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CASE REPORT

A very unusual complication of amniocentesis

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Key Clinical Message

The uses of amniocentesis are numerous, including determination of chromosomal abnormalities, lung maturity, and infections. A common complication of amniocentesis is loss of the pregnancy, but rare complications should be considered. The role of patient history and clinical observation of uncommon presentations are critical in the management of the patient.

Keywords

Amniocentesis, complications, procedure, teratoma.

Amniocentesis is a well-known procedure performed during pregnancy for diagnostic and therapeutic purposes. Typical complications of the procedure include infection of the amniotic sac, preterm labor, respiratory distress, fetal deformities, trauma, alloimmunization, and failure of the puncture wound to heal properly. We present the case of a 13-month-old male who developed this primary tumor in the left hand thought to be a result of an amniocentesis complication during the second trimester of pregnancy. The diagnosis of this case proved difficult with multiple consultations due to the unconventional presentation of symptoms. The clinical impact of this case is evident as knowledge of this complication in fetal life can lead to a swifter and more accurate diagnosis as well as prevent further complications associated with malignant lesions.

Background

Amniocentesis is a routine procedure that is rarely the cause of a teratoma. Teratoma is a congenital tumor that is commonly formed by tissue represented by the three germ layers: the outer most layer ectoderm, the inner layer called the endoderm and mesoderm. The word teratoma has originated from the Greek language. The Greek word "*teraton*," refers to monster and was used by the German physician Rudolph C. Virchow in his book on tumors, which was published in 1863 [1]. These tumors are either monodermal and highly specialized or in rare cases undergo malignant transformation [2]. Typically, teratomas are classified as benign, heterogeneous collection of well-differentiated cells (mature), undifferentiated cells of embryonic origin (immature), teratoma with malignant transformation, and mixed germ cell tumors, which also includes malignant mixed type (also referred to as teratocarcinoma) [2]. The specific type of tumor and its location depends on the age of the patient.

Benign teratomas usually do not spread but rather they tend to grow aggressively. On the other hand, malignant teratomas are characterized by metastasis in addition to local aggressive growth [3]. Specifically; teratomas in newborns are generally benign and local in their growth behavior. Some teratomas can gain malignancy, "depending on the maturity and other types of cells that may be involved [3]."

The most common congenital tumor in a newborn is a sacrococcygeal teratoma, which has a highly favorable prognosis due to its early diagnosis (grossly visible), allowing for timely treatment [3]. Unique germ cell sites such as the ovary, testicle, and even some unusual sites as the neck, stomach, and portions of the retroperitoneum may be sites for teratomas [3].

Another common tumor in children is the yolk sac tumor, also known as endodermal sinus tumor (EST).

The EST is the most common testicular tumor in children under 3 years of age [4]. These tumors bear some histological resemblance to the yolk sac, allantois, and the embryonic mesenchyme, hence the nomenclature. Several studies have studied gene abnormalities associated with EST, one being hypermethylation of the RUNX3 gene (a transcription factor) and the second being associated with the overexpression of GATA-4, another transcription factor that regulates the differentiation of the yolk sac endoderm [5, 6]. Adult yolk sac tumors are often found in conjunction with other kinds of germ cell tumors, mostly teratomas, and embryonal carcinomas, however, infants usually present with the pure form of teratomas [7].

The presentation of these tumors usually consists of no specific set of symptoms except for a painless testicular mass. Pure teratomas are usually benign, while the EST is malignant. Metastasis of the tumor is uncommon and results in >10% of the affected population of patients. Elevated levels of α feta protein occur in 75% of patients with teratocarcinoma (mixed germ cell tumor), embryonal cell carcinoma, and yolk sac carcinoma [4, 7]. Aggressive tumors in children are commonly treated with surgical resection and adjuvant chemotherapy, which has excellent prognostic results. However, other factors such as lower levels of alpha fetal protein and earlier stages of disease also carry a better prognosis. The following case is an atypical presentation of a mixed teratoma (teratocarcinoma) with yolk sac component secondary to a procedure.

Case Presentation

We report a 13-month-old African American male who presented with a mass on the dorsal left hand. The mass was not noticed until recently as it grew rapidly in size. No report of redness or pain associated with the lesion. The patient had no significant past medical history according to his parents. A complete prenatal, natal, and postnatal history revealed a complicated amniocentesis during the second trimester in 2012. The amniocentesis was done in a private clinic setting and much detailed information was not available. After further inquiry, the mother explained that there was minor contact of the needle with the fetus but no trauma or abnormalities were noted at the time or after several follow-ups.

Investigations

A biopsy was performed of the lesion and a fleshy, yellow-tan soft tissue measuring $2.5 \times 2 \times 0.5$ cm was excised. The initial microscopic description of the soft tissue sections revealed that the tumor resembled Schiller–Duvall bodies. Further description of the mass included: pleomorphic malignant neoplasm with focal areas of the tumor that resembled differentiated and tubular structures and eosinophilic hyaline droplets present extracellularly within the cytoplasm that contained alpha fetoprotein. Chest X-ray (including skull), computed tomography (CT) scan, and abdominal ultrasound were unremarkable.

Differential diagnosis

The differential diagnoses considered in this case included synovial sarcoma, unusual sweat gland carcinoma, unusual appearing hemangiopericytoma, and a glomus tumor. Multiple consultations resulted in the final diagnosis to be mixed teratoma (teratocarcinoma) with yolk sac component.

Treatment

Based on the information at hand and the diagnosis it was determined to proceed with amputation of the left hand. A biopsy of the left axillary nodes was also performed at the time as well.

