An 8-year-old boy collapsed on a trampoline and could not be resuscitated. The boy was a dizygotic twin born at 36 weeks’ gestational age. Medical history included a chest wall nodule, pigmented ocular lesions, anxiety, mild hypospadias, and idiopathic toe-walking. Left conjunctival biopsy at age 7 years and left caruncular biopsy at age 8 years both showed hyperpigmentation without melanocytic atypia . Minor malar and perioral freckling was observed but not documented with photography; both the father and paternal relatives also had freckling of the face and mouth. There was no history of sudden unexpected death in childhood, sudden cardiac death, or any known genetic condition in relatives within 3 degrees of relation to the child.

Multifocal left ventricular fibrous scars, coronary arterial “recanalized thrombi,” and borderline cardiomegaly (215 g, 89-190 g expected) were reported on the original forensic autopsy. Our secondary review identified embolic cardiac myxoma within the coronary arteries and left ventricular subendocardial replacement fibrosis consistent with healed infarcts likely originating from the aortic valve that had been harvested by an organ procurement team before autopsy.

Exome sequencing on the child-parent trio identified a paternally inherited pathogenic variant in PRKAR1A (c.682C>T, p.R228\*), confirming a diagnosis of CNC. Cascade testing was unavailable in the paternal relatives.