Open Peer Review on Qeios

Cardiofaciocutaneous syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Cardiofaciocutaneous syndrome</u>. ORPHA:1340

Cardiofaciocutaneous (CFC) syndrome is a RASopathy characterized by craniofacial dysmorphology, congenital heart disease, dermatological abnormalities (most commonly hyperkeratotic skin and sparse, curly hair), growth retardation and intellectual disability.