Biotinidase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Biotinidase deficiency. ORPHA:79241

Biotinidase deficiency is a late-onset form of multiple carboxylase deficiency (see this term), an inborn error of biotin metabolism that, if untreated, is characterized by seizures, breathing difficulties, hypotonia, skin rash, alopecia, hearing loss and delayed development.