Extra-renal, extra-neural atypical teratoid/rhabdoid tumor (ATRT): imaging Features of uncommon tumor

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Learning objectives

Learning Objectives

• To review the epidemiological, clinical and pathological features of the ATRT.
• To increase the awareness of the radiological features of the ATRT in the renal and CNS.
• To demonstrate the radiological features of the extra-renal extra neural ATRT.
Background

Atypical teratoid rhabdoid AT/RTs tumor is an uncommon aggressive pediatric neoplasm of uncertain origin. It was first identified in the kidney of infants and children and was described in 1978 as rhabdomyosarcomatoid variant of Wilms’ tumor. It was not recognized as a distinct entity until the 80s. It commonly occurs in central nervous system and kidney. Recently some reports demonstrate its occurrence in other anatomical location rather than the kidney and CNS. These poorly differentiated neoplasms may pose a great deal of difficulty in the diagnosis particularly when occur in unusual locations. Along the past 3 years we record about ten cases of ATRT at extra-neural and extra-renal locations.

The histopathologic diagnosis of extrarenal rhabdoid tumor is based on the presence of characteristic features similar to those found in kidney ATRT. These include large oval to polygonal cells with abundant eosinophilic cytoplasm, large vesicular nuclei with prominent nucleoli, and conspicuous cytoplasmic inclusions. Ultrastructurally, cytoplasmic inclusions are composed of concentric arrays of parallel intermediate filaments, 6-9 nm in diameter. The filamentous cytoplasmic inclusions are not membrane bound and occasionally incorporate lipid droplets or mitochondria.
Findings and procedure details

Imaging Findings

The ATRT occur in young children (median age is less than 2-3 years). It usually presented by palpable masses. US is usually the first screening tool followed by CT. For CNS lesions the MRI is the primary imaging tool. The large heterogeneous solid mass is the common radiological presentation. The lymphatic deposits are rare.

1. Renal atypical teratoid/rhabdoid tumor (ATRT)

Renal tumors comprise 7-8% of all pediatrics tumors. Rhabdoid tumor of kidney is a rare neoplasm of childhood comprising about 2% of renal tumors of childhood. Previously it is thought to be a sarcoma variant of Wilms' tumor, now it is recognized as a distinct pathologic entity. Infants are more affected, with a median age of 11 months and over 80% of patients are under 2 years with male predominate by a ratio of 1.5:1. Associated synchronous or metachronous primary intracranial masses or brain metastases has been established as a distinctive feature (Fig.2&3). The brain lesion is usually near the midline and often in the posterior fossa or pineal region.

Clinically: ATRT usually present as a lion mass, patients may develop hypercalcemia secondary to elevated parathormone levels. The serum calcium level tends to normalize after surgical resection.

Imaging demonstrates a large, centrally located, heterogeneous soft-tissue mass involving the renal hilum with indistinct margins. Its appearance is closely resemble other renal masses specially the Wilms tumor, yet there are some features may help in differentiation: subcapsular fluid collections (Fig.1) and the lobular appearance of the tumor. These tumor lobules also were often separated by intervening low-density areas of hemorrhage or necrosis. Calcification is more common in ATRT than Wilms' tumors and the clear cell sarcoma. Also, calcifications in rhabdoid tumors tend to be hyperdense linear areas outlining the tumor lobules. Vascular invasion, metastases, local invasion, or distinctness of the tumor margin do not show any specific pattern to help distinguish a rhabdoid tumor from other renal tumors. Lack of liver metastasis at the time of diagnosis may be important for therapy and prognosis but will not help differentiate a rhabdoid tumor from other pediatric renal tumors. There is need to search for a synchronous CNS lesion, which seems frequent in rhabdoid tumors.

