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| | HP_LACTIC_ACIDOSIS, HP_LACTIC_ACIDOSIS |
| | HP_ABNORMALITY_OF_THE_MITOCHONDRION, HP_ABNORMALITY_OF_THE_MITOCHONDRION |
| | HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN, HP_ABNORMAL_ACTIVITY_OF_MITOCHONDRIAL_RESPIRATORY_CHAIN |
| | HP_INCREASED_CSF_LACTATE, HP_INCREASED_CSF_LACTATE |
| | HP_ABNORMAL_CSF_METABOLITE_LEVEL, HP_ABNORMAL_CSF_METABOLITE_LEVEL |
| | HP_LEUKODYSTROPHY, HP_LEUKODYSTROPHY |
| | HP_SLOPING_FOREHEAD, HP_SLOPING_FOREHEAD |
| | HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION, HP_MUSCLE_ABNORMALITY_RELATED_TO_MITOCHONDRIAL_DYSFUNCTION |
| | 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_CAFE_AU_LAIT_SPOT, HP_CAFE_AU_LAIT_SPOT |
| | HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION, HP_FOCAL_T2_HYPERINTENSE_BASAL_GANGLIA_LESION |
| | HP_MITOCHONDRIAL_MYOPATHY, HP_MITOCHONDRIAL_MYOPATHY |
| | HP_PIGMENTARY_RETINOPATHY, HP_PIGMENTARY_RETINOPATHY |
| | HP_OPTIC_DISC_PALLOR, HP_OPTIC_DISC_PALLOR |
| | HP_INFERTILITY, HP_INFERTILITY |
| | HP_ABNORMAL_URINE_PH, HP_ABNORMAL_URINE_PH |
| | HP_DECREASED_ACTIVITY_OF_THE_PYRUVATE_DEHYDROGENASE_COMPLEX, HP_DECREASED_ACTIVITY_OF_THE_PYRUVATE_DEHYDROGENASE_COMPLEX |
| | HP_ABNORMALITY_OF_HEPATOBIILIARY_SYSTEM_PHYSIOLOGY, HP_ABNORMALITY_OF_HEPATOBIILIARY_SYSTEM_PHYSIOLOGY |
| | HP_LEUKEMIA, HP_LEUKEMIA |
| | HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE, HP_ABNORMAL_MITOCHONDRIA_IN_MUSCLE_TISSUE |
| | HP_LIPID_ACCUMULATION_IN_HEPATOCYTES, HP_LIPID_ACCUMULATION_IN_HEPATOCYTES |
| | HP_HYPOPIGMENTATION_OF_THE_SKIN, HP_HYPOPIGMENTATION_OF_THE_SKIN |
| | HP_INCREASED_SERUM_PYRUVATE, HP_INCREASED_SERUM_PYRUVATE |
| | HP_ACUTE_LEUKEMIA, HP_ACUTE_LEUKEMIA |
| | HP_HYPERAMMONEMIA, HP_HYPERAMMONEMIA |
| | HP_LOWER_LIMB_SPASTICITY, HP_LOWER_LIMB_SPASTICITY |
| | HP_RAGGED_RED_MUSCLE_FIBERS, HP_RAGGED_RED_MUSCLE_FIBERS |
| | HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY, HP_ABNORMAL_BRAINSTEM_MRI_SIGNAL_INTENSITY |
| | HP_ABSENT_SEPTUM_PELLUCIDUM, HP_ABSENT_SEPTUM_PELLUCIDUM |
| | HP_VASCULAR_TORTUOSITY, HP_VASCULAR_TORTUOSITY |
| | HP_METABOLIC_ACIDOSIS, HP_METABOLIC_ACIDOSIS |
| | HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_AMINO_ACID_CONCENTRATION |
| | HP_RETINAL_ARTERIAL_TORTUOSITY, HP_RETINAL_ARTERIAL_TORTUOSITY |
| | HP_OPTIC_NEUROPATHY, HP_OPTIC_NEUROPATHY |
| | HP_MITOCHONDRIAL_INHERITANCE, HP_MITOCHONDRIAL_INHERITANCE |
| | HP_LEBER_OPTIC_ATROPHY, HP_LEBER_OPTIC_ATROPHY |
| | HP_PROGRESSIVE_SPASTIC_PARAPLEGIA, HP_PROGRESSIVE_SPASTIC_PARAPLEGIA |
| | HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM, HP_ABNORMALITY_OF_KREBS_CYCLE_METABOLISM |
| | HP_HEPATIC_FAILURE, HP_HEPATIC_FAILURE |
| | HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS, HP_MITOCHONDRIAL_RESPIRATORY_CHAIN_DEFECTS |
| | HP_FETAL_ULTRASOUND_SOFT_MARKER, HP_FETAL_ULTRASOUND_SOFT_MARKER |
| | HP_SPASTIC_TETRAPARESIS, HP_SPASTIC_TETRAPARESIS |
| | HP_CENTROCECAL_SCOTOMA, HP_CENTROCECAL_SCOTOMA |
| | HP_SHORT_PALPEBRAL_FISSURE, HP_SHORT_PALPEBRAL_FISSURE |
| | HP_MULTIFACTORIAL_INHERITANCE, HP_MULTIFACTORIAL_INHERITANCE |
| | HP_PSYCHOMOTOR_RETARDATION, HP_PSYCHOMOTOR_RETARDATION |
| | HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL, HP_ABNORMAL_URINE_CARBOXYLIC_ACID_LEVEL |
| | HP_ABNORMAL_RENAL_GLOMERULUS_MORPHOLOGY, HP_ABNORMAL_RENAL_GLOMERULUS_MORPHOLOGY |
| | HP_EPISODIC_VOMITING, HP_EPISODIC_VOMITING |
| | HP_DECREASED_FERTILITY_IN_MALES, HP_DECREASED_FERTILITY_IN_MALES |
| | HP_FETAL_DISTRESS, HP_FETAL_DISTRESS |
| | HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY, HP_ABNORMAL_CORPUS_STRIATUM_MORPHOLOGY |
| | HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_IV, HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_IV |
| | HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS, HP_PAROXYSMAL_INVOLUNTARY_EYE_MOVEMENTS |
| | HP_AMINOACIDURIA, HP_AMINOACIDURIA |
| | HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM, HP_ABNORMALITY_OF_THE_SEPTUM_PELLUCIDUM |
| | HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_CARBOHYDRATE_CONCENTRATION |
| | HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION, HP_FOCAL_T2_HYPERINTENSE_BRAINSTEM_LESION |
| | HP_MALE_INFERTILITY, HP_MALE_INFERTILITY |
| | HP_POOR_HEAD_CONTROL, HP_POOR_HEAD_CONTROL |
| | HP_ABNORMAL_CIRCULATING_SERINE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_SERINE_FAMILY_AMINO_ACID_CONCENTRATION |
| | HP_ABNORMALITY_OF_CORNEAL_STROMA, HP_ABNORMALITY_OF_CORNEAL_STROMA |
| | HP_ABNORMAL_CIRCULATING_GLUTAMINE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_GLUTAMINE_FAMILY_AMINO_ACID_CONCENTRATION |
| | HP_PROGRESSIVE_EXTERNAL_OPTHALMOPLÉGIA, HP_PROGRESSIVE_EXTERNAL_OPTHALMOPLÉGIA |
| | HP_3_METHYLGLUTAConIC_ACIDURIA, HP_3_METHYLGLUTAConIC_ACIDURIA |
| | HP_MULTIPLE_GLOMERULAR_CYSTS, HP_MULTIPLE_GLOMERULAR_CYSTS |
| | HP_PROGRESSIVE_CEREBELLAR_ATAxia, HP_PROGRESSIVE_CEREBELLAR_ATAxia |
| | HP_RECURRENT_URINARY_TRACT_INFECTIOnS, HP_RECURRENT_URINARY_TRACT_INFECTIOnS |
| | HP_INTESTINAL_POLYP, HP_INTESTINAL_POLYP |
| | HP_INCREASED_MUSCLE_LIPID_CONTENT, HP_INCREASED_MUSCLE_LIPID_CONTENT |
