Laurence-Moon Syndrome

National Cancer Institute

Source


A rare genetic syndrome with an autosomal recessive pattern of inheritance. Mutations in the BB55 and MKKS genes of chromosome 11 have been observed in some cases which has lead to comparisons with Bardet-Biedl syndrome and McKusick-Kaufman syndrome. These syndromes have similar etiologies and presentations but are considered separate entities. Clinical signs of Laurence-Moon syndrome include ataxia, intellectual delay, retinitis pigmentosa and hypogonadism. The clinical course follows a progression to growth retardation, spastic paraplegia and optic atrophy with eventual vision loss.