# Chediak-Higashi syndrome: pathognomonic feature

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A 2-year-old girl presented to us with recurrent infections, hepatosplenomegaly, and photophobia. On examination she had blond hair with a metallic sheen. The blood smear showed giant lysosomes in the white blood cells (figure) and we diagnosed Chediak-Higashi syndrome, a rare autosomal recessive disease (gene *CHS1/LYST* at 1q42.1-2). There have been around 200 cases reported, and giant cytoplasmic granules are pathognomonic. Death often occurs before the age of 7 years because of the so-called accelerated phase, with hepatosplenomegaly, lymphadenopathy, and pancytopenia or severe recurrent bacterial infections.

The laboratory diagnosis was supported by partial oculocutaneous albinism and typical microscopic granules in hair shafts.

A new mutation was identified and our patient was found to be homozygous for a sequence change in the LYST gene: p.G3583R:c.10747G>. She currently waits for a compatible haemopoietic cell transplantation donor. Her parents and brother are not HLA-identical.

Our patient is now 7 years old, and is well, without serious infections, and with normal neurological function, which might be related to a milder clinical phenotype.

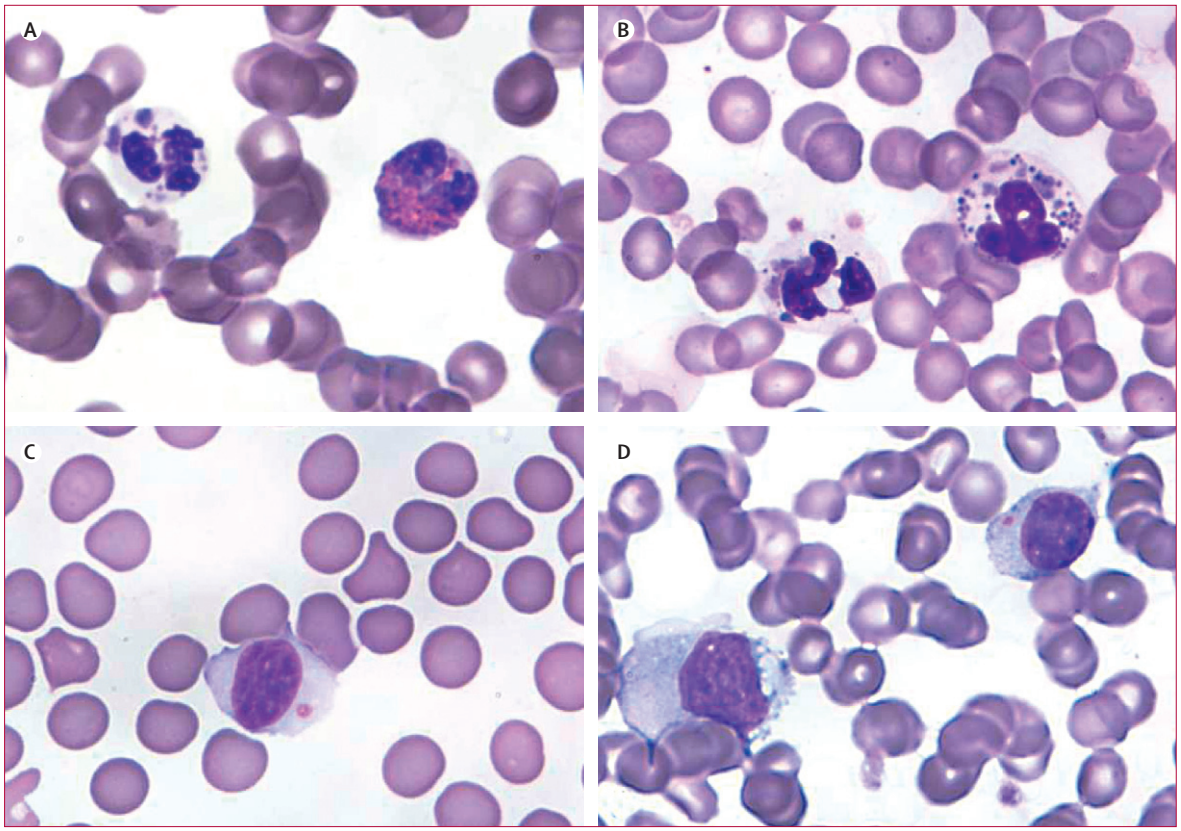


***Figure:*** **Chediak-Higashi syndrome**
Blood film showing pathognomonic giant cytoplasmic granules in neutrophils and eosinophils (A), neutrophils (B), and lymphocytes (C, D).

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