

THE MITOCHONDRION, HP\_ABNORMALITY\_OF\_THE MITOCHONDRION

- HP\_INCREASED\_CSF\_LACTATE, HP\_INCREASED\_CSF\_LACTATE
- HP\_LACTIC\_ACIDOSIS, HP\_LACTIC\_ACIDOSIS
- HP\_ABNORMAL\_ACTIVITY\_OF\_MITOCHONDRIAL\_RESPIRATORY\_CHAIN, HP\_ABNORMAL\_ACTIVITY\_OF\_MITOCHONDRIAL\_RESPIRATORY\_CHAIN
- HP\_LEUKODYSTROPHY, HP\_LEUKODYSTROPHY
- HP\_ABNORMAL\_CSF\_METABOLITE\_LEVEL, HP\_ABNORMAL\_CSF\_METABOLITE\_LEVEL
- HP\_OPTIC\_NEUROPATHY, HP\_OPTIC\_NEUROPATHY
- HP\_DECREASED\_ACTIVITY\_OF\_MITOCHONDRIAL\_COMPLEX\_I, HP\_DECREASED\_ACTIVITY\_OF\_MITOCHONDRIAL\_COMPLEX\_I
- HP\_LOWER\_LIMB\_SPASTICITY, HP\_LOWER\_LIMB\_SPASTICITY
- HP\_LETHARGY, HP\_LETHARGY
- HP\_ABNORMALITY\_OF\_HEPATOBIILIARY\_SYSTEM\_PHYSIOLOGY, HP\_ABNORMALITY\_OF\_HEPATOBIILIARY\_SYSTEM\_PHYSIOLOGY
- HP\_ABNORMALITY\_OF\_THE\_FRONTAL\_HAIRLINE, HP\_ABNORMALITY\_OF\_THE\_FRONTAL\_HAIRLINE
- HP\_FOCAL\_T2\_HYPERINTENSE\_BRAINSTEM\_LESION, HP\_FOCAL\_T2\_HYPERINTENSE\_BRAINSTEM\_LESION
- HP\_ABNORMAL\_GLIAL\_CELL\_MORPHOLOGY, HP\_ABNORMAL\_GLIAL\_CELL\_MORPHOLOGY
- HP\_OPTIC\_DISC\_PALLOR, HP\_OPTIC\_DISC\_PALLOR
- HP\_SPASTIC\_PARAPLEGIA, HP\_SPASTIC\_PARAPLEGIA
- HP\_INFERTILITY, HP\_INFERTILITY
- HP\_GLIOSIS, HP\_GLIOSIS
- HP\_ABNORMAL\_BRAINSTEM\_MRI\_SIGNAL\_INTENSITY, HP\_ABNORMAL\_BRAINSTEM\_MRI\_SIGNAL\_INTENSITY
- HP\_ABNORMAL\_URINE\_PH, HP\_ABNORMAL\_URINE\_PH
- HP\_NEURODEGENERATION, HP\_NEURODEGENERATION
- HP\_ARTERIAL\_STENOSIS, HP\_ARTERIAL\_STENOSIS
- HP\_ABNORMAL\_PATELLA\_MORPHOLOGY, HP\_ABNORMAL\_PATELLA\_MORPHOLOGY
- HP\_ABNORMAL\_AORTIC\_VALVE\_PHYSIOLOGY, HP\_ABNORMAL\_AORTIC\_VALVE\_PHYSIOLOGY
- HP\_PROGRESSIVE\_NEUROLOGIC\_DETERIORATION, HP\_PROGRESSIVE\_NEUROLOGIC\_DETERIORATION
- HP\_BRUISING\_SUSCEPTIBILITY, HP\_BRUISING\_SUSCEPTIBILITY
- HP\_ABNORMALITY\_OF\_HEAD\_BLOOD\_VESSEL, HP\_ABNORMALITY\_OF\_HEAD\_BLOOD\_VESSEL
- HP\_NEOPLASM\_OF\_THE\_PANCREAS, HP\_NEOPLASM\_OF\_THE\_PANCREAS
- HP\_MULTIFACTORIAL\_INHERITANCE, HP\_MULTIFACTORIAL\_INHERITANCE
- HP\_BRAIN\_IMAGING\_ABNORMALITY, HP\_BRAIN\_IMAGING\_ABNORMALITY
- HP\_RENAL\_TUBULAR\_ACIDOSIS, HP\_RENAL\_TUBULAR\_ACIDOSIS
- HP\_ORGANIC\_ACIDURIA, HP\_ORGANIC\_ACIDURIA
- HP\_CRANIOSYNOSTOSIS, HP\_CRANIOSYNOSTOSIS
- HP\_PSYCHOMOTOR\_RETARDATION, HP\_PSYCHOMOTOR\_RETARDATION
- HP\_HYPOTHERMIA, HP\_HYPOTHERMIA
- HP\_ABNORMAL\_CARDIAC\_VENTRICULAR\_FUNCTION, HP\_ABNORMAL\_CARDIAC\_VENTRICULAR\_FUNCTION
