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MEDNIK syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. MEDNIK syndrome. ORPHA:171851

MEDNIK syndrome, previously known as Erythrokeratoderma Variabilis type 3 (EKV3), is characterized by intellectual deficit, enteropathy, sensorineural hearing loss, peripheral neuropathy, lamellar and erythrodermic ichthyosis, and keratoderma (MEDNIK stands for Mental retardation, Enteropathy, Deafness, peripheral Neuropathy, Ichthyosis, Keratoderma).