Winchester syndrome is an extremely rare connective tissue disorder believed by some scientists to be closely related to the mucopolysaccharidoses, which is a group of hereditary metabolic diseases caused by the absence or malfunction of certain enzymes, leading to the accumulation in cells and tissues of molecules that would normally be broken down into smaller units. This syndrome is characterized by short stature, arthritis-like symptoms, nodules under the skin (subcutaneous), coarse facial features, and eye and teeth abnormalities. Winchester syndrome is believed to be inherited as an autosomal recessive trait. Since the original description of this syndrome in 1969, only about a dozen cases have been described in the medical literature. Those identified with the syndrome to date have included individuals of Mexican, Indian, Puerto Rican and Iranian descent. Additional affected individuals may be undiagnosed or misdiagnosed. Diagnostic criteria for Winchester syndrome were developed to include characteristic skeletal radiologic findings plus at least two of the following signs: short stature, progressive fusion (contractures) of the joints, cataracts, coarse facial features, darkened patches of skin (hyperpigmentation), and the growth of hair in the leathery patches of skin.