DISCUSSION

Malignant extrarenal rhabdoid tumours are highly malignant soft tissue tumours that are analogous to the rhabdoid tumours of the kidney or atypical teratoid/rhabdoid tumors of the central nervous system. These occur in the paediatric population, arise in multiple anatomical locations and may develop sporadically or as part of a hereditary syndrome called 'Rhabdoid tumour predisposition syndrome' (RTPS)[1].

These tumours are extremely rare with an incidence of less than one per million of paediatric population. Only sixteen cases of extrarenal rhabdoid tumours presenting as a neck mass have been reported in literature.

Histopathologically, the tumour cells show a characteristic 'rhabdoid' morphology which comprises of presence of eccentric, vesicular nuclei; prominent nucleoli, well-defined cytoplasmic borders and abundant eosinophilic cytoplasm containing occasional pale cytoplasmic inclusion bodies. Brisk mitotic activity is usually seen[2].

'Composite' extrarenal rhabdoid tumours may display a combination of cellular elements consisting of undifferentiated ''small round blue cells,'' mesenchymal and epithelial components besides the classic rhabdoid phenotype, and sampling limitations of this rhabdoid component can pose as a diagnostic challenge.

Extrarenal rhabdoid tumours are polyphenotypic neoplasms exhibiting consistent and diffuse immunopositivity for vimentin and variable immunopositivity for epithelial and neuroectodermal markers. Tumour cells are immunonegative for CD34, desmin, HMB-45 and myogenin; and consistently lose nuclear expression of INI-1 protein due to mutations or deletions of the SMARCB1 gene.

It is important to distinguish extrarenal rhabdoid tumours from other soft tissue and epithelial neoplasms which can have cells with a "rhabdoid appearance," including proximal-type epithelioid sarcomas (SMARCB1 deficient, however tumours are CD34+ve in 50% of cases; usually occur in young adults, less aggressive compared to extrarenal rhabdoid tumours), chordomas (SMARCB1 deficient, occurs in sacral region, anti-brachury +ve), epithelioid malignant peripheral nerve sheath tumours (S-100+ve, CD34 +ve), myoepithelial carcinomas(SMARCB1 deficient, S-100+ve, p63+ve, diffuse panCK+ve), pleomorphic rhabdomyosarcoma (show myogenic differentiation with desmin, myogenin, MyoD1+ve), malignant melanoma (S-100, HMB-45, MelanA+ve) and primitive neuroectodermal tumours (retained expression of INI-1)[3].