Rhabdoid tumor has the worst prognosis of all renal tumors. It is highly aggressive and metastasizes early, most commonly to the lungs and less often to the liver, abdomen, lymph nodes, or skeleton. Usually most patients presenting with advanced disease.
2. CNS atypical teratoid/rhabdoid tumor (ATRT)

Atypical teratoid/rhabdoid tumor of the CNS is an aggressive infantile neoplasm of uncertain origin. In 1995, Rorke et al described a special type of CNS tumor that partially or totally consisted of rhabdoid cells resembling a malignant rhabdoid tumor of the kidney and usually also contained areas resembling primitive neuroectodermal tumor (PNET) and sometimes foci of cells indicating a mesenchymal or epithelial origin. They named it "atypical teratoid/rhabdoid" tumor. The average age of presentation of primary ATRT of brain is two years (with a range of one month to twelve years) with 1.9:1 male predominance. The infratentorial compartment is the most common location (Figs.5) but it was documented in the supratentorial and intra ventricular compartments (Fig. 6) as well. Clinical presentations are non specific; depend upon the site of occurrence. It carries a poor prognosis with frequent local recurrences and dissemination through CSF pathways, about one-third of tumors already have CNS dissemination at presentation.

The imaging features usually are non-specific, ATRT usually present as a large heterogeneous space occupying lesion. CT shows heterogeneous lesion with zones of iso or slight hyperdensity alternating with cystic and necrotic hypodense areas, presence of calcification (Fig. 3) or hemorrhage. On MRI, the lesion appears hypointense on T1WI and iso-to-hyper intense on T2WI, with inhomogeneous enhancement. Its heterogeneous appearance is because of presence of cystic and necrotic areas with occasional hemorrhagic changes and moderate to marked surrounding edema. It also shows marked restricted diffusion with reduced ADC value reflecting the high cellularity and aggressiveness of such tumor. Obstruction of CSF pathways can lead to hydrocephalus (Fig.4).

The main differential diagnosis of the posteriopr fossa ATRT is medulloblastoma, the location of the tumor and pattern of tumor growth are important differential points, the medulloblastoma usually arises from the vermian area and grows into the fourth ventricle, while ATRT may arise from the cerebellar hemisphere and grow into the adjacent cisternal space instead of filling the fourth ventricle (Fig.4). Compression and displacement of the fourth ventricle by the mass effect of the tumor is very unusual in medulloblastoma.

Clinically, the patient's age is another differential point, Medulloblastoma is rare in infants, and the median age for diagnosis is about 6 years while the ATRT is a tumor of infancy and childhood presents at a younger age (median age at diagnosis being 16.5 months).

Other tumors like ependymoma, choroid plexus papilloma, supratenterial PNET. The earlier age of onset, larger size and polymorphic appearance help in proper diagnosis.

3. Orbital atypical teratoid/rhabdoid tumor (ATRT).
Orbital ATRT is a extremely rare but highly malignant neoplasm. First reported by Rootman et al. in 1989, there have been few published cases of ATRT of the orbit.

Although ATRT characteristically has an early age at onset, ATRT occasionally presenting at birth is referred to a congenital.

We report a case of orbital ATRT in a male at age of 1 month, presented clinically by a large right ocular swelling. MRI of the head and orbits revealed a large, well-delineated right orbital mass eliciting iso to hypo intense signal on both T1WI and T2WI with heterogeneous enhancement on post Gd series. No evidence of surrounding bony erosion or intracranial extension (Fig. 7).

The patient developed multiple subcutaneous solid masses, the largest at the right axilla also in the right groin (Fig.8). These lesions appear uniformly hypo dense with no internal calcification or cystic degeneration. The underlying bone is intact.

