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HP_LACTIC_ACIDOSIS, HP_LACTIC_ACIDOSIS
HP ABNORMAL CSF METABOLITE LEVEL, HP ABNORMAL CSF METABOLITE LEVEL
HP_INCREASED_CSF_LACTATE, HP_INCREASED_CSF_LACTATE
HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN, HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN
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_LEUKODYSTROPHY, HP_LEUKODYSTROPHY
HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION, HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION
HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION
HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE, HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE
HP_LETHARGY, HP_LETHARGY
HP_ABNORMAL_BASAL_GANGLIA_MRI_SIGNAL_INTENSITY, HP_ABNORMAL_BASAL_GANGLIA_MRI_SIGNAL_INTENSITY
HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY, HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY
HP ABNORMALITY OF HEPATOBILIARY SYSTEM PHYSIOLOGY, HP ABNORMALITY OF HEPATOBILIARY SYSTEM PHYSIOLOGY
HP_SLOPING_FOREHEAD, HP_SLOPING_FOREHEAD
HP_PIGMENTARY_RETINOPATHY, HP_PIGMENTARY_RETINOPATHY
HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION, HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION
HP_ABNORMAL_GLIAL_CELL_MORPHOLOGY, HP_ABNORMAL_GLIAL_CELL_MORPHOLOGY
HP_ABNORMAL_SOCIAL_BEHAVIOR, HP_ABNORMAL_SOCIAL_BEHAVIOR
HP_FETAL_DISTRESS, HP_FETAL_DISTRESS
HP_INCREASED_SERUM_PYRUVATE, HP_INCREASED_SERUM_PYRUVATE
HP_ABNORMAL_URINE_PH, HP_ABNORMAL_URINE_PH
HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION, HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION
HP_MITOCHONDRIAL_MYOPATHY, HP_MITOCHONDRIAL_MYOPATHY
HP_OPTIC_NEUROPATHY, HP_OPTIC_NEUROPATHY
HP_ABNORMAL_LOCALIZATION_OF_KIDNEY, HP_ABNORMAL_LOCALIZATION_OF_KIDNEY
HP_GINGIVAL_OVERGROWTH, HP_GINGIVAL_OVERGROWTH
HP_SMOOTH_PHILTRUM, HP_SMOOTH_PHILTRUM
HP_DECREASED_ACTIVITY_OF_THE_PYRUVATE_DEHYDROGENASE_COMPLEX, HP_DECREASED_ACTIVITY_OF_THE_PYRUVATE_DEHYDROGENASE_COMPLEX
HP_CAFE_AU_LAIT_SPOT, HP_CAFE_AU_LAIT_SPOT
HP_ABNORMAL_BRAIN_LACTATE_LEVEL_BY_MRS, HP_ABNORMAL_BRAIN_LACTATE_LEVEL_BY_MRS
HP_PROXIMAL_TUBULOPATHY, HP_PROXIMAL_TUBULOPATHY
HP_OPTIC_DISC_PALLOR, HP_OPTIC_DISC_PALLOR
HP_POOR_HEAD_CONTROL, HP_POOR_HEAD_CONTROL
HP_MULTIFACTORIAL_INHERITANCE, HP_MULTIFACTORIAL_INHERITANCE
HP_IMPAIRED_SOCIAL_INTERACTIONS, HP_IMPAIRED_SOCIAL_INTERACTIONS
HP_LIPID_ACCUMULATION_IN_HEPATOCYTES, HP_LIPID_ACCUMULATION_IN_HEPATOCYTES
HP_MYELODYSPLASIA, HP_MYELODYSPLASIA
HP_LOWER_LIMB_SPASTICITY, HP_LOWER_LIMB_SPASTICITY
HP_POOR_EYE_CONTACT, HP_POOR_EYE_CONTACT
HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVATIVE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVATIVE
