the pregnant woman. Each case should be considered individually in regard to the occurrence of possible accompanying malformations and to the consequences of the presence and growth of the tumour. Currently, it is believed that fetal tumours are rarely accompanied by anomalies of the fetal karyotype. Therefore, in cases such as these, routine amniocentesis is not recommended as it additionally endangers the fetus with complications.[7] In our case the fetal karyotype was normal. Is it possible that agenesis of the fetal right hand was caused by fetal needle injury during diagnostic amniocentesis? There is, however, insufficient evidence of this in the medical literature. In cases with an increase of the symptoms associated with the tumour, intrauterine therapy has been suggested in order to remove part of the lesion or its complete resection or coagulation of the main providing vessel.[8] In most cases prognosis in FIF as well as in teratoma is very good and after the removal of the lesion no local recurrence has been observed. Myelomeningocele should also be taken into consideration in the differential diagnosis of the tumour in the sacrococcygeal area. In the presented case, the continuity of the spine and the image of the central nervous system did not raise any doubts.

The failure to detect a malformation of the fetal upper limb in prenatal imagining should be emphasized. This fact was overlooked both in the first trimester of the pregnancy, in which it was the easiest to assess the skeletal system, as well as in further US examinations and fetal MRI. This proves how difficult it may be to recognize diagnostically the anomalies of the fetal palms and forearms. To compare, having obtained the parents' consent, we present the photograph of the 3-month-old girl with her elder sister (Figure 7).

The ultrasonography and echocardiographic examination in the presented case did not only help to establish the final diagnosis but primarily to monitor the tumour growth and the effects of its growth on the fetus and, in particular, on the circulation system. The fetus was active to the end of the pregnancy, all endocardiac flows were normal, good development of the fetal lungs were registered and there was no need for premature delivery despite the presence of a large tumour qualified for surgical intervention after birth. Echocardiography confirmed well-being of the fetus and provided additional safety for the obstetrician and the pregnant woman.

Figure 7. The 3-month-old girl with her elder sister.

Particular attention should be paid that one defect does not preclude the coexistence of others—such as the lack of a forearm in this case. Also we need to know that lack of a spine or its elements in the picture of the ultrasound or MRI do not exclude the diagnosis of disease.

Declaration of conflicting interests

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Ethical approval

Local approval, project no. 1. Written consent was obtained from the patient to publish the images and medical details of the case.

Guarantor

Katarzyna Zych-Krekora

Contributorship

MK and KZK researched the literature and conceived the study. MS, MRL and MB designed the audit. KSz, SŁ and APP did the data analysis. KZK wrote the first draft of the manuscript. MK wrote the final version of the manuscript. All authors reviewed and approved the final version of the manuscript.

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