- HP\_WIDELY\_SPACED\_TEETH, HP\_WIDELY\_SPACED\_TEETH
- HP\_ABNORMAL\_RETINAL\_ARTERY\_MORPHOLOGY, HP\_ABNORMAL\_RETINAL\_ARTERY\_MORPHOLOGY
- HP\_LIPID\_ACCUMULATION\_IN\_HEPATOCYTES, HP\_LIPID\_ACCUMULATION\_IN\_HEPATOCYTES
- HP\_PELVIC\_GIRDLE\_MUSCLE\_WEAKNESS, HP\_PELVIC\_GIRDLE\_MUSCLE\_WEAKNESS
- HP\_ABNORMAL\_FEMORAL\_NECK\_HEAD\_MORPHOLOGY, HP\_ABNORMAL\_FEMORAL\_NECK\_HEAD\_MORPHOLOGY
- HP\_MITOCHONDRIAL\_RESPIRATORY\_CHAIN\_DEFECTS, HP\_MITOCHONDRIAL\_RESPIRATORY\_CHAIN\_DEFECTS
- HP\_FOCAL\_T2\_HYPERINTENSE\_BASAL\_GANGLIA\_LESION, HP\_FOCAL\_T2\_HYPERINTENSE\_BASAL\_GANGLIA\_LESION
- HP\_RETINAL\_VASCULAR\_TORTUOSITY, HP\_RETINAL\_VASCULAR\_TORTUOSITY
- HP\_ABNORMAL\_BASAL\_GANGLIA\_MRI\_SIGNAL\_INTENSITY, HP\_ABNORMAL\_BASAL\_GANGLIA\_MRI\_SIGNAL\_INTENSITY
- HP\_ARTERIOSCLEROSIS, HP\_ARTERIOSCLEROSIS
- HP\_METABOLIC\_ACIDOSIS, HP\_METABOLIC\_ACIDOSIS
- HP\_UPPER\_EXTREMITY\_JOINT\_DISLOCATION, HP\_UPPER\_EXTREMITY\_JOINT\_DISLOCATION
- HP\_INCREASED\_SERUM\_PYRUVATE, HP\_INCREASED\_SERUM\_PYRUVATE
- HP\_HEPATIC\_FAILURE, HP\_HEPATIC\_FAILURE
- HP\_ABNORMAL\_SPEECH\_PROSODY, HP\_ABNORMAL\_SPEECH\_PROSODY
- HP\_WEBBED\_NECK, HP\_WEBBED\_NECK
- HP\_ABNORMAL\_RENAL\_GLOMERULUS\_MORPHOLOGY, HP\_ABNORMAL\_RENAL\_GLOMERULUS\_MORPHOLOGY
- HP\_PROXIMAL\_TUBULOPATHY, HP\_PROXIMAL\_TUBULOPATHY
- HP\_RECURRENT\_SINOPULMONARY\_INFECTIONS, HP\_RECURRENT\_SINOPULMONARY\_INFECTIONS
- HP\_ABNORMAL\_PHARYNX\_MORPHOLOGY, HP\_ABNORMAL\_PHARYNX\_MORPHOLOGY
- HP\_PERSEVERATION, HP\_PERSEVERATION
- HP\_CENTROCECAL\_SCOTOMA, HP\_CENTROCECAL\_SCOTOMA
- HP\_MITOCHONDRIAL\_INHERITANCE, HP\_MITOCHONDRIAL\_INHERITANCE
- HP\_ABNORMAL\_METABOLIC\_BRAIN\_IMAGING\_BY\_MRS, HP\_ABNORMAL\_METABOLIC\_BRAIN\_IMAGING\_BY\_MRS
- HP\_ABNORMAL\_MITOCHONDRIA\_IN\_MUSCLE\_TISSUE, HP\_ABNORMAL\_MITOCHONDRIA\_IN\_MUSCLE\_TISSUE
- HP\_ABNORMAL\_RENAL\_TUBULE\_MORPHOLOGY, HP\_ABNORMAL\_RENAL\_TUBULE\_MORPHOLOGY
- HP\_DECREASED\_ACTIVITY\_OF\_THE\_PYRUVATE\_DEHYDROGENASE\_COMPLEX, HP\_DECREASED\_ACTIVITY\_OF\_THE\_PYRUVATE\_DEHYDROGENASE\_COMPLEX
- HP\_VENTRICULAR\_PREEXCITATION, HP\_VENTRICULAR\_PREEXCITATION
- HP\_RECURRENT\_PAROXYSMAL\_HEADACHE, HP\_RECURRENT\_PAROXYSMAL\_HEADACHE
- HP\_MITOCHONDRIAL\_MYOPATHY, HP\_MITOCHONDRIAL\_MYOPATHY
- HP\_CUTIS\_MARMORATA, HP\_CUTIS\_MARMORATA