| | HP_ABNORMAL_CIRCULATING_PROLINE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PROLINE_CONCENTRATION |
| HP_O_SERUM_LACTATE, HP_INCREASED_SERUM_LACTATE | HP_DECREASED_TESTICULAR_SIZE, HP_DECREASED_TESTICULAR_SIZE |
| | HP_INTESTINAL_PSEUDO_OBSTRUCTION, HP_INTESTINAL_PSEUDO_OBSTRUCTION |
| | HP_MyALGIA, HP_MyALGIA |
| | HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVATIVE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PROTEINOGENIC_AMINO_ACID_DERIVATIVE_CONCENTRATION |
| | HP_ABNORMAL_RENAL_CORTEX_MORPHOLOGY, HP_ABNORMAL_RENAL_CORTEX_MORPHOLOGY |
| | HP_DEMENTIA, HP_DEMENTIA |
| | HP_SKIN_PIT, HP_SKIN_PIT |
| | HP_ACNE, HP_ACNE |
| | HP_NEOPLASM_OF_THE_BREAST, HP_NEOPLASM_OF_THE_BREAST |
| | HP_PROMINENT_NOSE, HP_PROMINENT_NOSE |
| | HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_PYRUVATE_FAMILY_AMINO_ACID_CONCENTRATION |
| | HP_POLYNEUROPATHY, HP_POLYNEUROPATHY |
| | HP_FLUCTUATIOnS_IN_CONSCIOUSNESS, HP_FLUCTUATIOnS_IN_CONSCIOUSNESS |
| | HP_ABNORMALITY_OF_THE_PERIVENTRICULAR_WHITE_MATTER, HP_ABNORMALITY_OF_THE_PERIVENTRICULAR_WHITE_MATTER |
| | HP_ECTOPIC_ANUS, HP_ECTOPIC_ANUS |
| | HP_SPASTIC_PARAPLEGIA, HP_SPASTIC_PARAPLEGIA |
| | HP_ABNORMALITY_OF_THE_TESTIS_SIZE, HP_ABNORMALITY_OF_THE_TESTIS_SIZE |
| | HP_CLEFT_LIP, HP_CLEFT_LIP |
| | HP_MELANOMA, HP_MELANOMA |
| | HP_INSULIN_RESISTANCE, HP_INSULIN_RESISTANCE |
| | HP_RETINAL_TELANGIECTASIA, HP_RETINAL_TELANGIECTASIA |
| | HP_POOR_EYE_CONTACT, HP_POOR_EYE_CONTACT |
| | HP_ABNORMAL_SACRUM_MORPHOLOGY, HP_ABNORMAL_SACRUM_MORPHOLOGY |
| | HP_DELAYED_CRANIAL_SUTURE_CLOSURE, HP_DELAYED_CRANIAL_SUTURE_CLOSURE |
| | HP_PLEURAL_EFFUSION, HP_PLEURAL_EFFUSION |
| | HP_BREAST_CARCINOMA, HP_BREAST_CARCINOMA |
| | HP_BULBAR_SIGNS, HP_BULBAR_SIGNS |
| | HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION, HP_SEGMENTAL_PERIPHERAL_DEMYELINATION_REMYELINATION |
| | HP_OVARIAN_NEOPLASM, HP_OVARIAN_NEOPLASM |
| | HP_HIGHLY_ARCHED_EYEBROW, HP_HIGHLY_ARCHED_EYEBROW |
| | HP_GENERALIZED_LIMB_MUSCLE_ATROPHY, HP_GENERALIZED_LIMB_MUSCLE_ATROPHY |
| | HP_FREQUENT_FALLS, HP_FREQUENT_FALLS |
| | HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER, HP_APLASIA_HYPOPLASIA_OF_THE_CEREBRAL_WHITE_MATTER |
| | HP_JOINT_HYPERFLEXIBILITY, HP_JOINT_HYPERFLEXIBILITY |
| | HP_DEATH_IN_CHILDHOOD, HP_DEATH_IN_CHILDHOOD |
| | HP_SMOOTH_PHILTRUM, HP_SMOOTH_PHILTRUM |
| | HP_MICROVESICULAR_HEPATIC_STEATOSIS, HP_MICROVESICULAR_HEPATIC_STEATOSIS |
| | HP_DUPLICATION_OF_THUMB_PHALANX, HP_DUPLICATION_OF_THUMB_PHALANX |
| | HP_PROGRESSIVE_MUSCLE_WEAKNESS, HP_PROGRESSIVE_MUSCLE_WEAKNESS |