4. Neck atypical teratoid rhabdoid tumor (ATRT)

ATRT have been described in multiple anatomic locations. According to the literature, less than 10 cases with ATRT in the neck soft tissue were reported without any metastasis at the time of diagnosis. The cases were between 5-54 months old at the time of diagnosis. We report a case of ATRT at the right side of the neck in a 7 years old male patient. MRI (Fig.9) showed an ill defined heterogenous mass lesion seen at the right aspect of the neck extending deeply into the parapharyngeal space showing heterogenous intensity and enhancement with restricted diffusion and reduced ADC value reflecting the aggressive biological behavior of the tumor. The mass was rapidly growing; CT done 1 week after MRI (Fig. 10) the mass appeared huge infiltrating the subcutaneous fat with ulceration of the overlying skin. It appears heterogeneously hypo dense with no internal calcification.

Another case of lower cervical and upper mediastinal ATRT is also encountered in a 2 months male patient present with respiratory distress because the mass compress the air way. (Fig.11). It had a poor prognosis as the baby died after 2 months.

5. Hepatic atypical teratoid/rhabdoid tumor (ATRT)

Liver rhabdoid tumors are a rare histological finding. Infant and young children are the most affected patients with a median age of diagnosis of 16.7 months. Moreover nearly all cases (89%) occurred in the first 2 years of life, with no gender difference. The outcome of the other published cases was uniformly fatal, despite aggressive treatment. The overall mortality rate was 89% with a mean survival of 15.3 weeks.
We report a case of a huge heterogeneously hypodense hepatic mass with no internal calcification or fat density. It is seen involving the right diaphragmatic copula associated with pulmonary deposits on presentation. (Fig. 12)

6. Abdomino-pelvic atypical teratoid/rhabdoid tumor (ATRT)

The ATRT in the pelvic cavity is an extremely rare, up to our knowledge there is no report in literature describing the imaging features of the pelvic ATRT.

We present three cases of abdominal and pelvic ATRT:

A 1 month male patient presenting with rapidly growing abdominal swelling. CT shows a huge abdominal mass lesion is seen inseparable from the left hepatic lobe inferior surface, displacing the bowel all around and causing bulge of the abdominal wall. The lesion appears hypo dense and poorly enhanced with no internal calcification. Associated with few metastatic hepatic focal lesions also noted at presentation (Fig. 13).

A 4 month male patient presented with a lower abdominal mass lesion. US showed a heterogeneously hypo echoic solid mass lesion, CT showed a huge heterogeneous solid deeply seated pelvic mass lesion, no internal calcification. No definite evidence of invasion of surrounding structures. The mass compress the left ureter with mild back pressure changes (Fig. 14).

10 years old female patient presented with rapidly growing abdominal mass, CT showed a huge well-defined heterogeneous hypodense solid mass is seen occupying the lower abdomen and pelvis. It is seen extending into the right retroperitoneal region extending behind the right psoas muscle compressing it and infiltrating it. No intrinsic calcifications. No intraspinal extension. Associated moderate amount of ascites is noticed with peritoneal thickening and nodularity. Surgical excision and pathological assessment revealed ATRT with peritoneal deposits (Fig. 15).

7. Extremities atypical teratoid/rhabdoid tumor (ATRT)

ATRT in the upper and lower limbs resemble the soft tissue sarcomas both in clinical and radiological appearance, it diagnosed on pathological background. We report two cases of ATRT one on the axilla and another one in the calf muscle.

- 2 year old female patient presented with rapidly growing right axillary swelling. CT showed a large right axillary heterogeneous soft tissue mass lesion. No internal calcification. No intra-thoracic extension. No significantly enlarged lymph nodes. No evidence of distant metastases at the time of presentation. (Fig 16)
- 4 year female patient presented with right leg mass. MRI showed a large rather defined solid soft tissue mass lesion seen occupying the right popliteal fossa in the posterior aspect of the knee extending into the upper leg. The lesion is seen involving both heads of gastrocnemius muscle. It elicits low signal in T1wi that changed to high signal in T2 with evident heterogeneous post contrast enhancement (Fig.17). CT done and confirm absence of internal calcification and intact cortex of the adjacent bones. The patient developed metastatic pulmonary nodules later on (Fig.18).
A rather defined large mass lesion is seen consuming the left kidney. No internal calcification. The mass crossing the midline. No evidence of invasion of surrounding structures or enlarged lymph nodes.

