Mulvihill-Smith syndrome is an extremely rare disorder characterized by low birth weight; growth delays leading to short stature (dwarfism); and/or a prematurely aged facial appearance. Other findings may include additional abnormalities of the head and facial (craniofacial) areas, multiple deeply-colored skin lesions (pigmented nevi), hearing impairment, and/or mental retardation. Eventually, some affected individuals may develop diminished capabilities to resist and fight off repeated infections (primary immunodeficiency). The range and severity of symptoms varies from case to case. All reported cases of Mulvihill-Smith syndrome have occurred as isolated cases. It is possible that this condition is due to a new dominant gene mutation. Mulvihill-Smith syndrome is an extremely rare disorder that, in theory, affects males and females in equal numbers. Most of the reported cases, however, have been males. Only seven cases have been reported. The diagnosis of Mulvihill-Smith Syndrome may be suspected upon the identification of characteristic physical features and findings (e.g., low birth weight, lack of subcutaneous fat in the face, etc.). A diagnosis may be confirmed based upon a thorough clinical evaluation, a detailed patient history, and a variety of specialized tests. For example, hearing tests may be performed to determine the range and severity of hearing impairment in each individual. Pigmented nevi may be present at birth (congenital) or shortly after birth. Other characteristic findings (e.g., short stature) may not be apparent until a child is older.