Abetalipoproteinemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Abetalipoproteinemia. ORPHA:14

Abetalipoproteinemia/ homozygous familial hypobetalipoproteinemia (ABL/HoFHBL) is a severe form of familial hypobetalipoproteinemia (see this term) characterized by permanently low levels (below the 5th percentile) of apolipoprotein B and LDL cholesterol, and by growth delay, malabsorption, hepatomegaly, and neurological and neuromuscular manifestations.