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HP_ABNORMALITY_OF_THE_MITOCHONDRION, HP_ABNORMALITY_OF_THE_MITOCHONDRION
HP_LACTIC_ACIDOSIS, HP_LACTIC_ACIDOSIS
HP_SLOPING_FOREHEAD, HP_SLOPING_FOREHEAD
HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN, HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN
HP_CRANIOSYNOSTOSIS, HP_CRANIOSYNOSTOSIS
HP_INFERTILITY, HP_INFERTILITY
HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_I, HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_I
HP_DEMENTIA, HP_DEMENTIA
HP_SPASTIC_PARAPLEGIA, HP_SPASTIC_PARAPLEGIA
HP_LOWER_LIMB_SPASTICITY, HP_LOWER_LIMB_SPASTICITY
HP_VASCULAR_TORTUOSITY, HP_VASCULAR_TORTUOSITY
HP_GASTROINTESTINAL_DYSMOTILITY, HP_GASTROINTESTINAL_DYSMOTILITY
HP_INTESTINAL_PSEUDO_OBSTRUCTION, HP_INTESTINAL_PSEUDO_OBSTRUCTION
HP ABNORMAL SPEECH PROSODY, HP ABNORMAL SPEECH PROSODY
HP_SARCOMA, HP_SARCOMA
HP_MITOCHONDRIAL_INHERITANCE, HP_MITOCHONDRIAL_INHERITANCE
HP_PERIPHERAL_AXONAL_DEGENERATION, HP_PERIPHERAL_AXONAL_DEGENERATION
HP_CAFE_AU_LAIT_SPOT, HP_CAFE_AU_LAIT_SPOT
HP_SENSORIMOTOR_NEUROPATHY, HP_SENSORIMOTOR_NEUROPATHY
HP_DEMYELINATING_PERIPHERAL_NEUROPATHY, HP_DEMYELINATING_PERIPHERAL_NEUROPATHY
HP_ABNORMAL_PERIPHERAL_MYELINATION, HP_ABNORMAL_PERIPHERAL_MYELINATION
HP_PERIPHERAL_DEMYELINATION, HP_PERIPHERAL_DEMYELINATION
HP_GLOBAL_BRAIN_ATROPHY, HP_GLOBAL_BRAIN_ATROPHY
HP_LEUKODYSTROPHY, HP_LEUKODYSTROPHY
HP_BULBAR_SIGNS, HP_BULBAR_SIGNS
HP_LEBER_OPTIC_ATROPHY, HP_LEBER_OPTIC_ATROPHY
HP_ABNORMAL_LOCALIZATION_OF_KIDNEY, HP_ABNORMAL_LOCALIZATION_OF_KIDNEY
HP_TRIANGULAR_SHAPED_PHALANGES_OF_THE_HAND, HP_TRIANGULAR_SHAPED_PHALANGES_OF_THE_HAND
HP_INCREASED_CSF_LACTATE, HP_INCREASED_CSF_LACTATE
HP_RECURRENT_PAROXYSMAL_HEADACHE, HP_RECURRENT_PAROXYSMAL_HEADACHE
HP_SENSORY_NEUROPATHY, HP_SENSORY_NEUROPATHY
HP_MIXED_DEMYELINATING_AND_AXONAL_POLYNEUROPATHY, HP_MIXED_DEMYELINATING_AND_AXONAL_POLYNEUROPATHY
HP_ABNORMALITY_OF_HEPATOBILIARY_SYSTEM_PHYSIOLOGY, HP_ABNORMALITY_OF_HEPATOBILIARY_SYSTEM_PHYSIOLOGY
HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION, HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION
HP_ABNORMAL_RENAL_GLOMERULUS_MORPHOLOGY, HP_ABNORMAL_RENAL_GLOMERULUS_MORPHOLOGY
HP_CENTRAL_SCOTOMA, HP_CENTRAL_SCOTOMA
HP_PSYCHOMOTOR_RETARDATION, HP_PSYCHOMOTOR_RETARDATION
HP_ALOPECIA, HP_ALOPECIA
HP_PROGRESSIVE_EXTERNAL_OPHTHALMOPLEGIA, HP_PROGRESSIVE_EXTERNAL_OPHTHALMOPLEGIA
HP ABNORMAL BRAINSTEM MRI SIGNAL INTENSITY, HP ABNORMAL BRAINSTEM MRI SIGNAL INTENSITY
HP_SHORT_PHILTRUM, HP_SHORT_PHILTRUM
HP_MULTIPLE_GLOMERULAR_CYSTS, HP_MULTIPLE_GLOMERULAR_CYSTS
HP_PRENATAL_MATERNAL_ABNORMALITY, HP_PRENATAL_MATERNAL_ABNORMALITY
HP_LEUKEMIA, HP_LEUKEMIA
HP_DISTAL_PERIPHERAL_SENSORY_NEUROPATHY, HP_DISTAL_PERIPHERAL_SENSORY_NEUROPATHY
HP_INFANTILE_SPASMS, HP_INFANTILE_SPASMS
HP_VENTRICULAR_PREEXCITATION, HP_VENTRICULAR_PREEXCITATION
HP_GAIT_ATAXIA, HP_GAIT_ATAXIA
HP_RAGGED_RED_MUSCLE_FIBERS, HP_RAGGED_RED_MUSCLE_FIBERS
HP_MITOCHONDRIAL_MYOPATHY, HP_MITOCHONDRIAL_MYOPATHY