| | HP_ABNORMAL_CIRCULATING_GLYCINE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_GLYCINE_CONCENTRATION |
| | HP_ABNORMAL_LABIA_MORPHOLOGY, HP_ABNORMAL_LABIA_MORPHOLOGY |
| | HP_RECURRENT_MyCOBACTERIAL_INFECTIOnS, HP_RECURRENT_MyCOBACTERIAL_INFECTIOnS |
| | HP_DEATH_IN_INFANCY, HP_DEATH_IN_INFANCY |
| | HP_ABNORMAL_CAUDATE_NUCLEUS_MORPHOLOGY, HP_ABNORMAL_CAUDATE_NUCLEUS_MORPHOLOGY |
| | HP_ABNORMAL_GLIAL_CELL_MORPHOLOGY, HP_ABNORMAL_GLIAL_CELL_MORPHOLOGY |
| | HP_HYPOPIGMENTED_SKIN_PATCHES, HP_HYPOPIGMENTED_SKIN_PATCHES |
| | HP_ABNORMAL_GLOBUS_PALLIDUS_MORPHOLOGY, HP_ABNORMAL_GLOBUS_PALLIDUS_MORPHOLOGY |
| | HP_RENAL_FANCONI_SYNDROME, HP_RENAL_FANCONI_SYNDROME |
| | HP_BLURRED_VISION, HP_BLURRED_VISION |
| | HP_ABSENCE_OF_SECONDARY_SEX_CHARACTERISTICS, HP_ABSENCE_OF_SECONDARY_SEX_CHARACTERISTICS |
| | HP_HYPOMIMIC_FACE, HP_HYPOMIMIC_FACE |
| | HP_ABNORMAL_TRACHEA_MORPHOLOGY, HP_ABNORMAL_TRACHEA_MORPHOLOGY |
| | HP_ABNORMAL_URETER_PHYSIOLOGY, HP_ABNORMAL_URETER_PHYSIOLOGY |
| | HP_THICK_LOWER_LIP_VERMILION, HP_THICK_LOWER_LIP_VERMILION |
| | HP_UNDERDEVELOPED_SUPRAORBITAL_RIDGES, HP_UNDERDEVELOPED_SUPRAORBITAL_RIDGES |
| | HP_ANAL_ATRESIA, HP_ANAL_ATRESIA |
| | HP_ABNORMAL_SPEECH_PROSODY, HP_ABNORMAL_SPEECH_PROSODY |
| | HP_HYPERVENTILATION, HP_HYPERVENTILATION |
| | HP_INCREASED_INTRAMYOCeLLULAR_LIPID_DROPLETS, HP_INCREASED_INTRAMYOCeLLULAR_LIPID_DROPLETS |
| | HP_ARTERIAL_STENOSIS, HP_ARTERIAL_STENOSIS |
| | HP_CHOREOATHETOSIS, HP_CHOREOATHETOSIS |
| | HP_PSYCHOTIC_MENTATION, HP_PSYCHOTIC_MENTATION |
| | HP_ABNORMALITY_OF_THE_MAXILLA, HP_ABNORMALITY_OF_THE_MAXILLA |
| | HP_ABNORMAL_CIRCULATING_CITRULLINE_CONCENTRATION, HP_ABNORMAL_CIRCULATING_CITRULLINE_CONCENTRATION |
| | HP_STUTTERING, HP_STUTTERING |
| | HP_ABNORMAL_CIRCULATING_FATTY_ACID_CONCENTRATION, HP_ABNORMAL_CIRCULATING_FATTY_ACID_CONCENTRATION |
| | HP_TRACHEOESOPHAGEAL_FISTULA, HP_TRACHEOESOPHAGEAL_FISTULA |
| | HP_ABNORMAL_CAROTID_ARTERY_MORPHOLOGY, HP_ABNORMAL_CAROTID_ARTERY_MORPHOLOGY |
| | HP_CHOLELITHIASIS, HP_CHOLELITHIASIS |
| | HP_ABNORMAL_LOCALIZATION_OF_KIDNEY, HP_ABNORMAL_LOCALIZATION_OF_KIDNEY |
| | HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY, HP_SEVERE_GLOBAL_DEVELOPMENTAL_DELAY |
| | HP_ABNORMAL_LARGE_INTESTINE_PHYSIOLOGY, HP_ABNORMAL_LARGE_INTESTINE_PHYSIOLOGY |
| | HP_CONGENITAL_SENSORINEURAL_HEARING_IMPAIRMENT, HP_CONGENITAL_SENSORINEURAL_HEARING_IMPAIRMENT |
| | HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_III, HP_DECREASED_ACTIVITY_OF_MITOCHONDRIAL_COMPLEX_III |
| | HP_PROXIMAL_TUBULOPathY, HP_PROXIMAL_TUBULOPathY |