De Sanctis-Cacchione syndrome is an extremely rare disorder characterized by the skin and eye symptoms of xeroderma pigmentosum (XP) occurring in association with neurological abnormalities, mental retardation, unusually short stature (dwarfism), and underdevelopment of the testes or ovaries (hypogonadism). Xeroderma pigmentosum is a group of rare inherited skin disorders characterized by a heightened reaction to ultraviolet light (photosensitivity), skin discolorations, and the possible development of several types of eye disorders and skin cancers. The most common neurological abnormalities associated with De Sanctis-Cacchione syndrome are low intelligence, an abnormally small head (microcephaly), the loss of ability to coordinate voluntary movement (ataxia), and/or absent (areflexia) or weakened (hyporeflexia) reflexes. De Sanctis-Cacchione syndrome is inherited as an autosomal recessive trait. De Sanctis-Cacchione syndrome is an extremely rare disorder that affects males and females in equal numbers. Although about 200 cases have been reported in Western medical literature, the exact number of cases of this disorder is not known. Onset of symptoms usually occurs during the first year of life, but in rare cases may appear during early or late childhood. Onset of some neurological symptoms may occur as late as five to 10 years of age or even in the second decade of life. De Sanctis-Cacchione syndrome was first described in the medical literature in 1932.