Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pulmonary fibrosis-hepatic hyperplasia-bone marrow hypoplasia syndrome. ORPHA:210136

Pulmonary fibrosis - hepatic hyperplasia - bone marrow hypoplasia, also named trimorphic syndrome (i.e. three (inherited) morbidities, pulmonary, hepatic and cytopenia), is a rare disease reported in 4 cases to date, manifesting with idiopathic pulmonary fibrosis, hepatic nodular regenerative hyperplasia leading to portal hypertension and thrombocytopenia due to bone marrow hypoplasia. The condition was associated with 100% mortality.