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Case Report: When bias clouds judgment in diagnosing a pediatric mixed germ cell tumor

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Background: Cognitive bias is increasingly recognized as a significant factor contributing to diagnostic errors in the medical field. While interventions to reduce biases have mainly been studied in adult hospital or acute care settings, the impact of cognitive bias on diagnostic errors in pediatrics is also noteworthy.
Case presentation: We discuss a 17-year-old who presented for treatment of a clinically and radiographically diagnosed macrocystic lymphatic malformation. Overcoming the challenges of diagnostic anchoring and medical mimicry, medical evaluation was abruptly adjusted, and surgical resection revealed a diagnosis of immature teratoma.
Conclusion: The case helps highlight how cognitive biases can contribute to diagnostic delay and exemplifies methods employed to overcome bias. Understanding and addressing cognitive biases are essential for improving diagnostic accuracy and patient care in pediatric settings.

KEYWORDS
cognitive bias, diagnostic mimic, interventional radiology (IR), lymphatic malformation (LM), outside hospital (OSH), pediatric germ cell tumor, teratoma

1 Introduction

Pediatric cervical neck masses can be caused by a range of inflammatory, neoplastic, and congenital processes. Whereas some diagnoses are benign, such as congenital or acquired cysts, others like solid tumors demand more acute intervention. Accurate and timely diagnosis requires comprehensive clinical history and exam and may require radiographic, pathologic, or genetic evaluation. However, there can be significant overlap in clinical presentations, and radiology reports can range from nonspecific to overly committed. Cognitive shortcuts and biases may also influence diagnostic decisions, potentially resulting in incorrect diagnoses or treatment delays.

We present a case of a pediatric cervical neck mass that illustrates diagnostic mimicry, wherein clinical intuition played a pivotal role in overcoming confirmation bias. A structured, multidisciplinary approach helped identify the correct diagnosis of a mixed germ cell tumor that required immediate intervention. We emphasize the importance of recognizing cognitive biases to improve diagnostic accuracy and patient outcomes.

2 Case description

2.1 Patient information & clinical findings

A seventeen-year-old male presented in rural Montana with a newly appearing left neck mass causing progressive discomfort and airway compression. Magnetic resonance imaging (MRI) showed a left lower neck multi-cystic mass extending into the supraclavicular and subcarinal regions of the mediastinum, consistent with macrocystic lymphatic malformation. Local Otolaryngology attempted aspiration, but insufficient fluid was obtained for relief, prompting transfer out of state for multidisciplinary vascular anomaly center (VAC) care. Past medical, family, and psychosocial history were non-contributory, with no known genetic syndromes or prior malignancy.

2.2 Diagnostic assessment

VAC evaluation confirmed left cervical cystic masses, the largest measuring 7 × 7 cm. The lesion was non-tender but firm, raising concern for intralesional hemorrhage. Additional history revealed a 40–pound weight loss, emesis, and early satiety over the preceding months. On exam, the abdomen was markedly distended with a palpable mass in the left upper and lower quadrants. Full-body MRI/MRA revealed a large multiloculated cystic abdominal mass suggestive of additional lymphatic malformation sites (Figure 1A). The differential diagnosis included complex lymphatic anomaly, venous-lymphatic malformation, and germ cell tumor, with evolving clinical and procedural findings progressively arguing against a vascular etiology.

Interventional Radiology was consulted for aspiration and possible sclerotherapy. Abdominal fluid was brown-tinged, and cervical fluid was clear, both atypical for lymph and chyle (Figure 1B). Sclerotherapy was initially attempted on the cervical lesion; however, fluid analysis (triglyceride 13 mg/dl; lymphocytes 2%) was not consistent with lymphatic fluid, so further attempts were discontinued.