- HP\_VASCULAR\_DILATATION, HP\_VASCULAR\_DILATATION
- HP\_AORTIC\_REGURGITATION, HP\_AORTIC\_REGURGITATION
- HP\_LEBER\_OPTIC\_ATROPHY, HP\_LEBER\_OPTIC\_ATROPHY
- HP\_ABNORMALITY\_OF\_THE\_INCISOR, HP\_ABNORMALITY\_OF\_THE\_INCISOR
- HP\_FACIAL\_DIPLEGIA, HP\_FACIAL\_DIPLEGIA
- HP\_JOINT\_HYPERFLEXIBILITY, HP\_JOINT\_HYPERFLEXIBILITY
- HP\_FEMALE\_INFERTILITY, HP\_FEMALE\_INFERTILITY
- HP\_EPISODIC\_RESPIRATORY\_DISTRESS, HP\_EPISODIC\_RESPIRATORY\_DISTRESS
- HP\_ABNORMAL\_URINE\_SODIUM\_CONCENTRATION, HP\_ABNORMAL\_URINE\_SODIUM\_CONCENTRATION
- HP\_TRANSIENT\_ISCHEMIC\_ATTACK, HP\_TRANSIENT\_ISCHEMIC\_ATTACK
- HP\_LOW\_PLASMA\_CITRULLINE, HP\_LOW\_PLASMA\_CITRULLINE
- HP\_ABNORMAL\_URINE\_POTASSIUM\_CONCENTRATION, HP\_ABNORMAL\_URINE\_POTASSIUM\_CONCENTRATION
- HP\_RETINAL\_TELANGIECTASIA, HP\_RETINAL\_TELANGIECTASIA
- HP\_DECREASED\_CIRCULATING\_ANTIBODY\_LEVEL, HP\_DECREASED\_CIRCULATING\_ANTIBODY\_LEVEL
- HP\_BASAL\_GANGLIA\_GLIOSIS, HP\_BASAL\_GANGLIA\_GLIOSIS
- HP\_PREMATURELY\_AGED\_APPEARANCE, HP\_PREMATURELY\_AGED\_APPEARANCE
- HP\_CEREBRAL\_CALCIFICATION, HP\_CEREBRAL\_CALCIFICATION
- HP\_ASTROCYTOSIS, HP\_ASTROCYTOSIS
- HP\_ABNORMALITY\_OF\_THE\_LYMPH\_NODES, HP\_ABNORMALITY\_OF\_THE\_LYMPH\_NODES
- HP\_PROGRESSIVE\_INABILITY\_TO\_WALK, HP\_PROGRESSIVE\_INABILITY\_TO\_WALK
- HP\_DICARBOXYLIC\_ACIDURIA, HP\_DICARBOXYLIC\_ACIDURIA
- HP\_PROGRESSIVE\_EXTERNAL\_OPHTHALMOPLEGIA, HP\_PROGRESSIVE\_EXTERNAL\_OPHTHALMOPLEGIA
- HP\_LACRIMATION\_ABNORMALITY, HP\_LACRIMATION\_ABNORMALITY
- HP\_ABNORMAL\_PHALANGEAL\_JOINT\_MORPHOLOGY\_OF\_THE\_HAND, HP\_ABNORMAL\_PHALANGEAL\_JOINT\_MORPHOLOGY\_OF\_THE\_HAND
- HP\_ABNORMAL\_FEMORAL\_HEAD\_MORPHOLOGY, HP\_ABNORMAL\_FEMORAL\_HEAD\_MORPHOLOGY
- HP\_TRANSPOSITION\_OF\_THE\_GREAT\_ARTERIES, HP\_TRANSPOSITION\_OF\_THE\_GREAT\_ARTERIES
- HP\_ABNORMALITY\_OF\_KREBS\_CYCLE\_METABOLISM, HP\_ABNORMALITY\_OF\_KREBS\_CYCLE\_METABOLISM
- HP\_HAND\_MUSCLE\_WEAKNESS, HP\_HAND\_MUSCLE\_WEAKNESS
- HP\_MACULAR\_PURPURA, HP\_MACULAR\_PURPURA
- HP\_ABNORMAL\_CEREBROSPINAL\_FLUID\_METABOLITE\_CONCENTRATION, HP\_ABNORMAL\_CEREBROSPINAL\_FLUID\_METABOLITE\_CONCENTRATION
- HP\_RECURRENT\_UPPER\_RESPIRATORY\_TRACT\_INFECTIONS, HP\_RECURRENT\_UPPER\_RESPIRATORY\_TRACT\_INFECTIONS
- HP\_BRONCHITIS, HP\_BRONCHITIS
- HP\_SLENDER\_FINGER, HP\_SLENDER\_FINGER
- HP\_FETAL\_DISTRESS, HP\_FETAL\_DISTRESS