HP_NEPHROPATHY, HP_NEPHROPATHY
HP_CENTROCECAL_SCOTOMA, HP_CENTROCECAL_SCOTOMA
HP_STROKE_LIKE_EPISODE, HP_STROKE_LIKE_EPISODE
HP_PARAPLEGIA, HP_PARAPLEGIA
HP_MULTIFACTORIAL_INHERITANCE, HP_MULTIFACTORIAL_INHERITANCE
HP_EPISODIC_VOMITING, HP_EPISODIC_VOMITING
HP_EMOTIONAL_LABILITY, HP_EMOTIONAL_LABILITY
HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION, HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION
HP_OPTIC_DISC_PALLOR, HP_OPTIC_DISC_PALLOR
HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER, HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER
HP_SPINAL_DYSRAPHISM, HP_SPINAL_DYSRAPHISM
HP_MYELODYSPLASIA, HP_MYELODYSPLASIA
HP_PALMOPLANTAR_KERATODERMA, HP_PALMOPLANTAR_KERATODERMA
HP_SPASTIC_TETRAPARESIS, HP_SPASTIC_TETRAPARESIS
HP_PROXIMAL_TUBULOPATHY, HP_PROXIMAL_TUBULOPATHY
HP_ABNORMAL_SPERM_MOTILITY, HP_ABNORMAL_SPERM_MOTILITY
HP_ERYTHEMA, HP_ERYTHEMA
HP_STUTTERING, HP_STUTTERING
HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION, HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION
HP_ABNORMAL_CSF_METABOLITE_LEVEL, HP_ABNORMAL_CSF_METABOLITE_LEVEL
HP_ABNORMAL_URINE_PH, HP_ABNORMAL_URINE_PH
HP_ABNORMAL_CIRCULATING_NON_PROTEINOGENIC_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_NON_PROTEINOGENIC_AMINO_ACID_CONCEN
HP_GRAY_MATTER_HETEROTOPIA, HP_GRAY_MATTER_HETEROTOPIA
HP_ABNORMALITY_OF_THE_HALLUX, HP_ABNORMALITY_OF_THE_HALLUX
HP_ACUTE_LEUKEMIA, HP_ACUTE_LEUKEMIA
HP_LOW_PLASMA_CITRULLINE, HP_LOW_PLASMA_CITRULLINE
HP_FACIAL_HYPERTRICHOSIS, HP_FACIAL_HYPERTRICHOSIS
HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY, HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY
HP_HIGH_NARROW_PALATE, HP_HIGH_NARROW_PALATE
HP_POSTNATAL_MICROCEPHALY, HP_POSTNATAL_MICROCEPHALY
HP_BREAST_CARCINOMA, HP_BREAST_CARCINOMA
HP_INCREASED_SERUM_PYRUVATE, HP_INCREASED_SERUM_PYRUVATE
HP_PROMINENT_FOREHEAD, HP_PROMINENT_FOREHEAD
HP_SPARSE_BODY_HAIR, HP_SPARSE_BODY_HAIR
HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS, HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS
HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM, HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM
HP_OPTIC_NEUROPATHY, HP_OPTIC_NEUROPATHY
HP PIGMENTARY RETINOPATHY, HP PIGMENTARY RETINOPATHY
HP_BILATERAL_SINGLE_TRANSVERSE_PALMAR_CREASES, HP_BILATERAL_SINGLE_TRANSVERSE_PALMAR_CREASES
HP MUSCLE SPASM, HP MUSCLE SPASM
HP_POSTURAL_TREMOR, HP_POSTURAL_TREMOR
HP_ABNORMAL_METABOLIC_BRAIN_IMAGING_BY_MRS, HP_ABNORMAL_METABOLIC_BRAIN_IMAGING_BY_MRS
HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM, HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM
HP_RETINAL_ARTERIAL_TORTUOSITY, HP_RETINAL_ARTERIAL_TORTUOSITY
HP_ABNORMAL_MITOCHONDRIAL_MORPHOLOGY, HP_ABNORMAL_MITOCHONDRIAL_MORPHOLOGY
HP_FLUCTUATIONS_IN_CONSCIOUSNESS, HP_FLUCTUATIONS_IN_CONSCIOUSNESS
HP_SMOOTH_PHILTRUM, HP_SMOOTH_PHILTRUM
HP_FREQUENT_FALLS, HP_FREQUENT_FALLS
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D_SERUM_LACTATE, HP_INCREASED_SERUM_LACTATE

