GENETICS AND GENODERMATOSES

WHITE TODAY, WHITE TOMORROW, WHITE FOREVER: AN UNRELATED PARTIAL ALBINISM

Sujana Lakkireddy Gari [(1)]

Santosh Hospital, Skin And Vd, Ghaziabad, India [(1)]

Background: Skin and hair colour are important visible sociocultural characteristics of an individual, and any deviation in the pigmentation is a significant concern. Genetic disorders leading to pigment alteration are more common in pediatric age group and heighten the parents concern. Griscelli syndrome (GS) is a rare autosomal recessive disorder characterized by pigment dilution of skin, hair and immunodeficiency. Worldwide 60 cases have been reported. There are three types of GS. Type 3 GS, only3 cases have been reported in India with pigment dilution of skin and hair without neurological and immunodeficiency.

Observation: A 7yr old boy presented with complaints of white patches over abdomen since 5 months. On further evaluation, the child was born of non consanguineous marriage with silvery white hair all over the body by birth including scalp, eye brow and eye lashes. Developmental milestones and growth indices were normal. Systemic examination was unremarkable. No neurological abnormality or immune system problems. Hair microscopy showed small and large irregular clumps of melanin. Further tests couldn't be done due to financial constraints. Diagnosis of GS type 3was made on bases of clinical examination and hair shaft microscopy. Type 3 GS is characterized by partial albinism due to mutation of melanophilin which forms a protein complex which in turn helps in transfer of melanosomes from center to periphery of a melanocyte.

Key message: Focus on rare syndrome by cost effective simple hair microscopy.