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HP_LACTIC_ACIDOSIS, HP_LACTIC_ACIDOSIS
HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN, HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN
HP_ABNORMAL_CSF_METABOLITE_LEVEL, HP_ABNORMAL_CSF_METABOLITE_LEVEL
HP_LEUKODYSTROPHY, HP_LEUKODYSTROPHY
HP_INCREASED_CSF_LACTATE, HP_INCREASED_CSF_LACTATE
HP_ABNORMALITY_OF_THE_MITOCHONDRION, HP_ABNORMALITY_OF_THE_MITOCHONDRION
HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_I, HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_I
HP_LETHARGY, HP_LETHARGY
HP_ABNORMAL_BASAL_GANGLIA_MRI_SIGNAL_INTENSITY, HP_ABNORMAL_BASAL_GANGLIA_MRI_SIGNAL_INTENSITY
HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION, HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION
HP_SLOPING_FOREHEAD, HP_SLOPING_FOREHEAD
HP_HYPERAMMONEMIA, HP_HYPERAMMONEMIA
HP_CAFE_AU_LAIT_SPOT, HP_CAFE_AU_LAIT_SPOT
HP_PIGMENTARY_RETINOPATHY, HP_PIGMENTARY_RETINOPATHY
HP DECREASED ACTIVITY OF THE PYRUVATE DEHYDROGENASE COMPLEX, HP DECREASED ACTIVITY OF THE PYRUVATE DEHYDROGENASE COMPLEX
HP_OPTIC_DISC_PALLOR, HP_OPTIC_DISC_PALLOR
HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION, HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION
HP_INCREASED_SERUM_PYRUVATE, HP_INCREASED_SERUM_PYRUVATE
HP_AMINOACIDURIA, HP_AMINOACIDURIA
HP_INFERTILITY, HP_INFERTILITY
HP_LEBER_OPTIC_ATROPHY, HP_LEBER_OPTIC_ATROPHY
HP_MITOCHONDRIAL_MYOPATHY, HP_MITOCHONDRIAL_MYOPATHY
HP_LIPID_ACCUMULATION_IN_HEPATOCYTES, HP_LIPID_ACCUMULATION_IN_HEPATOCYTES
HP_RETINAL_ARTERIAL_TORTUOSITY, HP_RETINAL_ARTERIAL_TORTUOSITY
HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE, HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE
HP HYPOPIGMENTATION OF THE SKIN, HP HYPOPIGMENTATION OF THE SKIN
HP_ABSENT_SEPTUM_PELLUCIDUM, HP_ABSENT_SEPTUM_PELLUCIDUM
HP_VASCULAR_TORTUOSITY, HP_VASCULAR_TORTUOSITY
HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY, HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY
HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM, HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM
HP_RAGGED_RED_MUSCLE_FIBERS, HP_RAGGED_RED_MUSCLE_FIBERS
HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY, HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY
HP_POSTNATAL_MICROCEPHALY, HP_POSTNATAL_MICROCEPHALY
HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION
HP_CENTROCECAL_SCOTOMA, HP_CENTROCECAL_SCOTOMA
HP_LOWER_LIMB_SPASTICITY, HP_LOWER_LIMB_SPASTICITY
HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION
HP_POOR_HEAD_CONTROL, HP_POOR_HEAD_CONTROL
HP_BLURRED_VISION, HP_BLURRED_VISION
HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL, HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL
HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION, HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION
HP_MALE_INFERTILITY, HP_MALE_INFERTILITY
HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS, HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS
HP_DECREASED_FERTILITY_IN_MALES, HP_DECREASED_FERTILITY_IN_MALES
HP_ABNORMAL_URINE_PH, HP_ABNORMAL_URINE_PH
HP_METABOLIC_ACIDOSIS, HP_METABOLIC_ACIDOSIS
HP_ABNORMALITY_OF_CORNEAL_STROMA, HP_ABNORMALITY_OF_CORNEAL_STROMA
· HP_PROGRESSIVE_SPASTIC_PARAPLEGIA, HP_PROGRESSIVE_SPASTIC_PARAPLEGIA
HP_SHORT_PALPEBRAL_FISSURE, HP_SHORT_PALPEBRAL_FISSURE
HP_MITOCHONDRIAL_INHERITANCE, HP_MITOCHONDRIAL_INHERITANCE
HP_FETAL_DISTRESS, HP_FETAL_DISTRESS
HP PLEURAL EFFUSION, HP PLEURAL EFFUSION
HP_ABNORMAL_LABIA_MORPHOLOGY, HP_ABNORMAL_LABIA_MORPHOLOGY
HP_INTESTINAL_POLYP, HP_INTESTINAL_POLYP
HP_POOR_EYE_CONTACT, HP_POOR_EYE_CONTACT
HP_OPTIC_NEUROPATHY, HP_OPTIC_NEUROPATHY
HP_SPASTIC_TETRAPARESIS, HP_SPASTIC_TETRAPARESIS
HP_ABNORMAL_CIRCULATING_GLUTAMINE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_GLUTAMINE_FAMILY_AMINO_ACID_CONCENTRATION.
HP_3_METHYLGLUTACONIC_ACIDURIA, HP_3_METHYLGLUTACONIC_ACIDURIA
HP_SANDAL_GAP, HP_SANDAL_GAP
HP_ABNORMAL_CIRCULATING_NITROGEN_COMPOUND_CONCENTRATION, HP_ABNORMAL_CIRCULATING_NITROGEN_COMPOUND_CONCENTRATION
HP_ABNORMAL_CIRCULATING_PROLINE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PROLINE_CONCENTRATION
HP_SMALL_BASAL_GANGLIA, HP_SMALL_BASAL_GANGLIA
HP_MYELODYSPLASIA, HP_MYELODYSPLASIA
HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM, HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM
HP_DEATH_IN_INFANCY, HP_DEATH_IN_INFANCY
HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS, HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS
HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION, HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION
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HP\_ABNORMAL\_CIRCULATING\_PROTEINOGENIC\_AMINO\_ACID\_DERIVATIVE\_CONCENTRATION, HP\_ABNORMAL\_CIRCULATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATIVE\_CONCENTRATION\_ACID\_DERIVATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATIVE\_CONCENTRATI

