Sotos Syndrome

National Cancer Institute

Source

National Cancer Institute. Sotos Syndrome. NCI Thesaurus. Code C75019.

An autosomal dominant overgrowth syndrome caused by mutation(s) of the NSD1 or the NFIX gene, encoding H3 lysine-36 and H4 lysine-20 specific histone-lysine N-methyltransferase, and nuclear factor 1 X-type, respectively. The condition is characterized by a disproportionately large and long head with a slightly prominent forehead and pointed chin, hypertelorism, down-slanting eyes, large hands and feet, overgrowth in childhood, and developmental delay.