Multidisciplinary consensus led to surgical resection of the abdominal lesion. A 12-h operation resulted in removal of 95% of a 25 × 25 × 20 cm retroperitoneal mass. The mass had both solid and cystic components and encased critical structures, including the aorta, inferior vena cava, kidneys, ureters, and iliac vessels (Figure 2A). Pathology revealed a mixed germ cell tumor, approximately 98% immature teratoma (Figures 2B, E), with additional mature teratomatous elements including squamous, neural, and gastrointestinal-type epithelium (Figures 2C, D). Foci of embryonal carcinoma were identified (Figure 2F), along with areas consistent with yolk sac tumor (Figure 2G). Tumor markers were elevated: AFP 1,240 ng/ml (ref 0–8), HCG 268 mIU/ml (ref 0–5), confirming the diagnosis. No major diagnostic access barriers were reported.

Abbreviations: AFP, alpha fetal protein; ENT, Ear/Nose/Throat; HCG, human chorionic gonadotropin; LM, lymphatic malformation; MRI, magnetic resonance imaging; OSH, outside hospital; VAC, vascular anomaly center; BEP, Bleomycin, Etoposide, Cisplatin; YST, yolk sac tumor.

2.3 Therapeutic intervention

Following pathologic confirmation and tumor marker elevation, the patient was transitioned to oncology care and initiated on standard Bleomycin, Etoposide, and Cisplatin (BEP) chemotherapy, without dose modification or delay.

2.4 Follow-up and outcomes

The patient was followed clinically and radiographically following initiation of chemotherapy. Follow-up imaging demonstrated regression of residual disease, and tumor markers declined appropriately. Treatment was well tolerated without significant toxicity, dose delays, or interruptions. Adherence was supported through shared decision-making, frequent outpatient follow-up, and active patient engagement in treatment planning.

2.5 Timeline of clinical events from presentation to treatment

Schematic diagram showing the diagnostic and treatment timeline from symptom onset to initiation of chemotherapy is showed in Figure 3.

3 Discussion

This case report was prepared in accordance with the CARE (CAse REport) guidelines, and the completed CARE checklist is provided as supplementary material.

Pediatric cervical neck masses have a broad differential, including cystic, inflammatory, and neoplastic etiologies such as immature teratomas. These entities often share overlapping and nonspecific clinical and imaging features, increasing the risk of diagnostic error and underscoring the need for multidisciplinary evaluation. Diagnostic checkpoints that should prompt reconsideration of lymphatic malformation include: (1) systemic symptoms such as weight loss or early satiety, which are atypical for isolated lymphatic disease; (2) multifocal involvement across non-contiguous anatomic compartments; (3) aspirate characteristics inconsistent with chyle or lymph; and (4) lack of biochemical or clinical response to sclerotherapy. When an initial diagnosis fails to account for the full clinical picture, structured re-evaluation is essential to avoid diagnostic delay.

In this case, macrocystic lymphatic malformation was initially supported by imaging and subspecialty assessment, and evaluation at a VAC followed standard lymphatic pathways. However, the presence of multifocal disease, gastrointestinal symptoms, and significant weight loss deviated from typical lymphatic malformations. An atypical complex vascular anomaly, including CLA with possible intestinal lymphangiectasia, was initially considered.

Optimal evaluation in such cases requires early multidisciplinary involvement: radiology to reassess imaging patterns; interventional radiology to evaluate aspirate characteristics and procedural response; pediatric surgery to

