CASE REPORT

Unusual cause for testicular swelling in a child

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SUMMARY

Myeloid sarcoma is a rare solid tumour composed of primitive precursors of granulocytic series of white blood cells involving extramedullary anatomic site. Here we report the case of a 10-month-old with testicular swelling, who was finally diagnosed with granulocytic sarcoma. This case demonstrates that Granulocytic sarcoma should be considered as a part of the differential diagnosis for testicular swelling in spite of having normal peripheral counts and absence of hepatosplenomegaly.

BACKGROUND

Myeloid sarcoma (MS) arising in the testis is a rare clinical entity in children, which is more commonly seen in the adult population only.¹ It is an accumulation of myeloblasts in the extramedullary site.² According to Reinhardt and Creutzig,³ extramedullary manifestations in children with acute myeloid leukemia (AML) include 2%–25% of the paediatric population. In that series, only 2.5% of those patients presented with an isolated primary MS. Per the literature reviewed, only five cases of infantile testicular MS have been reported so far. Also in the cases reported so far, those with a negative bone marrow aspirate is very rare, hence we report this case.^{1,2}

HISTORY

A previously healthy 10-month-old male child, third born to non-consanguineous parents, presented with left testicular swelling noticed 2 months earlier. There was no history of trauma or other constitutional symptoms such as pain, fever, weight loss, bleeding manifestations or irritability. Physical examination revealed a healthy child who had a non-tender, hard mass in the testis, measuring 4×4 cm, and a normal right testis (figure 1). No lymphadenopathy or organomegaly was observed. A clinical diagnosis of primary testicular tumour was considered.

INVESTIGATIONS

Haematological examination revealed haemoglobin concentration of 95 g/L, with white cell count of $8.4 \times 10^9/L$, platelet count $160 \times 10^9/L$, and peripheral smear having microcytic hypochromic anaemia without any abnormal blasts. His alpha-fetoprotein, lactate dehydrogenase, uric acid, serum electrolytes, liver and renal function test values were within normal limits. Chest X-ray was normal.

Ultrasonography showed a left testicular hypoechoic mass with increased blood flow, measuring $4 \times 2.9 \times 2.1$ cm, suggestive of probable malignancy.

A biopsy of the testicular mass was performed and samples sent for histology and immunohistochemistry.

Histology report of the mass showed sections of testicular tissue displaying partially effaced architecture and few preserved seminiferous tubules with diffuse interstitial infiltration by neoplastic cells. The cells are monomorphic, medium sized, with scant to moderate granular eosinophilic cytoplasm, and round nuclei with fine vesicular chromatin and distinct single nucleolus. Brisk mitosis and apoptosis are noted. Interspersed infiltrate of mature small lymphocytes is also seen. Seminiferous tubules identified show spermatogonia and Sertoli cells. No infiltration into seminiferous tubules was observed.

Immunohistochemistry showed strongly positive for CD45, CD43, CD34, CD117, CD68 and CD56 with few neoplastic cells positive for myeloperoxidase (MPO), mild positive for CD4, and negative for ALK-1, Tdt, CD99, CD5, CD8, CD10, CD19 and CD20 (figure 2). This MS is reported to be a differentiated type, per the WHO classification.

Positron emission tomography-CT showed only bulky left testis with patchy increased metabolic activity without any involvement in any other parts. Bone marrow aspiration and its biopsy were normal.

Figure 1 Swelling of the left testis.

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Figure 2 (A) Testes parenchyma displaying diffuse infiltration by leukaemic cells (H&E, X100). (B) Strong CD43 immunopositivity (X100). (C) Patchy CD117 immunopositivity in tumour cells (X100). (D) Strong CD34 immunopositivity (X100).

DIFFERENTIAL DIAGNOSIS

- ▶ Primary testicular cancer
- ▶ Secondaries due to leukaemia and non-Hodgkin's lymphoma such as Burkitt's lymphoma and lymphoblastic lymphoma.

MANAGEMENT

Parents were counselled about the child's condition, the nature of the disease, the treatment option for chemotherapy and further follow-up after chemotherapy, and the side effects of the treatment. But in spite of detailed counselling, the family did not agree to treatment and the baby was lost to follow-up.

OUTCOME AND FOLLOW-UP

Lost to follow-up.

DISCUSSION

Testicular MS in children is a rare clinical entity. It is an extramedullary neoplasm of myeloid blasts. In infants to date, only five cases of testicular MS have been reported per literature (table 1). We present the sixth case of MS that presented with isolated testicular swelling.^{1 2 4-6}

MS is referred to by multiple names such as monocytic sarcoma, chloroma, myelosarcoma, extramedullary myeloid cell tumour, granulocytic sarcoma and myeloblastoma.

MS arises from multiple sites such as the skin, soft tissue, bones and lymph nodes; skin being the most common site.⁷ Though most of the cases including ours presented with a testicular swelling, there is also evidence of testicular MS that presented with testicular haemorrhage in the absence of a palpable testicular mass.⁷ Clinical findings that were observed in advanced cases of MS included hydronephrosis, retroperitoneal and mediastinal lymphadenopathy, or gynecomastia.¹

Per the literature reviewed by Douet-Guilbert *et al*, out of 19 children with MS, rearrangement of 11q23;t(9;11)(p21-22:23) was found in nine cases and was reported as the most frequent cytogenetic abnormality.⁸ Heerema-McKenney *et al*⁹ reported that cases with 11q23 translocation have a more aggressive

Table 1 All cases of infants with MS testicular involvement

Age (month)	Site of involvement	Bone marrow	Treatment	References
2	Right	AML	Disease free 12 months after transplant	Walker and Cartwright ⁴
3	Left	Normal	Disease free at 12 months of follow-up	Armstrong <i>et al</i> ⁵
8	Bilateral	Not available	Not available	Park <i>et al</i> ⁶
3	Right	Normal	Died few hours after family decided on supportive treatment	Fonseca <i>et al</i> ²
6	Left	AML	In remission after bone marrow transplant	Tran <i>et al</i> ¹
12	Left	AML	No follow-up	Our patient

AML, acute myeloid leukemia; MS, myeloid sarcoma.

clinical course.⁹ MS is most common with M5 subtype. Histological diagnosis of testicular MS is variable ranging from no differentiation to well-differentiated MS. MS cases have also been reported to be misdiagnosed for rhabdomyosarcoma or plasmacytoma.¹⁰

Shaïda and Berman¹¹ reported that percutaneous testicular biopsy is useful in situations where ultrasound findings are equivocal, where there is a discrepancy between clinical and radiological findings, where orchidectomy would be undesirable, or in cases of atrophic testis.

Management of testicular MS is controversial. But it generally involves radical orchiectomy with systemic chemotherapy. Many patients also receive local radiation therapy, followed by consolidation chemotherapy, or bone marrow transplant.¹⁰

Yamauchi and Yasuda¹² reported that all cases of MS should be treated with intensive chemotherapy, even those children who are already treated with surgery or radiotherapy and appear to be cured. Also many children suffered systemic or local relapse if they had not received systemic chemotherapy. Any delay in treatment showed unfavourable outcomes with increased rate of relapse and reduction in overall survival rate.³

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Competing interests None declared.

Patient consent Obtained.

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Learning points

- ▶ Although rare, myeloid sarcoma should be considered in the differential diagnosis of a child with isolated testicular swelling.
- ▶ Blood for peripheral smear examination is warranted in all cases of testicular swelling.
- ▶ Confirmation of the diagnosis of myeloid sarcoma should be done by immunohistochemistry.

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