Open Peer Review on Qeios

Exostoses-anetodermia-brachydactyly type E syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Exostoses-anetodermia-brachydactyly type E syndrome. ORPHA:1962

Exostoses-anetodermia-brachydactyly type E syndrome is an association reported in a single kindred characterized by the variable presence of the following features: anetodermia (macular atrophy of the skin), multiple exostoses, and brachydactyly type E (see this term). There have been no further descriptions in the literature since 1985.