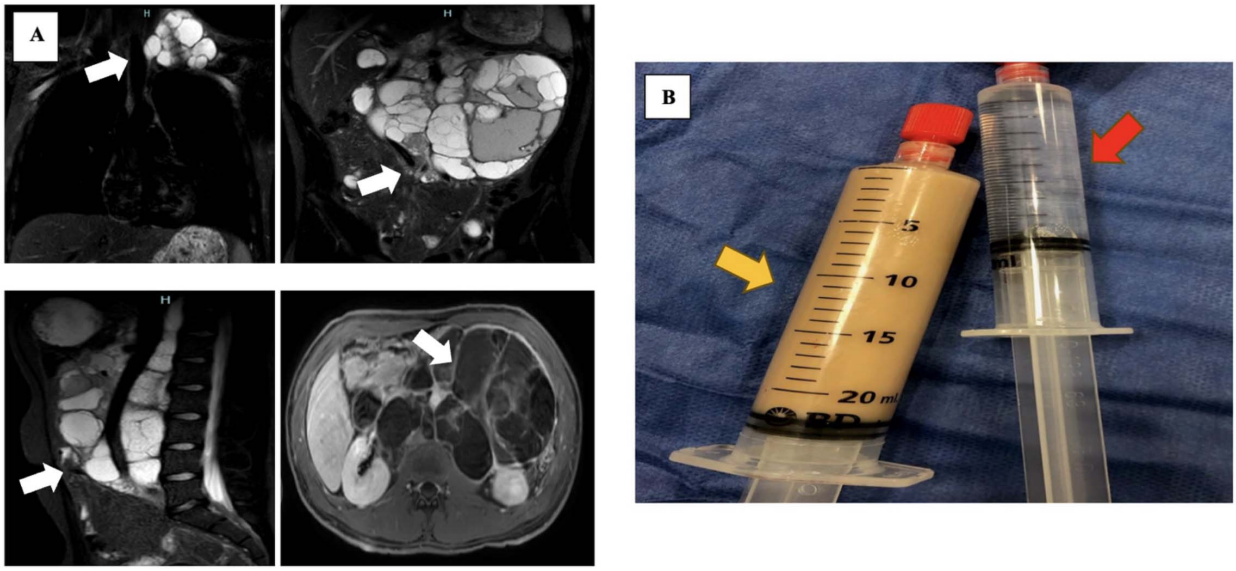

FIGURE 1
Initial radiologic and interventional findings suggesting but contradicting lymphatic malformation. **(A)** Full body MRI/MRA reported multifocal macro-cystic abdominal masses and a macro-cystic left neck mass with concern for generalized lymphatic anomaly. **(B)** Aspiration of the macro-cysts of the abdomen revealed brownish fluid less consistent with the typical milky white appearance of lymph or chyle [yellow arrow] and aspiration of the macro-cysts of the neck revealed atypical clear watery fluid [red arrow].

assess resectability and anatomic risk; pathology to establish definitive diagnosis; and oncology to integrate tumor markers and guide timely therapy.

3.1 Strengths and limitations

This case highlights the strength of multidisciplinary collaboration, where clinical reasoning, surgical insight, and reevaluation of imaging enabled diagnosis. However, a key limitation was the initial reliance on typical pathways without questioning atypical features, which delayed definitive treatment.

3.2 Comparison with literature

A limited number of reports describe teratomas mimicking vascular anomalies, but few address diagnostic delay related to cognitive bias. Unlike prior cases diagnosed early through surgery, this case illustrates how diagnostic mimicry and specialty silos can delay recognition without active re-evaluation. Diagnostic errors account for approximately 75% of malpractice lawsuits and 38% of total payouts (1), with cognitive biases, particularly confirmation bias, being major contributors. Pediatricians report diagnostic errors affecting patient outcomes at least annually (2–5), and misdiagnosis remains a leading cause of avoidable harm and cost (1, 4, 6, 7).

Lymphatic malformations, typically diagnosed through history, examination, and imaging (4), may demonstrate nonspecific radiologic features and be confused with teratomas (8, 9). Key diagnostic mimickers include complex lymphatic anomalies and congenital cystic neck masses, particularly when cystic morphology is interpreted without integrating systemic symptoms or treatment response. These entities require distinct management strategies, underscoring the importance of accurate differentiation (10–12).

In this case, atypical aspirate characteristics, systemic symptoms, and failure to respond to sclerotherapy prompted diagnostic re-evaluation. Definitive diagnosis of a mixed germ cell tumor was achieved only after surgical intervention, emphasizing the necessity of reassessing assumptions in light of evolving clinical and procedural data.