HP_HEPATIC_FAILURE, HP_HEPATIC_FAILURE
HP_ABNORMALITY_OF_THE_PERIVENTRICULAR_WHITE_MATTER, HP_ABNORMALITY_OF_THE_PERIVENTRICULAR_WHITE_MATTER
HP_DEMYELINATING_PERIPHERAL_NEUROPATHY, HP_DEMYELINATING_PERIPHERAL_NEUROPATHY
HP_HYPOPIGMENTATION_OF_THE_SKIN, HP_HYPOPIGMENTATION_OF_THE_SKIN
HP_ABNORMALITY_OF_THE_GINGIVA, HP_ABNORMALITY_OF_THE_GINGIVA
HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS, HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS
HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM, HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM
HP_GLIOSIS, HP_GLIOSIS
HP DECREASED ACTIVITY OF MITOCHONDRIAL COMPLEX IV, HP DECREASED ACTIVITY OF MITOCHONDRIAL COMPLEX IV
HP_PSYCHOMOTOR_RETARDATION, HP_PSYCHOMOTOR_RETARDATION
HP_IMMUNOLOGIC_HYPERSENSITIVITY, HP_IMMUNOLOGIC_HYPERSENSITIVITY
HP_CLEFT_SOFT_PALATE, HP_CLEFT_SOFT_PALATE
HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY, HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY
HP_SHORT_THUMB, HP_SHORT_THUMB
HP_HORSESHOE_KIDNEY, HP_HORSESHOE_KIDNEY
HP_GLOMERULAR_SCLEROSIS, HP_GLOMERULAR_SCLEROSIS
HP_ABNORMALITY_OF_THE_LYMPH_NODES, HP_ABNORMALITY_OF_THE_LYMPH_NODES
HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION
HP_RETINAL_TELANGIECTASIA, HP_RETINAL_TELANGIECTASIA
HP_PROGRESSIVE_SPASTIC_PARAPLEGIA, HP_PROGRESSIVE_SPASTIC_PARAPLEGIA
HP_APLASIA_HYPOPLASIA_OF_THE_THUMB, HP_APLASIA_HYPOPLASIA_OF_THE_THUMB
HP_RAGGED_RED_MUSCLE_FIBERS, HP_RAGGED_RED_MUSCLE_FIBERS
HP_METABOLIC_ACIDOSIS, HP_METABOLIC_ACIDOSIS
HP_INFERTILITY, HP_INFERTILITY
HP_GLOBAL_BRAIN_ATROPHY, HP_GLOBAL_BRAIN_ATROPHY
HP_ABNORMAL_URETER_PHYSIOLOGY, HP_ABNORMAL_URETER_PHYSIOLOGY
HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL, HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL
HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS, HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS
HP_RETINAL_ARTERIAL_TORTUOSITY, HP_RETINAL_ARTERIAL_TORTUOSITY
HP_EXERCISE_INTOLERANCE, HP_EXERCISE_INTOLERANCE
HP_ECTRODACTYLY, HP_ECTRODACTYLY
HP_SANDAL_GAP, HP_SANDAL_GAP
HP_GASTROINTESTINAL_HEMORRHAGE, HP_GASTROINTESTINAL_HEMORRHAGE
HP_HYPERAMMONEMIA, HP_HYPERAMMONEMIA
HP_ABNORMAL_CIRCULATING_NITROGEN_COMPOUND_CONCENTRATION, HP_ABNORMAL_CIRCULATING_NITROGEN_COMPOUND_CONCENTRATION
HP_ARTHRITIS, HP_ARTHRITIS
HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION.
HP_RENAL_TUBULAR_DYSFUNCTION, HP_RENAL_TUBULAR_DYSFUNCTION
HP ABNORMAL CIRCULATING FATTY ACID CONCENTRATION, HP ABNORMAL CIRCULATING FATTY ACID CONCENTRATION
HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER, HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER
HP_CENTROCECAL_SCOTOMA, HP_CENTROCECAL_SCOTOMA
HP_HYPERLORDOSIS, HP_HYPERLORDOSIS
