



GENETICS AND GENODERMATOSES

## CHEDIAK HIGASHI SYNDROME - CASE REPORT

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**Background:** Chediak Higashi syndrome is a rare autosomal recessive multisystem disorder characterized by partial ocular and cutaneous albinism, immunodeficiency, hematologic disorder, recurrent infections. The presence of large lysosomal-like organelles in granule containing cells are diagnostic.

**Case:** A one and half year boy presented with recurrent respiratory and systemic complaints. On examination, he had silvery grey hair, depigmentation over the face, hands and feet and hepatosplenomegaly. Complete blood count and peripheral blood smear showed anemia, thrombocytopenia, neutropenia and lymphocytosis. Bone marrow aspirate confirmed 'giant inclusion bodies' in cytoplasm of granulocytes. Histopathological examination of skin biopsy with H&E staining revealed sparse melanin pigment in keratinocytes in lower half of epidermis and giant melanocytes. Hair shaft microscopy showed evenly distributed large melanin granules.

**Discussion:** The presence of abnormal giant intracytoplasmic granules in neutrophils and their precursors are diagnostic of CHS. Bone marrow transplantation therapy is the only promising therapy, that too if given before the onset of the accelerated phase. This emphasizes the need for early identification of the disease by careful examination of the peripheral smear by an experienced hematopathologist, so that bone marrow transplantation can be suggested at the earliest.