3.3 Scientific rationale

Anchoring and confirmation bias (Table 1) played key roles. Anchoring refers to over- reliance on initial impressions, while confirmation bias involves selectively interpreting data to support those impressions. Both can coexist and compound diagnostic error (1, 13). Mitigation requires pausing premature closure, actively seeking contradictory evidence, and continuously re-evaluating data. Radiologists and clinicians alike must challenge early assumptions. When a diagnosis fails to fully explain the clinical course, it's essential to ask: What else could this be? What doesn't fit? (1, 13).

Metacognition and cognitive forcing strategies can support more objective decision-making. Avoiding the pseudo-diagnostic effect, relying on irrelevant data to support a diagnosis, is vital. Clinicians should not hesitate to revisit or revise an initial diagnosis if clinical evolution no longer fits. Helpful reflection questions

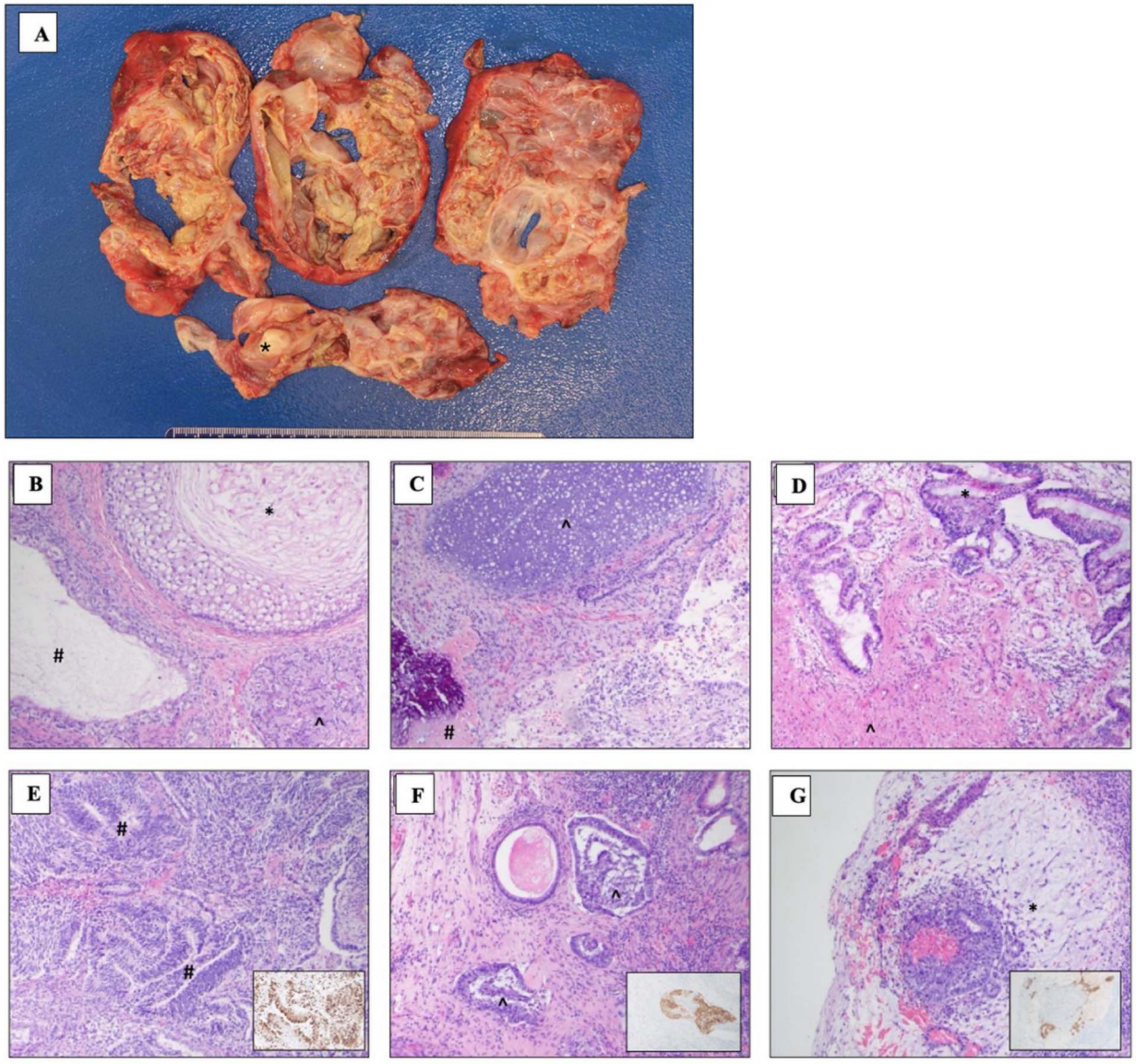

FIGURE 2
Gross and histopathologic features of a mixed germ cell tumor **(A)** Cross-sections of large, multiloculated, predominantly cystic mass with smooth, fibrous septa. Scattered amorphous tan solid nodules are present (*) comprising <2% of the total volume. **(B-D)** Microscopic examination. Mature teratoma component consisting of **B**. squamous epithelium (*), ganglion cell cluster (), and ciliated respiratory- like epithelium (#) (H&E, 10x); **(C)** cartilage () and bone (#) (H&E, 10x); and **D**. small bowel-type epithelium (*) with adjacent necrotic tissue () (H&E, 10x). **(E)** Immature teratoma component consisting of immature neuroepithelium (#) (H&E, 10x) exhibiting a high proliferative index (inset, Ki-67, 4x). **(F)** Embryonal carcinoma component () consisting of primitive epithelial cells with prominent cellular polymorphism (H&E, 10x), highlighted by CD30 immunohistochemistry (inset, CD30, 4x). **(G)** Yolk sac tumor component consisting of sparse stellate cells in a myxoid background (*) (H&E, 10x), highlighted by AFP immunohistochemistry (inset, AFP, 4x).

include: What parts of this picture are inconsistent? Have I overlooked alternatives? (10).

Enhancing clinical reasoning means balancing intuitive and analytical thinking (Table 1) while remaining vigilant for bias. Physicians face barriers to consistent analytical reasoning, such as time pressures, sleep deprivation (10), and cognitive overload. Medical education should emphasize debiasing strategies, guided reflection, and structured re-evaluation. Cultivating open discussion, a questioning mindset, and adopting technology such as clinical decision support systems or AI tools can help minimize diagnostic error and improve care (14).

4. Takeaway message

Improving the diagnostic process is a clinical and moral imperative. Cognitive biases significantly contribute to diagnostic errors and delays, particularly in pediatric neck masses where

