3 Clinical Features
to Consider a Diagnosis of
Leukocyte Adhesion Deficiency-I (LAD-I)

1. History of umbilical cord complications
    - Delayed cord separation or
    - Omphalitis (infection)

2. Higher than normal neutrophil count (Granulocytosis)

3. Bacterial or fungal infection early in life
    <u>Including</u>: pneumonia or other respiratory tract, sepsis, otitis media, oral infections, perianal skin infections, necrotic skin ulcers and others

> ▸ LAD-I can be diagnosed by flow-cytometry of peripheral blood neutrophils for CD18 (and CD11) expression
>
> ▸ Genetic testing for *ITGB2* mutation is confirmatory.

▸ Severe LAD-I is characterized by neutrophil CD18 expression <2% of normal, and is frequently fatal in the first 2 years of life.

▸ Stem cell (bone marrow) transplant may be curative.

▸ Gene therapy trials for severe LAD-I are underway in the US & EU.

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