Extramedullary haematopoiesis resembling a blueberry muffin, in a neonate

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DESCRIPTION

A 5 h old baby girl born prematurely presented with fever and rash all over the body since birth. Her mother had a history of fever without rash for 1 week during the second trimester. On physical examination, the baby had microcephaly, symmetrical intrauterine growth retardation, rash all over the body, palpable orbital swelling surrounded by ecchymosis (figure 1), jaundice and hepatosplenomegaly. The rash consisted of bluish-red, firm non-blanching macules and papules 0.5–1.0 cm in diameter over the face (figure 1) and entire body (figure 2).

Laboratory investigations revealed a haemoglobin level of 8.4 g/dL, white cell count of 44 000 cells/mm³ with lymphocytosis (86%), platelet count of 34 000 cells/mm³ and conjugated hyperbilirubinaemia (total bilirubin 17.5 mg/dL, direct bilirubin 5.5 mg/dL). A peripheral smear showed features of disseminated intravascular haemolysis and no

evidence of blast cells. The baby's coagulation profile was deranged (prothrombin time 36 s (control 14), international normalised ratio (INR)

Figure 2 Bluish-red, firm non-blanching maculopapular rashes over the body.

Figure 1 Orbital swelling surrounded by ecchymosis and bluish-red, firm non-blanching maculopapular rash over the face.

Learning points

1. Detailed evaluation of a baby with blueberry muffin lesions is needed since it is an initial presenting feature of various aetiologies.
2. Extramedullary haematopoiesis should be differentiated from chloroma, which is usually isolated.
3. Chloromas presenting as periorbital swelling (raccoon eyes) are more commonly seen with neuroblastoma, and very infrequently in acute lymphoblastic lymphoma and acute myeloid leukaemia.³

Table 1 Differential diagnosis for blueberry muffin lesions

Infections	(Congenital rubella infection, toxoplasmosis, herpes virus, parvovirus, coxsackievirus, Epstein-Barr virus, syphilis) ²
Haematological	Hereditary spherocytosis, fetomaternal transfusion, alloimmunisation, (Rh incompatibility, haemolysis due to anti-Kp (a)), treatment with recombinant erythropoietin, transient neonatal myeloproliferative disorder
Malignancies	Congenital leukaemia, Extraosseal Ewing sarcoma, alveolar rhabdomyosarcoma, neuroblastoma
Systemic	Langerhans cell histiocytosis, Hashimoto-Pritzker disease (congenital self-healing reticulohistiocytosis), lupus
Vascular malformation	Haemangiomas
Endocrine	Congenital transient neonatal hyperparathyroidism
Lysosomal storage disorder	Mucopolidosis type II (I-cell disease)
Neurodevelopmental disorder	Aicardi-Goutières syndrome

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2.5, partial thromboplastin time 102.3 s (29.5–42.2)). Results of cytomegalovirus IgM (2.83 IU/mL) and IgG (174 IU/mL) tests were positive. Despite intensive treatment, the child did not survive.

Blueberry muffin lesion occurs due to dermal extramedullary haematopoiesis.¹ The most common cause is congenital cytomegalovirus infection. Table 1 below shows the differential diagnosis of blueberry muffin lesions.

Contributors BK prepared the manuscript draft and managed the case. SV prepared the pictures. RR managed the case and reviewed the manuscript and reviewed the manuscript. RS reviewed the manuscript.

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