Leukocyte adhesion defect type 1: An uncommon immunodeficiency

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Introduction:

Leukocyte adhesion deficiency type 1 (LAD-1) is a rare autosomal recessive disorder of neutrophil function, caused by mutations in the $\beta 2$ integrin gene, characterized by recurrent bacterial infections.

Background and aims:

We describe here two cases in infants who presented with recurrent infections, delayed separation of umbilical cord and marked leucocytosis.

Case 1 : A four months old girl, child of consanguineous parents, hospitalised for fever.

- omphalitis and bacterial meningitis in neonatal period.
- One of his siblings died in infancy due to recurrent infections.
- The liver was palpable 8 cm below the costal margin. The spleen was palpable 6 cm along its long axis below costal margin.
- Investigations: markedly raised leukocyte count ($112.800/\text{mm}^3$).
- Clinical presentation + family history + flow cytometric analysis → diagnosis of LAD 1 type.
- Bone marrow transplant has not been done, due to non-availability of donor. She had multiple episodes of pyrexia, that required multiple hospitalizations.
- The patient succumbed to disseminating infection and died at the age of six.

Case 2: A 47 days -old girl, child of non-consanguineous parents

- had history of bacterial meningitis, omphalitis 5 days after birth and his umbilical cord had been separated 16 days after birth.
- Hospitalized for fever and umbilical swelling, leaving pus whose culture isolated a pseudomonas.
- On examination, the spleen was palpable 4 cm along its long axis below costal margin.
- Laboratory data: marked leukocytosis ($70.300/\text{mm}^3$).
- Her serum immunoglobulin profile and flow cytometric analysis revealed diagnosis of LAD 1 type.
- The child was managed with antibiotics and discharged on trimethoprim/sulfamethoxazole and fluconazole prophylaxis.
- She is waiting for bone marrow transplant.

discussion:

- ✓ Only about 3000 cases have been reported worldwide.
- ✓ Autosomal recessive disorder.

- ✓ Clinical features:
 - Delayed detachment of umbilical cord
 - Omphalitis
 - Impaired wound healing
 - Marked leukocytosis
 - Susceptibility to recurrent bacterial and fungal infections.

- ✓ Evolution ⚡
 - septicemia
 - generalized edeme
 - renal failure
 - death

- ✓ Treatment: stem-cell or bone marrow transplantation.

Conclusion:

The rarity of this disease requires that physicians have a high index of suspicion in a child with history of delayed umbilical cord separation, repeated infections and marked leukocytosis, even in the absence of infections. The diagnosis can be established by flow cytometric analysis of neutrophils surface integrins. Bone marrow transplantation is current options in treatment.