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HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION.
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HP_FETAL_DISTRESS, HP_FETAL_DISTRESS

HP_BROAD_TOE, HP_BROAD_TOE

HP_PREAXIAL_POLYDACTYLY, HP_PREAXIAL_POLYDACTYLY HP_PSYCHOSIS, HP_PSYCHOSIS

HP_MICROVESICULAR_HEPATIC_STEATOSIS, HP_MICROVESICULAR_HEPATIC_STEATOSIS

HP_APLASIA_HYPOPLASIA_OF_THE_SKIN, HP_APLASIA_HYPOPLASIA_OF_THE_SKIN

HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVATIVE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVA

HP_DILATED_CARDIOMYOPATHY, HP_DILATED_CARDIOMYOPATHY HP_PAPULE, HP_PAPULE

HP_EPILEPTIC_SPASM, HP_EPILEPTIC_SPASM

HP STROKE, HP STROKE

HP ACTION TREMOR, HP ACTION TREMOR HP_PRURITUS, HP_PRURITUS

HP_ABNORMAL_CORONARY_ARTERY_MORPHOLOGY, HP_ABNORMAL_CORONARY_ARTERY_MORPHOLOGY HP_ABSENT_SEPTUM_PELLUCIDUM, HP_ABSENT_SEPTUM_PELLUCIDUM

HP_NEOPLASM_OF_THE_BREAST, HP_NEOPLASM_OF_THE_BREAST

HP_CONVEX_NASAL_RIDGE, HP_CONVEX_NASAL_RIDGE

HP_WIDELY_SPACED_TEETH, HP_WIDELY_SPACED_TEETH HP_ABNORMAL_SPERMATOGENESIS, HP_ABNORMAL_SPERMATOGENESIS

HP_ABNORMAL_CSF_PROTEIN_LEVEL, HP_ABNORMAL_CSF_PROTEIN_LEVEL

HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE, HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE

HP_ABNORMALITY_OF_THE_PLANTAR_SKIN_OF_FOOT, HP_ABNORMALITY_OF_THE_PLANTAR_SKIN_OF_FOOT HP_DEATH_IN_CHILDHOOD, HP_DEATH_IN_CHILDHOOD

HP_WIDENED_CEREBRAL_SUBARACHNOID_SPACE, HP_WIDENED_CEREBRAL_SUBARACHNOID_SPACE

HP_ABNORMAL_CORTICAL_BONE_MORPHOLOGY, HP_ABNORMAL_CORTICAL_BONE_MORPHOLOGY

HP_ABNORMALITY_OF_THE_PHALANGES_OF_THE_TOES, HP_ABNORMALITY_OF_THE_PHALANGES_OF_THE_TOES HP_APLASIA_HYPOPLASIA_OF_THE_RIBS, HP_APLASIA_HYPOPLASIA_OF_THE_RIBS

HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION

HP_ABNORMAL_CAROTID_ARTERY_MORPHOLOGY, HP_ABNORMAL_CAROTID_ARTERY_MORPHOLOGY HP_JOINT_HYPERFLEXIBILITY, HP_JOINT_HYPERFLEXIBILITY

HP_WEBBED_NECK, HP_WEBBED_NECK HP_ABNORMALITY_OF_THE_CHOANAE, HP_ABNORMALITY_OF_THE_CHOANAE

HP_ABNORMAL_AORTIC_VALVE_PHYSIOLOGY, HP_ABNORMAL_AORTIC_VALVE_PHYSIOLOGY

HP_NEONATAL_HYPOGLYCEMIA, HP_NEONATAL_HYPOGLYCEMIA

HP_LYMPHEDEMA, HP_LYMPHEDEMA

HP_DOLICHOCEPHALY, HP_DOLICHOCEPHALY

HP_BASAL_GANGLIA_CALCIFICATION, HP_BASAL_GANGLIA_CALCIFICATION

HP_LACTICACIDURIA, HP_LACTICACIDURIA

HP_WIDE_NOSE, HP_WIDE_NOSE

HP_ABNORMAL_MIDDLE_PHALANX_MORPHOLOGY_OF_THE_HAND, HP_ABNORMAL_MIDDLE_PHALANX_MORPHOLOGY_OF_THE_HAND HP_TRIPHALANGEAL_THUMB, HP_TRIPHALANGEAL_THUMB

HP_RETINAL_TELANGIECTASIA, HP_RETINAL_TELANGIECTASIA

HP_OSTEOARTHRITIS, HP_OSTEOARTHRITIS