

[Open Peer Review on Qeios](#)

Thumb deformity-alopecia-pigmentation anomaly syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Thumb deformity-alopecia-pigmentation anomaly syndrome. ORPHA:2251*

Thumb deformity-alopecia-pigmentation anomaly syndrome is a rare, genetic, congenital limb malformation syndrome characterized by short stature, sparse scalp hair, hypoplastic, proximally-placed thumbs, and skin hyperpigmentation with areas of 'raindrop' depigmentation. Presence of a single, upper central incisor has also been reported. There have been no further descriptions in the literature since 1988.