Multifocal Macrocystic Lesions: A Case of Diagnostic Mimicry

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BACKGROUND

- Although disease classification for the vascular anomalies has improved, diagnostic imaging mimicry continues to be challenging.
- Malignant pathology can appear similar to a benign lesion and delay appropriate therapy.
- Multifocal cystic lesions can be particularly challenging to characterize.
- We present a case of diagnostic mimicry where clinical intuition was able to overcome conformational bias to identify appropriate diagnosis.

CASE REPORT

A 17-year-old boy was referred for left neck macrocystic lymphatic malformation. Initial visit identified the subcutaneous cystic neck lesion and additionally a markedly distended abdomen with a large palpable mass. He reported abdominal pain and unintended weight loss.

The patient underwent sclerotherapy of the left neck and intra-abdominal macrocysts [Figures 5] with consideration for surgical debulking if symptoms persisted or masses did not respond. Aspiration of the macrocysts of the abdomen revealed brownish fluid less consistent with the typical milky white appearance of lymph or chyle [Figure 6: yellow arrow] and aspiration of the macrocysts of the neck revealed atypical clear watery fluid [Figure 6: red arrow].

A near-complete surgical resection of the abdominal lesions was performed. Formal pathology returned consistent with a mixed germ cell tumor His care was transitioned to the care of the solid tumor oncology team to initiate therapy with bleomycin, etoposide, cisplatin (BEP) chemotherapy [Figures 8-13].

Several data points of initial evaluation were overemphasized resulting in conformational bias:
- Outside diagnosis of a neck lymphatic malformation
- MRI read of lymphatic malformation
- Intra-op pathology review of lymphatic malformation
- Several data points in subsequent evaluation refuted the diagnosis:
  - Lack of response to sclerotherapy
  - Fluid aspirate that appeared inconsistent with lymphatic fluid

Accurate diagnosis required looking beyond the area of chief concern on initial exam, pausing to re-evaluate diagnosis with a lack or response to first-line therapy and investigating further to provide a more accurate pathologic diagnosis. Improving the diagnostic process represents a moral, professional and public health imperative.

CONCLUSIONS