Hypohidrotic Ectodermal Dysplasia with Immune Deficiency

National Cancer Institute

Source

National Cancer Institute. Hypohidrotic Ectodermal Dysplasia with Immune Deficiency. NCI Thesaurus. Code C118844.

A rare disorder caused by mutations either in the IKBKG gene resulting in an X-linked recessive inheritance pattern or in the NFKBIA gene resulting in an autosomal dominant inheritance pattern. It is characterized by abnormal development of ectodermal tissues including the skin, hair, teeth, and sweat glands and immune system deficiency. It results in dry and wrinkled skin, sparse scalp and body hair, missing teeth, and reduced ability to sweat. Patients have abnormally low levels of antibodies causing inability to fight infections.