CASE REPORT

Malignant rhabdoid tumour of the neck in a neonate

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SUMMARY

Fetal neck masses are rare, and present a challenge antenatally, during delivery and in making a diagnosis. In this report, we detail the presentation of a neonate with a neck mass diagnosed in the third trimester. The baby was delivered by ex utero intrapartum therapy (EXIT). Investigations sent included an MRI with limited CT cuts, and a biopsy, which lead to the diagnosis of a malignant rhabdoid tumour. This is rare and the overall survival is low.

BACKGROUND

Neck masses in the fetus are rare and present a challenge at various steps. Differentials vary based on the site of the tumour and the consistency—solid or cystic. Non-renal, non-central nervous system (CNS) rhabdoid tumours are rare in neonates.

CASE PRESENTATION

A baby was transferred in utero from a secondary hospital in view of an antenatal ultrasound done at 36 weeks of gestation which showed polyhydramnios (amniotic fluid index: 44 cm, single largest pocket: 14 cm) and a mass lesion 7 cm × 6 cm over the left side of the neck. This was the first baby of a 27-year-old non-consanguineously married mother. As seen in the antenatal ultrasound, the mass was predominantly solid with cystic spaces and internal septations, with increased vascularity. The trachea could not be visualised. Since the mother did not have an early dating scan, and with the baby's expected weight being 1800 g (with the tumour), the mother was given two doses of antenatal steroids. As oesophageal and tracheal obstruction was anticipated, the baby was delivered by ex utero intrapartum therapy (EXIT) under general anaesthesia.¹ An airway was established in 7 min under direct laryngoscopy with a 3 size endotracheal tube after an assistant lifted the tumour away from the field. The baby's heart rate remained above 60 beats per min throughout the procedure. Following the establishment of airway, the baby was transported to the neonatal unit using T-piece resuscitator with a peak inspiratory pressure of 20 cm H₂O and positive end-expiratory pressure of 5 cm H₂O. The baby had a neck mass of 10 × 7 cm on the left side of the face extending to neck. She was also noticed to have a nodule in the right popliteal fossa. The baby was sedated and ventilated during the duration of admission.

INVESTIGATIONS

Initial investigations at 1 hour of life showed metabolic acidosis with elevated lactate levels. Total

count was 34 600/mm³, with a leucoerythroblastic blood picture with elevated nucleated red blood cells (130/100 leucocytes). Liver enzymes were normal. Initial alpha fetoprotein was 58 000 IU/mL and Creatine Phosphokinase (CPK) was 392 IU/L. Screening echocardiography carried out on day 1 showed myocardial dysfunction with hypertrophic cardiomyopathy.

MRI of the neck (along with limited cuts of CT) was carried out at 24 hours of life, which showed a large lesion on the left side of the neck extending from the base of skull and temporal scalp region up to the clavicles (figures 1 and 2). There was associated rarefaction of the floor of middle cranial fossa and the left carotid sheath vessels were embedded within the tumour (figures 3 and 4). There was evidence of compression of the nasopharynx and oropharynx. A similar lesion was seen on the right side of the neck as well, deep to the right sternocleidomastoid muscle. Biopsy of the tumour and the nodule in the right popliteal fossa was carried out on day 2.

The biopsy showed cores of a cellular tumour composed of sheets of atypical cells with mild to moderately pleomorphic round to oval nuclei, irregular nuclear membrane, many with visible nucleoli and moderate amounts of eosinophilic to clear cytoplasm (figure 5). On immunohistochemistry, the tumour cells were negative for CD3, CD20, CD34, MPO, CD30, desmin and synaptophysin. Tumour cells were positive for cytokeratin (figure 6) with globular accentuation and focal staining for Epithelial Membrane Antigen (EMA). There was loss of Integrator Interactor 1 (INI-1) in the tumour cells (figure 7).

Over the course of admission, the baby had severe myocardial dysfunction necessitating three inotropes (dopamine, dobutamine and vasopressin). Milrinone was also commenced by day 2. She had low urine output from day 1 with biochemical evidence of acute kidney injury and was anuric from day 3. She also developed clinical and biochemical features of tumour lysis syndrome. The details of the tumour lysis syndrome and its management have been described separately.² She was started on treatment for tumour lysis syndrome from day 3 with hydration, allopurinol and rasburicase.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis of massive congenital neck masses that were considered for the baby were soft tissue sarcoma, undifferentiated teratoma and cervical neuroblastoma.

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Figure 1 T2-weighted image showing the extent of the tumour from the middle cranial fossa to the clavicles.

OUTCOME AND FOLLOW-UP

In view of the highly probable malignant nature of the tumour with tumour lysis syndrome and very guarded prognosis, the father made an informed decision not to pursue intensive care. Supports were withdrawn and the baby expired on day 4. Autopsy was advised but was not carried out since the parents did not consent.