HP_ABNORMAL_CIRCULATING_GLYCINE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_GLYCINE_CONCENTRATION
HP_INTESTINAL_PSEUDO_OBSTRUCTION, HP_INTESTINAL_PSEUDO_OBSTRUCTION
HP_FIBROUS_TISSUE_NEOPLASM, HP_FIBROUS_TISSUE_NEOPLASM
HP_ABNORMAL_RENAL_TUBULE_MORPHOLOGY, HP_ABNORMAL_RENAL_TUBULE_MORPHOLOGY
HP_AMINOACIDURIA, HP_AMINOACIDURIA
HP_MALIGNANT_NEOPLASM_OF_THE_CENTRAL_NERVOUS_SYSTEM, HP_MALIGNANT_NEOPLASM_OF_THE_CENTRAL_NERVOUS_SYSTEM
HP_INTESTINAL_POLYP, HP_INTESTINAL_POLYP
HP_STUTTERING, HP_STUTTERING
HP_ASTHMA, HP_ASTHMA
HP_GRAY_MATTER_HETEROTOPIA, HP_GRAY_MATTER_HETEROTOPIA
HP LEBER OPTIC ATROPHY, HP LEBER OPTIC ATROPHY
HP_GASTROINTESTINAL_DYSMOTILITY, HP_GASTROINTESTINAL_DYSMOTILITY
HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY, HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY
HP_PERIPHERAL_AXONAL_DEGENERATION, HP_PERIPHERAL_AXONAL_DEGENERATION
HP_BILATERAL_SINGLE_TRANSVERSE_PALMAR_CREASES, HP_BILATERAL_SINGLE_TRANSVERSE_PALMAR_CREASES
HP_CONGENITAL_DIAPHRAGMATIC_HERNIA, HP_CONGENITAL_DIAPHRAGMATIC_HERNIA
HP_ABNORMAL_NIPPLE_MORPHOLOGY, HP_ABNORMAL_NIPPLE_MORPHOLOGY
HP_TRIPHALANGEAL_THUMB, HP_TRIPHALANGEAL_THUMB
HP_EMOTIONAL_LABILITY, HP_EMOTIONAL_LABILITY
HP_NEOPLASM_OF_THE_SKELETAL_SYSTEM, HP_NEOPLASM_OF_THE_SKELETAL_SYSTEM
HP_APHASIA, HP_APHASIA
HP_WADDLING_GAIT, HP_WADDLING_GAIT
HP_ABNORMAL_PERICARDIUM_MORPHOLOGY, HP_ABNORMAL_PERICARDIUM_MORPHOLOGY
HP_ADRENAL_INSUFFICIENCY, HP_ADRENAL_INSUFFICIENCY
HP_VASCULAR_TORTUOSITY, HP_VASCULAR_TORTUOSITY
HP_OSTEOARTHRITIS, HP_OSTEOARTHRITIS
HP_LYMPHEDEMA, HP_LYMPHEDEMA
HP_PROMINENT_NOSE, HP_PROMINENT_NOSE
HP_ABNORMAL_CEREBROSPINAL_FLUID_METABOLITE_CONCENTRATION, HP_ABNORMAL_CEREBROSPINAL_FLUID_METABOLITE_CONCENTRATION
HP_NEONATAL_HYPOGLYCEMIA, HP_NEONATAL_HYPOGLYCEMIA
HP_NEURODEGENERATION, HP_NEURODEGENERATION
HP_RECURRENT_SKIN_INFECTIONS, HP_RECURRENT_SKIN_INFECTIONS
HP_ABNORMALITY_OF_RENAL_EXCRETION, HP_ABNORMALITY_OF_RENAL_EXCRETION
HP_BRAIN_IMAGING_ABNORMALITY, HP_BRAIN_IMAGING_ABNORMALITY
HP_ABNORMAL_METABOLIC_BRAIN_IMAGING_BY_MRS, HP_ABNORMAL_METABOLIC_BRAIN_IMAGING_BY_MRS
HP_MELANOCYTIC_NEVUS, HP_MELANOCYTIC_NEVUS
HP_BULBAR_SIGNS, HP_BULBAR_SIGNS
HP_PROGRESSIVE_EXTERNAL_OPHTHALMOPLEGIA, HP_PROGRESSIVE_EXTERNAL_OPHTHALMOPLEGIA
HP_CHOREOATHETOSIS, HP_CHOREOATHETOSIS
HP_PERSONALITY_CHANGES, HP_PERSONALITY_CHANGES
HP_BRISK_REFLEXES, HP_BRISK_REFLEXES
HP_LACTICACIDURIA, HP_LACTICACIDURIA
HP_ABNORMALITY_OF_THE_ULNA, HP_ABNORMALITY_OF_THE_ULNA
HP_MICROVESICULAR_HEPATIC_STEATOSIS, HP_MICROVESICULAR_HEPATIC_STEATOSIS
HP_3_METHYLGLUTACONIC_ACIDURIA, HP_3_METHYLGLUTACONIC_ACIDURIA
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D\_SERUM\_LACTATE, HP\_INCREASED\_SERUM\_LACTATE

