Coffin-Siris Syndrome

National Cancer Institute

Source

National Cancer Institute. Coffin-Siris Syndrome. NCI Thesaurus. Code C35321.

A rare genetic disorder with an undetermined pattern of inheritance affecting mostly females. Clinical signs at birth include recurrent respiratory infections, poor feeding, hypotonia, joint laxity and characteristic shortened fifth digits with hypoplastic or absent nails and craniofacial appearance: microcephaly, wide nose and lips, sparse scalp hair but thick eyebrows and eyelashes. The clinical course includes developmental delays in motor skills and speech with associated moderate mental retardation.