RET-Rearranged Small Round Cell Sarcoma

Sir,

Undifferentiated small round cell sarcoma (uSRCS) often poses diagnostic challenges.[1] CIC-rearranged undifferentiated small round cell sarcoma (CIC-rearranged sarcoma) is a subtype of small round cell sarcoma (SRCS), characterised by genetic rearrangement, most commonly between the CIC and DUX4 genes (CIC-DUX4 fusion).[2] The studies have found that round cell sarcomas occurring in bones or soft tissues exhibit unique clinical, morphological, and immunophenotypic characteristics.[3] The RET-rearranged round cell sarcoma described in this paper is extremely rare and is detailed below.

A one-year-old male infant was initially treated at a local hospital one month ago for symptoms included crying, body swelling, and intestinal obstruction. Despite receiving symptomatic treatment, his condition did not improve. He was subsequently referred to a tertiary care hospital, where MRI findings revealed a suspected neurogenic malignant tumour, minimal ascites, bilateral pleural effusion, and diffuse soft tissue oedema within the abdominal cavity (Figure 1A). Based on the imaging findings and the patient's condition — and obtaining consent from the family — it was decided to perform tumour resection and biopsy for pathological confirmation.

Relevant laboratory tests and examinations were conducted. Tumour resection and biopsy were performed under general anaesthesia. During surgery, the tumour was completely dissected and resected (Figure 1B). The resected tumour was subjected to pathological examination, immunohistochemical analysis, and genetic testing. The morphological findings, combined with immunohistochemistry and molecular testing, were consistent with RET-rearranged SRCS (Figure 1C-G). Genetic testing results showed an RET gene mutation, specifically CCDC6-RET fusion. The mutation types included C2: R12 (466 fusion reads) and C2: R11, with variant abundance of 14.52%. RET gene rearrangement detected by Fluorescence *in situ* hybridisation (FISH) was positive (Figure 1H).

RET-rearranged SRCS and CIC-rearranged sarcoma are extremely similar in morphology, therefore, careful differentiation is required. The patient is currently receiving chemotherapy and symptomatic treatment, with radiotherapy included during the treatment cycle. Targeted therapy agents may be used when it is needed. Follow-up is essential to monitor changes in the condition.

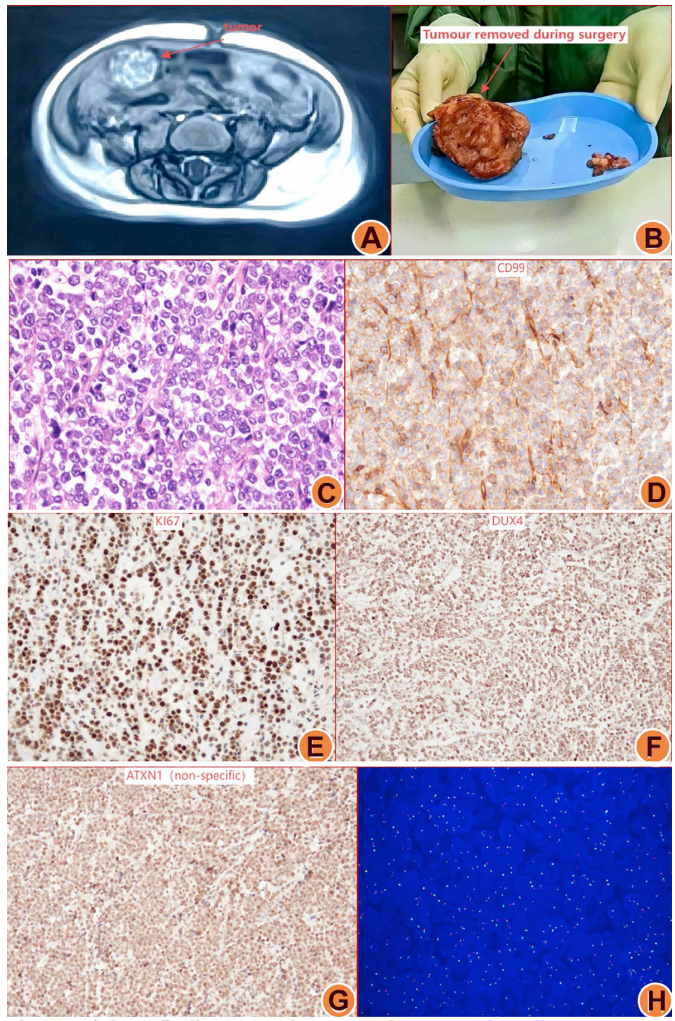

Figure 1: (A) MRI findings reveal suspected neurogenic malignant tumour (red arrow), minimal ascites, bilateral pleural effusion, and diffuse soft tissue oedema within the abdominal cavity. (B) During surgery, the tumour was completely resected. (C-G) The morphological factors, combined with immunohistochemistry and molecular testing, were consistent with RET-rearranged SRCS. (H) RET gene rearrangement by FISH was positive.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

BA, YY: Drafted, revised, and edited the manuscript.
XL: Conducted data collection, analysis, and interpretation.
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