HP\_ABNORMAL\_CIRCULATING\_PYRUVATE\_FAMILY\_AMINO\_ACID\_CONCENTRATION, HP\_ABNORMAL\_CIRCULATING\_PYRUVATE\_FAMILY\_AMINO\_ACID\_CONCENTRATION.

HP\_ABNORMAL\_CIRCULATING\_SERINE\_FAMILY\_AMINO\_ACID\_CONCENTRATION, HP\_ABNORMAL\_CIRCULATING\_SERINE\_FAMILY\_AMINO\_ACID\_CONCENTRATION

## D\_SERUM\_LACTATE, HP\_INCREASED\_SERUM\_LACTATE

HP\_WIDE\_MOUTH, HP\_WIDE\_MOUTH

HP\_HYPOMIMIC\_FACE, HP\_HYPOMIMIC\_FACE

HP\_ECTRODACTYLY, HP\_ECTRODACTYLY

HP\_INFANTILE\_SPASMS, HP\_INFANTILE\_SPASMS

HP\_CHOREOATHETOSIS, HP\_CHOREOATHETOSIS

HP\_EPISODIC\_VOMITING, HP\_EPISODIC\_VOMITING

HP\_ECTOPIC\_ANUS, HP\_ECTOPIC\_ANUS

HP\_SKIN\_PIT, HP\_SKIN\_PIT

HP\_HYPERVENTILATION, HP\_HYPERVENTILATION HP\_ENLARGED\_THORAX, HP\_ENLARGED\_THORAX

HP\_TETRALOGY\_OF\_FALLOT, HP\_TETRALOGY\_OF\_FALLOT

HP\_RECURRENT\_MYCOBACTERIAL\_INFECTIONS, HP\_RECURRENT\_MYCOBACTERIAL\_INFECTIONS

HP\_UNDERDEVELOPED\_SUPRAORBITAL\_RIDGES, HP\_UNDERDEVELOPED\_SUPRAORBITAL\_RIDGES

HP\_ABNORMAL\_C\_REACTIVE\_PROTEIN\_LEVEL, HP\_ABNORMAL\_C\_REACTIVE\_PROTEIN\_LEVEL

HP MICROVESICULAR HEPATIC STEATOSIS, HP MICROVESICULAR HEPATIC STEATOSIS

HP\_DECREASED\_ACTIVITY\_OF\_MITOCHONDRIAL\_COMPLEX\_IV, HP\_DECREASED\_ACTIVITY\_OF\_MITOCHONDRIAL\_COMPLEX\_IV

HP\_ABNORMALITY\_OF\_HEPATOBILIARY\_SYSTEM\_PHYSIOLOGY, HP\_ABNORMALITY\_OF\_HEPATOBILIARY\_SYSTEM\_PHYSIOLOGY

HP\_EPISODIC\_RESPIRATORY\_DISTRESS, HP\_EPISODIC\_RESPIRATORY\_DISTRESS

HP\_PSYCHOMOTOR\_RETARDATION, HP\_PSYCHOMOTOR\_RETARDATION

HP\_GLOBAL\_BRAIN\_ATROPHY, HP\_GLOBAL\_BRAIN\_ATROPHY

HP\_LOW\_PLASMA\_CITRULLINE, HP\_LOW\_PLASMA\_CITRULLINE

HP\_MULTIPLE\_GLOMERULAR\_CYSTS, HP\_MULTIPLE\_GLOMERULAR\_CYSTS
HP\_RENAL\_TUBULAR\_DYSFUNCTION, HP\_RENAL\_TUBULAR\_DYSFUNCTION