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HP_ABNORMAL_CIRCULATING_NON_PROTEINOGENIC_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_NON_PROTEINOGENIC_AMINO_ACID_CONCEN
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HP\_IRREGULAR\_HYPERPIGMENTATION, HP\_IRREGULAR\_HYPERPIGMENTATION

HP\_PERIPHERAL\_AXONAL\_NEUROPATHY, HP\_PERIPHERAL\_AXONAL\_NEUROPATHY

HP\_ABNORMAL\_AORTIC\_VALVE\_MORPHOLOGY, HP\_ABNORMAL\_AORTIC\_VALVE\_MORPHOLOGY

HP\_ABNORMALITIES\_OF\_PLACENTA\_OR\_UMBILICAL\_CORD, HP\_ABNORMALITIES\_OF\_PLACENTA\_OR\_UMBILICAL\_CORD

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

HP\_FOCAL\_WHITE\_MATTER\_LESIONS, HP\_FOCAL\_WHITE\_MATTER\_LESIONS

HP\_MITOCHONDRIAL\_INHERITANCE, HP\_MITOCHONDRIAL\_INHERITANCE

HP\_UNUSUAL\_FUNGAL\_INFECTION, HP\_UNUSUAL\_FUNGAL\_INFECTION

HP\_ABNORMAL\_PLEURA\_MORPHOLOGY, HP\_ABNORMAL\_PLEURA\_MORPHOLOGY

HP\_ABNORMAL\_SIZE\_OF\_THE\_CLITORIS, HP\_ABNORMAL\_SIZE\_OF\_THE\_CLITORIS

HP\_SLOW\_DECREASE\_IN\_VISUAL\_ACUITY, HP\_SLOW\_DECREASE\_IN\_VISUAL\_ACUITY

HP\_GENERALIZED\_ONSET\_MOTOR\_SEIZURE, HP\_GENERALIZED\_ONSET\_MOTOR\_SEIZURE

HP\_NEOPLASM\_OF\_THE\_THYROID\_GLAND, HP\_NEOPLASM\_OF\_THE\_THYROID\_GLAND

HP\_ABNORMAL\_CIRCULATING\_IGG\_LEVEL, HP\_ABNORMAL\_CIRCULATING\_IGG\_LEVEL

HP\_FLUCTUATIONS\_IN\_CONSCIOUSNESS, HP\_FLUCTUATIONS\_IN\_CONSCIOUSNESS

HP\_ABNORMALITY\_OF\_ADRENAL\_PHYSIOLOGY, HP\_ABNORMALITY\_OF\_ADRENAL\_PHYSIOLOGY

HP\_ABNORMAL\_METATARSAL\_MORPHOLOGY, HP\_ABNORMAL\_METATARSAL\_MORPHOLOGY

HP\_SOMATIC\_MUTATION, HP\_SOMATIC\_MUTATION

HP\_STROKE\_LIKE\_EPISODE, HP\_STROKE\_LIKE\_EPISODE

HP\_AVASCULAR\_NECROSIS, HP\_AVASCULAR\_NECROSIS

HP\_NARROW\_NASAL\_BRIDGE, HP\_NARROW\_NASAL\_BRIDGE

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

HP\_NEOPLASM\_OF\_FATTY\_TISSUE, HP\_NEOPLASM\_OF\_FATTY\_TISSUE

HP\_RECURRENT\_PANCREATITIS, HP\_RECURRENT\_PANCREATITIS

HP\_DENTAL\_MALOCCLUSION, HP\_DENTAL\_MALOCCLUSION

HP\_EPISODIC\_VOMITING, HP\_EPISODIC\_VOMITING

HP\_SPINAL\_RIGIDITY, HP\_SPINAL\_RIGIDITY

HP\_LEUKEMIA, HP\_LEUKEMIA

HP\_MACULE, HP\_MACULE

HP\_MENINGITIS, HP\_MENINGITIS

HP\_BULBOUS\_NOSE, HP\_BULBOUS\_NOSE

HP\_AORTIC\_VALVE\_STENOSIS, HP\_AORTIC\_VALVE\_STENOSIS