Rare disease

Genital ulceration in a 4 year old – a case of safeguarding? From social services to pathology...

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Summary

This case of genital ulceration and social concerns in a preschool aged child was highly suspicious for child sexual abuse. However, the lesions presumed to be herpes simplex did not respond to antiviral medication. Specialist input from paediatric oncology and endocrinology resulted in a rare diagnosis of langerhans cell histiocytosis. Furthermore, the complication of diabetes insipidus later developed, making this the youngest child described to our knowledge with vulval lesions of langerhans cell histiocytosis and a central nervous system complication.

BACKGROUND

Langerhans cell histiocytosis (LCH) is an extremely rare multi-factorial condition, with even fewer cases of vulval LCH with central nervous system involvement reported in children. Vulval lesions presenting in preschool children are highly alarming for safeguarding issues and this case is an interesting representation of the multi-disciplinary approach required by today's modern doctor. The sensitive story described encompasses communication with families, co-operation with social services and demonstrable pathology in order to reach a timely diagnosis and management plan. Reporting on rare diagnoses such as this is important to promote awareness of pathology outside of our common clinical knowledge, as diagnostic challenges frequently lead to delays in treatment.

CASE PRESENTATION

A 4-year-old child was admitted to the paediatric ward with bilateral vulval raised vesicular lesions, with surrounding erythema (figure 1). An alarming, provisional clinical diagnosis of herpes simplex with superadded bacterial infection was made and swabs – both bacterial and viral – were taken. Some of the lesions left deep ulcerations on healing. On retrospective questioning of the social history, the parents volunteered that they had recently re-located and that there was a significant family history of learning difficulties. Moreover, the family was known to social services, both for neglect and possible child sexual abuse (CSA) concerns in mothers' childhood. Careful discussions with the family included mode of transmission of herpes, including the possibility of sexual transmission. They were informed that there would be a referral to social services. The unusual nature warranted a referral to dermatology and gynaecology services, which advised excision biopsy of these lesions. Treatment with antiviral medication did not result in any significant improvement.

Figure 1 Vulval ulcerating lesions.

INVESTIGATIONS

Swabs and PCR for herpes simplex virology were negative. Biopsy of the lesion included histological features, an eosinophil-rich infiltrate admixed with histiocytes containing irregular nuclei with prominent folds together with occasional multi-nucleate forms. Immunocytochemical stains showed positive staining with both S100 and CD1a in the mononucleated histiocytic and multi-nucleate forms (figure 2). A diagnosis of LCH was made. Staging investigations, including a skeletal survey did not highlight any further disease process at the time of diagnosis. A subsequent MRI head was reported normal.

DIFFERENTIAL DIAGNOSIS

The admitting paediatric team considered differential diagnoses of herpes simplex, candida and bullous dermatosis.

Figure 2 CD1a positive staining confirming langerhans cell histiocytosis.

further suggested differentials from gynaecology and dermatology included ulcerative lichen planus, Behçet's disease and juvenile bullous disease.

TREATMENT

Our patient was treated with steroids (hydrocortisone 1%) locally and has been started on the multisystem LCH protocol with weekly vinblastine and steroids.

OUTCOME AND FOLLOW-UP

While under regular follow-up, our patient developed polydipsia and drinking upto 3 litres of water per day. Early morning urine osmolality was 83 mosmol/kg with a serum osmolality of 312 mosmol/kg. A water deprivation test was not done due to obvious symptoms. An MRI brain performed was normal with a normal pituitary stalk. A short synacthen test was normal for other hormones showing normal adrenocorticotrophic hormone and cortisol. Desmopressin to treat diabetes insipidus has therefore been added to her treatment regime.

DISCUSSION

LCH is described as a heterogeneous clinical entity, with proliferation of clonal dendritic cells derived from bone marrow. These antigen presenting cells are found within the dermis, but the cause of their proliferation remains unknown.¹ Pan *et al* suggest that the aetiology is multifactorial, including viral triggers, genetics and immune-mediated inflammatory interaction.^{2–5} LCH is rare and predominantly a disease of childhood, with one paper

reporting estimates of 3–5 cases/million children.⁶ Affected organs in children include bone, skin, lymph node, lung and the central nervous system. Disease categories can be divided into single-system and systemic LCH, with single system divided further into uni- or multi-focal involvement. Systemic involvement includes haematopoietic system, liver and spleen.⁷ Furthermore, central nervous system involvement is well documented and is most commonly diabetes insipidus, which occurs in almost a quarter of patients.⁸

To our knowledge, this is the only case of such a young child described with vulval LCH and central nervous system involvement. Due to the heterogeneity of LCH, treatment and prognosis are highly variable. Treatment options in previous cases have included steroids, chemotherapy, radiotherapy and surgery to various degrees of success.⁶

Learning points

- ▶ Although pattern recognition is a core clinical skill, we must also consider rare diagnoses outside of our common clinical knowledge.
- ▶ Issues around safeguarding children should always be considered and coordinating relevant services through information sharing is essential.
- ▶ Careful communication with families is pertinent to optimise clinical care in similar cases.
- ▶ Where swabs are negative, histological diagnosis is a gold standard.

Competing interests None.

Patient consent Obtained.

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