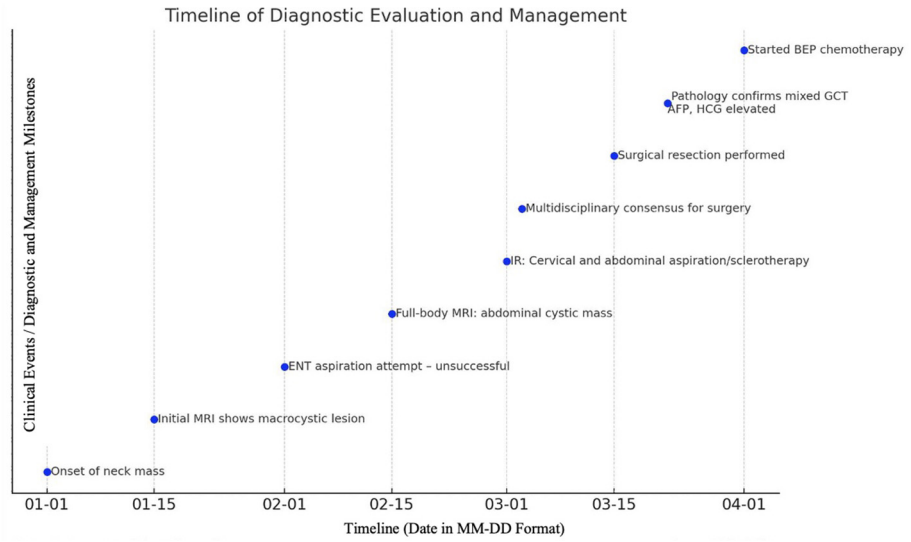

FIGURE 3
A structured chronology of key milestones in diagnosis and management, including symptom onset, initial imaging, multidisciplinary evaluations, interventional procedures, surgical resection, and initiation of chemotherapy.

TABLE 1 Key definitions related to cognitive bias and diagnostic reasoning.

TERM	DEFINITIONS
Anchoring bias	A cognitive bias in which individuals rely too heavily on initial information or the first piece of evidence they encounter when making decisions or judgments. Once the anchor is set, subsequent information is often interpreted or adjusted in relation to the initial reference point, leading to potential errors and overlooking alternative possibilities.
Confirmation bias	A cognitive bias that involves seeking, interpreting, or favoring information that confirms or supports preexisting beliefs, hypotheses, or expectations, while disregarding or downplaying evidence that contradicts them. It can hinder objective and comprehensive evaluation, leading delayed consideration of alternative diagnoses.
Metacognition	A healthcare professional's ability to actively reflect on their clinical decision- making processes. It involves being aware of their thoughts, biases, and reasoning strategies during patient evaluation and treatment planning. By practicing metacognition, providers can improve diagnostic accuracy, identify potential cognitive errors, and continuously improve their medical judgment.
Perspective seeking	Actively seeking and considering alternative viewpoints and expert opinions. This is particularly valuable in complex cases or when encountering ambiguous medical information. By seeking different perspectives, providers can make well informed and comprehensive assessments and potentially avoid diagnostic errors.
Cognitive forcing strategies	Methods used to stimulate creative thinking and foster comprehensive evaluation. These strategies encourage providers to think beyond initial impressions, consider various diagnostic possibilities, and explore potential treatment options thoroughly.
	• Case-based discussions: engaging in peer discussions. • Red team reviews: inviting external expert review. • Prospective hindsight: imagining future outcomes to anticipate potential pitfalls. • The five whys technique: repeatedly asking "why" to identify root cause. • Decision trees and flowcharts: Visual representations to consider multiple decision pathways. • Deliberate consideration of contrary evidence: actively seeking and evaluating evidence that contradicts initial hypotheses. • Simulation-based reviews: using virtual scenarios to test critical thinking and adaptability.
Intuitive thinking	The rapid processing of clinical information based on experience and expertise to make quick decisions in urgent or familiar situations.
Analytical thinking	Systematically analyzing patient data, medical records, and diagnostic test results emphasizing evidence-based reasoning and critical evaluation to arrive at wellsupported conclusions.
Guided reflection	The structured process of healthcare professionals reviewing challenging cases or patient outcomes with the guidance of mentors, peers, or review committees.

anatomy, radiology, and subspecialist access complicate evaluation. Addressing these biases is essential to improving accuracy and outcomes. This case illustrates how challenging confirmation and anchoring bias, combined with a multidisciplinary approach, led to a life- saving diagnosis. Recognizing diagnostic mimicry and applying critical thinking throughout the clinical course can improve care in complex pediatric conditions such as mixed germ cell tumors and lymphatic malformations.

Data availability statement

The original contributions presented in the study are included in the article/supplementary material, further inquiries can be directed to the corresponding author.

Ethics statement

Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article.

Author contributions

EE: Writing – original draft, Visualization, Writing – review & editing. LM: Writing – original draft. AK: Writing – review & editing. AA: Visualization, Writing – review & editing. BS: Writing – review & editing, Visualization. TN: Supervision, Writing – review & editing, Resources.

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