Adrenomyodystrophy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Adrenomyodystrophy. ORPHA:977

Adrenomyodystrophy is an extremely rare genetic endocrine disease characterized by primary adrenal insufficiency, dystrophic myopathy, hepatic steatosis, severe psychomotor delay, megalocornea, failure to thrive, chronic constipation, and terminal bladder ectasia which can lead to death. There have been no further descriptions in the literature since 1982.