Cystathioninuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cystathioninuria. ORPHA:212

Cystathioninuria is an autosomal recessive disorder caused by cystathionine gamma-lyase deficiency. It is usually pyridoxine-dependent, but in very rare cases it may be non-dependent. It is generally considered to be a benign condition without pathogenic relevance. However, association of cystathioninuria with intellectual impairment has been reported in several cases.