Fig. 1

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Fig. 2

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Fig. 3

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an ill defined heterogenous space occupying lesion seen at left lateral ventricle showing calcific foci, heterogenous enhancement and restricted diffusion
2 year old female patient presented with chronic headache.

large intra-axial heterogeneously enhanced space occupying lesion located at the right cerebellar hemisphere. WI shows restricted diffusion, with ADC value $0.7 \times 10^{-3} \text{ mm}^2/\text{sec}$

Fig. 4

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Fig. 5

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Fig. 6

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2 year female patient presented with headache

an ill-defined heterogenous space occupying lesion seen at right lateral ventricle showing isointense signal on T1&T2WI, heterogenous enhancement and restricted diffusion.
1 month male patient presented with right orbital mass

A large, well-delineated right orbital mass eliciting iso to hypo intense signal on both T1WI and T2WI with heterogeneous enhancement on post Gd series. No evidence of surrounding bony erosion or intracranial extension.

Fig. 7

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• Multiple subcutaneous solid soft tissue mass lesions are seen at the right axilla, right groin and right leg.

Fig. 8

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7 years old male patient presented with right cervical mass lesion

- An ill defined heterogenous mass lesion is seen at the right aspect of the neck extending deeply into the parapharyngeal space showing heterogenous intensity and enhancement with restricted diffusion reflecting the aggressive biological behavior of the tumor.

Fig. 9

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Fig. 10

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2 month male patient present with respiratory distress

An ill defined heterogenous superior mediastinal mass lesion is seen at the left aspect of the lower neck and upper mediastinum compressing the upper air way.

Fig. 11

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1 year male patient presented with right hypochondrial swelling.

A large heterogenous solid soft tissue mass lesion is seen involving the right hepatic lobe compressing the right kidney. No internal calcification. It shows faint enhancement. No venous extensions or lymph node deposits.

Fig. 12

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Fig. 13

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A huge well-defined heterogeneous hypodense solid mass is seen occupying the lower abdomen and pelvis. It is seen extending into the right retroperitoneal region extending behind the right psoas muscle compressing it and infiltrating it. No intrinsic calcifications. No intraspinal extension. Associated moderate amount of ascites is noticed with peritoneal thickening and nodularity.

Fig. 14

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Fig. 15

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- A huge heterogeneous solid deeply seated pelvic mass lesion, no definite evidence of invasion of surrounding structures. The mass compress the left ureter with mild back pressure changes.
Fig. 16

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2 year old female patient presented with rapidly growing right axillary mass

a large right axillary heterogeneous soft tissue mass lesion. No internal calcification. No intra-thoracic extension. No significantly enlarged lymph nodes. No evidence of distant metastases at the time of presentation.
4 years female presented with right leg mass

Fig. 17

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A large rather defined solid soft tissue mass lesion seen occupying the right popliteal fossa in the posterior aspect of the knee extending into the upper leg. The lesion is seen involving both heads of gastrocnemius muscle. It elicits low signal in T1wi that changed to high signal in T2 with evident heterogeneous enhancement.
Fig. 18

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CT confirmed the absence of calcification and intact bony cortex of adjacent bones. Multiple bilateral scattered pulmonary nodules.
Conclusion

Extracranial rhabdoid tumours are rare. Although the kidney is the most common site, they can occur anywhere in the body. It usually has a bad prognosis, so the awareness of the radiological features of ATRT particularly in the unusual locations and consider it in the differential diagnosis may help in proper early diagnosis thus improve the patient’s outcome.

Although, there is no specific diagnostic features can help in definite diagnosis of ATRT on radiological background, we found that most of the cases presented as a large sized ill defined heterogenous masses. None of them show internal calcification, none of them associated with lymphatic deposits. These features may put ATRT in the diagnostic list when radiologist assesses a rapidly growing mass in a young child.
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References