BLUEBERRY MUFFIN BABY

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Objectives

- 2 clinical examples
- Definition
- Etiology
- Pathology
Case # 1

• Baby born at term after an uneventful pregnancy
• At birth, multiples ecchymotic lesions face / nose / groin
• Hepatomegaly with hepatitis and cholestasis
• ↑ WBC 35.7 (lymphocytes) ↓ Platelets 99
• Infectious Contact: Brother had hand-foot-mouth a few weeks before childbirth
Case # 1
Case # 1

Seen at day 2 of life:
Multiple violaceous ecchymosis on the face
Bluish macules in the torso
Case #2

- Baby born at 36 weeks 4/7 after a normal pregnancy without particularity. Serology N.
- Vesicular and papular lesions J # 1 of life
Case #2
Case #2
Case #2
Case #2

(~ 30 very polymorphous scattered Papules inguinal area / head / scalp / limb / trunk

Hemorrhagic Papules <5 mm infiltrated purplish, some crusted

Cutaneous biopsy performed
Blueberry muffin syndrome

- **Definition**: Clinical presentation consisting of disseminated erythematous to violaceous papules and/or nodules in a neonate.

- Classically corresponds to extramedullary foci of hematopoiesis.
### Table 1  Differential Diagnosis for Blueberry Muffin Baby

<table>
<thead>
<tr>
<th>Infectious diseases</th>
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<tbody>
<tr>
<td>Congenital Toxoplasmosis</td>
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<tr>
<td>Congenital Syphilis</td>
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<td>Congenital rubella</td>
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<td>Congenital cytomegalovirus</td>
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<td>Hepatitis B</td>
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<td>Herpes Simplex</td>
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<td>Listeriosis</td>
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<table>
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<th>Haematologic</th>
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<tr>
<td>Immune processes: Rh incompatibility; ABO blood group incompatibility</td>
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<td>Non-immune processes: Alpha thalassaemia gene deletions; Intrauterine parvovirus infection; Congenital dyserythropoietic anemia; Diamond Blackfan anemia; Intrauterine or birth-related hypoxia; Twin-to-twin transfusion; Hereditary spherocytosis</td>
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<td>Malignant/proliferative state: Neuroblastoma; Congenital rhabdomyosarcoma</td>
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<td>Histioctoses: Langerhans cell histiocytosis; Haemophagoctytic lymphohistiocytosis; Juvenile xanthogranuloma; Neonatal leukaemia</td>
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<td>Other: Neonatal lupus erythematosus</td>
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Histology

• Extramedullary hematopoiesis: Dermal infiltrate of immature red blood cells, white blood cell precursors and megakaryocytes centered on the vessels
Leukemia cutis: atypical mononuclear cell infiltrate: Dermal infiltration, adnexal structures and subcutaneous adipose tissue by atypical cells
Stain: TDT +, CD79a + and rare CD10 +
Case #1

US abdomen: N homogeneous liver
Testicular US: Important bilateral hydrocele. Heterogeneous aspect of the 2 testes
US of a lesion of the nose: Small dermal hematoma
Case #1

- Infectio: Hemoc-, CMV-, HSV-, Enterovirus-, EBV-, viral cultures-
- Normalization of leukocytosis and thrombocytopenia. Blood smear N
- PET-Scan 15/07: N except mild liver hyperplasia compatible with hepatitis
Case #1

• Episode of priapism
• Penile and testicular US: Penis N. Progression of testicular infiltration
• US abdo: Hepatomegaly heterogeneous aspect. Multiple hypoechoic lesions infiltrative of the lobe G
• Repeated blood smear: N
Case #1

• Bilateral submandibular adenopathies new
• Recurrence of hyperleucocytosis 41 with smear abnormal with blasts
• Bone marrow biopsy confirmed diagnosis LAL
Case #1

- The initial cutaneous biopsy was amended:
- Skin infiltrated by corresponding blast cells corresponding acute lymphoblastic leukemia pre-B
Skin biopsy

- Important dermal infiltrate of CD1a + and CD207 + cells
Case #2

• Infectious assessment: CMV-, Toxo-, HSV-

• PET-Scan 27/06: Lytic lesions in the skull (parietal and supra-orbital G). Involvement of cervical, paratracheal, abdominal and inguinal ganglia. Pulmonary involvement. Spinal Cord Injury

• Dx: Multisystemic Langerhans Histiocytosis
• Bx inguinal ganglion: CD1a + and CD207 +
• Treatment Vincristine and prednisone
Case #2

• PET-Scan No active disease
• Follow-up in the outpatient clinic:
• Resolution of cutaneous histiocytosis
References:
