Maternal phenylketonuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Maternal phenylketonuria. ORPHA:2209

Maternal phenylketonuria (PKU) is a rare disorder of phenylalanine metabolism (see this term), an inborn error of amino acid metabolism, characterized by the development of microcephaly, growth retardation, congenital heart disease, facial dysmorphism and intellectual disability in nonphenylketonuric offspring of mothers with excess phenylalanine (Phe) concentrations.