Hyperprolinemia type 2

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperprolinemia type 2. ORPHA:79101

Hyperprolinemia type 2 is an autosomal recessive proline metabolism disorder due to pyroline-5-carboxylate dehydrogenase deficiency. The condition is often benign but clinical signs may include seizures, intellectual deficit and mild developmental delay.