Outcome and follow-up

The patient recovered well postoperatively. The axillary lymph nodes yielded negative results for metastatic cells. The patient is now doing well and continuing to follow-up with his pediatrician.

Discussion

This case offered a patient with an extragonadal teratoma located in the upper extremity, inarguably an uncommon place. Extragonadal presentations of germ cell tumors are well recognized, occurring in the retroperitoneum, mediastinum, pineal, and suprasellar region [8]. There was no evidence of a primary source for this malignant tumor, normally discovered by lymph node biopsy and at long-term follow-up (greater than 10 years) there were no additional sites of tumor growth. A classic feature of malignant, germ cell tumors (of the gonads) is a supraclavicular mass also known as Virchow's Node due to lymphadenopathy via lymphatic drainage. Routine biopsy of supraclavicular nodes has indeed been advocated as a staging procedure for patients with testicular tumors [9, 10]. In this case there was no evidence of testicular tumor and after amputation of the effected hand the biopsy results were negative.

According to Colon and Upton, "diagnosing and treating pediatric hand tumors requires an understanding of unique features [11]." Most lesions of the upper extremities present with a definite history of trauma, but some,

as the case presented herein, do not. The palmar surface of the hand was the most common site of the lesions. It is widely accepted that hand tumors in pediatric patients are rare and when found, diagnostic tests such as, CT, magnetic resonance imaging, and ultrasonography can provide help to identify specific diagnoses [12–15]. However, most pediatric hand tumors are benign and painless and do not require surgical intervention [11].

Sobanko et al. stated that "soft tissue tumors of the hand arise from skin, subcutaneous tissue, tendons, nerve, and blood vessels [16]." Tumors of the hand usually present differently, therefore, these must be treated differently than similar tumors on other areas of the body. Only 1–2% of primary tumors located in the hand are considered to be malignant, additionally the most common of these malignant tumors are squamous cell carcinoma [16]. Metastatic tumors are rare in the hand, but they should be strongly considered in the differential as the prognosis is poor [10, 15]. Two-thirds of reported cases originate through malignancies of the lung, kidney, or head/neck, although not marked in this case [16].

The crucial element in this case was a thorough history and physical examination. Once the chart was reviewed and it was revealed that the patient had undergone a complicated amniocentesis procedure during the second trimester, the diagnosis became much more apparent. The amniotic fluid contains fetal tissues/cells, which was likely extracted from the amniotic sac and inadvertently placed into the patient's left hand.

Although amniocentesis is a routine procedure there are numerous complications that are associated with the procedure. Serious complications can result in miscarriage [17]. Amniocentesis carries a risk of miscarriage, which is thought to be 1 out of every 200 procedures, however, a recent study has shown that this risk may be reduced, 1 in 1600 [18, 19]. In comparison to chorionic villus sampling (CVS), the risk of miscarriage has reported to be more substantial, 1 out of 100 procedures. It is preferable to perform CVS up to four weeks earlier because the possibility of genetic defects is thought to be higher [20].

Although very uncommon, one of the risks mentioned is fetal injury, secondary to trauma from the needle. Fetal trauma case reports have commonly reported disfigurements on the skin; nonetheless, cases have been reported inciting direct trauma to central nervous system causing bleeds, ocular disturbances, and injury to internal organ [21]. In this clinical case, we believe that the fetus underwent trauma during the amniocentesis procedure, causing damage to the left hand of the fetus. Fetal limbs were found to be more commonly associated with needle contact compared to other fetal body parts [22]. Most amniocentesis-related injuries occur when the procedure is performed in the late 2nd and 3rd trimesters. Petrikovsky and Kaplan reported an incidence of 0.4% when using constant ultrasound-guided amniocentesis [22]. Prior to the use of synchronized ultrasound (U/S), a doctor's judgment was mainly based on the physical examination; it was performed blindly. Shortly after the restrictions of static U/S were uncovered, continuous visualization became the new movement in obstetric medicine. Jeanty et al. in 1983 described the use of real-time, simultaneous ultrasound-guided technique, with constant visualization of the needle, the amniotic fluid, and the fetus [23]. The currently accepted procedure helps to improve the needle induction in relation to closely located organs of the mother [24]. It also allows for better control and maneuverability of the needle angle during the procedure [24]."

An NICHD-sponsored investigation showed that about 3.5% of pregnant women that elected for an amniocentesis during 1972 up to 1975 suffered from miscarriage after the procedure, which was significantly different from the controls (3.2%) who did not elect to have amniocenteses [25, 26]. Conversely, a British collaborative study during the same period (1973–1976) reported rates of fetal after the procedure to be notably greater than its controls (2.6% vs. 1%) [27]. Similar results were found in a study from Denmark, where the miscarriage rate was 1.7% in females who had amniocentesis compared to 0.7% in the control group ($P \ll 0.05$) [25]. It is critical to realize that in all the previously discussed studies simultaneous ultrasound guidance was not used [22]. Overall, the attributable risk for loss of the fetus secondary to amniocentesis in a normal population is closely 0.5% [24].

Statistically, amniocentesis is considered to be a safe procedure with a complication rate affecting >2% of cases [21]. However, the complications that can occur can potentially become very damaging to a family such as a debilitating injury to the fetus or a miscarriage [21]. In this case, it proved detrimental to the patient as well as to his family. Therefore, amniocentesis should be performed with great care and attention to detail, to minimize complications that affect the lives of patients.

Conflict of Interest

None declared.

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