

Table 3 Observed differentiation between Sanjad–Sakati syndrome and Kenny–Caffey syndromes, type 1 and type 2

Feature	Sanjad–Sakati syndrome^a	Kenny–Caffey syndrome, type 1	Kenny–Caffey syndrome, type 2
Craniofacial	Microcephaly; micrognathia; deep-set eyes; long philtrum; posteriorly rotated ears	Microcephaly; broad cheeks; hypertelorism; dental caries	Macrocephaly; nanophthalmos; corneal and retinal calcification; congenital cataracts
Skeletal	Delayed bone age; patchy osteosclerosis; small hands and feet	Delayed bone age; poorly ossified skull bones; calvarial osteosclerosis; medullary stenosis of tubular bones; small hands and feet	Osteosclerosis; thickened cortex and narrow marrow cavities of long bones
Mental	Mental retardation (mild to moderate)	Mental retardation/normal mentality	Normal mentality
Other	Micropenis, cryptorchidism, ventricular dilatation	–	–
Laboratory findings	Hypocalcaemia; low parathyroid hormone; hyperphosphataemia; normal cell mediated immunity	Hypocalcaemia low parathyroid hormone; low to low-normal magnesium	Transient hypocalcaemia; low parathyroid hormone and iodine; transient hypophosphataemia; deficient T-cell function
Molecular pathology	Mutation in the tubulin-specific chaperone E gene (TCBE)	Mutation in the tubulin-specific chaperone E gene (TCBE)	Unknown
Inheritance	Autosomal recessive	Autosomal recessive	Autosomal dominant/X-linked

^a*Hypoparathyroidism–retardation–dysmorphism syndrome.*