A 35-year-old male visited our hospital complaining of rapidly worsening abdominal distention. Enhanced computed tomography showed a heterogeneous mass measuring 17 cm in diameter in the pelvic cavity, without any sign of peritoneal dissemination or distant metastasis. Pneumatic spaces were observed in the tumor suggesting its connection with the intestinal tract (Fig. 1; axial image, arrowhead). The tumor demonstrated mild hypo-intensity on T₁-weighted magnetic resonance image (MRI) and hyper-intensity on T₂-weighted MRI (Fig. 2, sagittal image).

Under a preoperative diagnosis of intra-abdominal mesenchymal tumor, such as sarcomas, gastrointestinal stromal tumor (GIST), desmoid tumor or neurogenic neoplasm, the patient underwent surgical removal of the tumor including the distal part of the ileum (Fig. 3A, the tumor had penetrated the ileum, arrowhead; Fig. 3B, cut surface of the specimen).

Histopathologically, the tumor consisted of solid and trabecular nests of small round cells partially surrounded by a desmoplastic stroma (Fig. 4). Immunohistochemically, the tumor was positive for cytokeratin AE1/AE3, desmin (Fig. 4, lower inset) and neuron-specific enolase (focal). Based on these findings, the tumor was diagnosed as a sarcoma most consistent with a desmoplastic small round cell tumor (DSRCT) arising from mesentery of the ileum. However, in fluorescence in situ hybridization analysis, only <30% of tumor cells demonstrated an Ewing sarcoma (EWS) gene rearrangement, while it is usually observed in >70% of tumor cells in typical DSRCTs.

The patient is doing well with no evidence of tumor recurrence for 8 months after the operation. Considering the less abundant stroma in the tumor, low frequency of EWS gene rearrangement and less aggressive clinical course, it was suggested that this tumor belonged to a low-malignant subtype of DSRCTs.