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Cardiofaciocutaneous Syndrome

National Cancer Institute

Source

National Cancer Institute. *Cardiofaciocutaneous Syndrome*. NCI Thesaurus. Code C84617.

A rare genetic syndrome most often caused by BRAF gene mutations. It is characterized by a distinctive facial appearance (high forehead, short nose, and widely spaced eyes), sparse and brittle hair, skin disorders, heart malformations, mental retardation and developmental delay.