

Neu-Laxova syndrome (NLS) is a rare genetic disorder that is inherited as an autosomal recessive trait. The syndrome is characterized by severe growth delays before birth (intrauterine growth retardation); low birth weight and length; and distinctive abnormalities of the head and facial (craniofacial) region. These may include marked smallness of the head (microcephaly), sloping of the forehead, widely spaced eyes (ocular hypertelorism), and other malformations, resulting in a distinctive facial appearance. NLS is also typically characterized by abnormal accumulations of fluid in tissues throughout the body (generalized edema); permanent flexion and immobilization of multiple joints (flexion contractures); other limb malformations; and/or abnormalities of the brain, skin, genitals, kidneys, and/or heart. NLS appears to affect males and females in relatively equal numbers. Since the disorder was originally described in three siblings in 1971 (Neu, RL) as well as three siblings in another family in 1972 (Laxova, R), over 30 additional cases have been reported. Investigators suggest that the disorder may have a higher frequency in Pakistanis than in other geographic or ethnic populations.