

Patient Five Details

Clinical findings-

General appearance: Pale, undernourished child

No icterus, cyanosis, clubbing, edema.

Skin: Partial albinism

Hair: Silvery-grey hair

Eyes: Photophobia, nystagmus

Lymph nodes: Mild generalized lymphadenopathy

Hepatosplenomegaly: Present

Investigations:

Laboratory Findings

- Hemoglobin: 9.2 g/dL
- Total leukocyte count: 3,200 cells/mm³
- Platelet count: 90,000/mm³
- Peripheral smear:
- Giant cytoplasmic granules in neutrophils, lymphocytes, and monocytes

Bone Marrow Aspiration

- Hypercellular marrow
- Presence of giant granules in myeloid precursor

Genetic Testing

- Mutation in LYST gene.