CASE REPORT

Not such a simple cyst: unusual presentation of sarcoma

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SUMMARY

A 10-year-old girl presented with a lump on her left upper anterior thigh. She presented to accident and emergency twice and to her general practitioner once. It was provisionally diagnosed as a cyst but it continued to grow in size. The lump had been noticed for 6 months, in paediatric clinic it was 5×5 cm, firm, tense and felt cystic. Systemically she was otherwise well. The overlying skin was blueish with prominent veins. The MRI also suggested a cyst but on excision it was revealed to be a high-grade sarcoma. After surgical excision she had a skin graft. Regular follow-ups with MRI have not shown any recurrence, she also has regular chest X-rays which have all been normal.

BACKGROUND

It took a long time and three presentations for this lump to be biopsied. It presented clinically as a cyst and MRI supported this diagnosis. It is important because it is an atypical presentation of a high-grade sarcoma which has obvious potentially severe consequences.

CASE PRESENTATION

A 10-year-old girl presented with a 6-month history of a lump on her left thigh, gradually increasing in size. When she presented to accident and emergency for a second time she was urgently sent to paediatrics. There was no history of trauma or infection in the area. The lump was on the anterior of the upper part of the left thigh and was noted to be 5×5 cm at time of presentation to the paediatric team. It was dome-like and felt firm, tense and cystic. The overlying skin was blueish with visible veins and a well-demarcated edge (figures 1 and 2).

INVESTIGATIONS

There were concerns about the origin of the lump so an MRI was performed. The report described a 3.7 cm cyst in the subcutaneous tissue of the left thigh with no local infiltration seen. She was then referred to a specialist paediatric centre where an excision biopsy with graft was performed. Histology revealed high-grade sarcoma, but it did not appear to fit into a typical embryological sarcoma category. "This specimen comprises a large

Figure 1 Lesion on patient's left thigh.

Figure 2 Closer view of lesion on patient's left thigh.

CrossMark

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well-circumscribed encapsulated intradermal lesion. The epidermis and overlying papillary dermis appear normal. A fibrous capsule is present surrounding the tumour that focally appears somewhat indistinct. The adjacent subcutaneous tissue shows focal areas of fat necrosis particularly in the lateral aspect.” Chromosome analysis of the tumour cells revealed abnormal karyotype of 45 chromosomes with one 13 chromosome missing as well as other translocations. This clone means that it is a cytogenically abnormal tumour. However, it did not show typical Ewing sarcoma abnormalities; however, the results do not entirely exclude a Ewing sarcoma. No metastatic disease was found.

DIFFERENTIAL DIAGNOSIS

Until the time of histology the main differential diagnosis was that of a cyst as she was systemically well and clinical examination and MRI supported this diagnosis.

OUTCOME AND FOLLOW-UP

Followed up with repeat MRI and chest X-ray at 1 year as well as being seen in specialist paediatric oncology and surgery

clinics. The skin graft has taken well and she continues to take good care of it. She remains well and asymptomatic.

Learning points

- ▶ Always be suspicious of lumps increasing in size.
- ▶ High-grade sarcoma can present without local invasion.
- ▶ The only way we can know the origin of a lump is by histology, and sometimes this gives surprising results.

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