DISCUSSION

Fetal neck masses are rare. The differential diagnoses considered is based on the anatomical location of the mass and other antenatal ultrasound findings depending on the solid or cystic nature of the tumour. In the anterior triangle, teratoma, goitre and haemangioma are the most common differentials considered for giant fetal neck masses. In the posterior triangle, cystic hygroma, cervical meningocele and occipital encephalocele are most common.³

After the diagnosis of a large neck mass, the decision was made to deliver the baby using the EXIT procedure. The use of EXIT during the delivery of a large congenital neck mass is well described.⁴ The size of the mass in conjunction with

Figure 3 CT cut showing rarefaction of the floor of middle cranial fossa.

polyhydramnios had suggested an increased risk of airway obstruction. This was proven in the MRI which showed compression of the nasopharynx and oropharynx. Antenatally, the differential diagnoses considered most likely were teratoma, neuroblastoma, cystic hygroma and haemangioma. Postnatally, in view of another lesion in popliteal fossa and the fact that the anomaly scan had not detected the tumour with rapid progression probably in the third trimester made the probability of a malignant tumour (neuroblastoma or a haematolymphoid malignancy) more likely. Based on the clinical examination and the MRI findings, other differentials considered were cervical neuroblastoma, soft tissue sarcoma and undifferentiated teratoma.

Initially described as a variant of Wilm's tumour, malignant rhabdoid tumours are extremely rare.

A review of the literature reported 72 fetal and neonatal rhabdoid tumours.⁵ Of these, 24 neonates and 9 fetuses were reported to have non-renal non-CNS tumours, and only 4 were identified as having a primary tumour in the neck similar to the case we report here. Described in neonates with early and multiple metastases and a rapid downhill course, the average gestational

Figure 2 MRI: T2-weighted image showing the extent of the tumour from the middle cranial fossa to the clavicles.

Figure 4 Short T1 inversion recovery images showing encasement of the carotid sheath within the tumour.

Figure 5 Large polygonal tumour cells with vesicular nuclei and prominent nucleoli and abundant amounts of eosinophilic cytoplasm.

age of neonates with non-renal, non-CNS rhabdoid tumours is 33 weeks, with a mean birth weight of 2696g. The average tumour weight is 382g (range: 120–980g). The most common location described is the skin and soft tissues as compared with the older infant with primaries in the kidney or CNS. Metastases are described at presentation in 70% of cases, with the most common site being the skin.

The characteristic finding on histology of the rhabdoid tumour is the rhabdoid tumour cell, which is described with a round vesicular nucleus, prominent nucleolus. On electron microscopy, a round to oval eosinophilic inclusion containing intermediate filaments is seen. Genetic analysis shows deletions in chromosome 22q11 and mutations of the *hSNF5/INI1* gene, which add to the diagnosis. In our patient, while blood was taken for DNA banking, genetic analysis had not been carried out at the time of reporting.

Overall survival is low (9.1%). Three survivors have been described, with localised skin or orbital rhabdoid tumours, which were treated with surgical resection and chemotherapy.

In our patient, the findings of the carotid sheath encased with tumour and the extension into the middle cranial fossa limited

Figure 6 Staining with cytokeratin (×400). Tumour cells with cytoplasmic positivity for cytokeratin with perinuclear accentuation were seen.

Figure 7 Staining with INI-1 (×400). Tumour cells display loss of INI-1.

Patient's perspective

Translated from the patient's father's language:

I was devastated to learn that my baby has a mass in the neck. I know that this is not common. I feel privileged to be considered to contribute to the larger body of knowledge by permitting my baby's story to be published, hoping that some other baby somewhere may benefit from this. I do understand my privacy will not be violated by this publication.

Learning points

- ▶ Ex utero intrapartum therapy has to be considered during the delivery of congenital neck masses.
- ▶ Differentials in a neonate with a giant neck mass are teratoma, goitre and haemangioma in the anterior triangle and cystic hygroma, cervical meningocele and occipital encephalocele in the posterior triangle.
- ▶ Rhabdoid tumours are to be considered when confronted with evidence of metastases and rapid progression.
- ▶ Genetic analysis is to be considered when attempting to make a diagnosis, for prognostication and management.

the surgical alternatives. The presence of tumour lysis syndrome together with the rapidly progressive uric acid nephropathy made it impossible to consider chemotherapy, leaving dialysis as the only option in view of the renal shut down.

In summary, despite the rarity of the occurrence, rhabdoid tumours have to be considered in the setting of a tumour in the neck with evidence of metastases and rapid progression in a neonate.

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