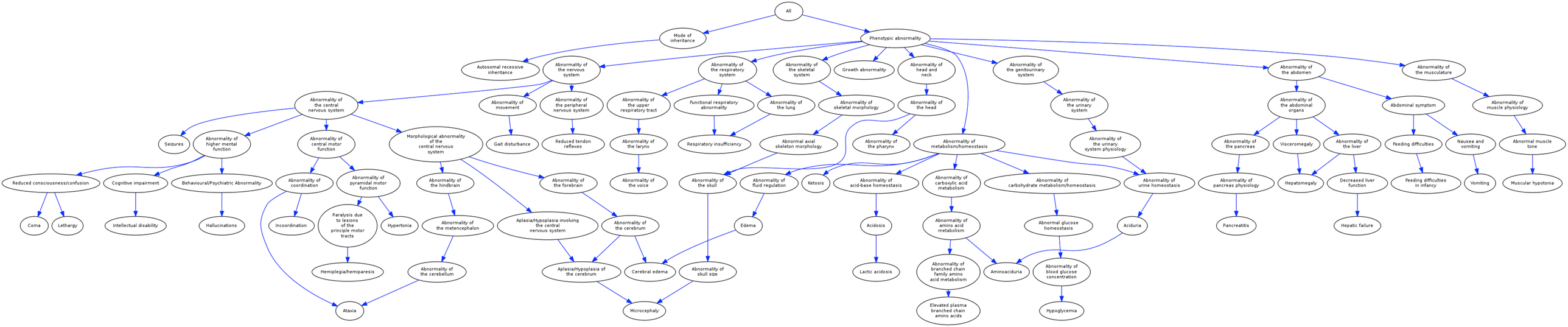
All
Mode of inheritance
Autosomal recessive inheritance
Phenotypic abnormality
Abnormality of the nervous system
Abnormality of the respiratory system
Abnormality of the skeletal system
Growth abnormality
Abnormality of head and neck
Abnormality of the genitourinary system
Abnormality of the abdomen
Abnormality of the musculature
Abnormality of the central nervous system
Abnormality of movement
Abnormality of the peripheral nervous system
Abnormality of the upper respiratory tract
Functional respiratory abnormality
Abnormality of the lung
Abnormality of skeletal morphology
Abnormality of the head
Abnormality of the urinary system
Abnormality of the abdominal organs
Abdominal symptom
Abnormality of muscle physiology
Seizures
Abnormality of higher mental function
Abnormality of central motor function
Morphological abnormality of the central nervous system
Gait disturbance
Reduced tendon reflexes
Abnormality of the larynx
Respiratory insufficiency
Abnormal axial skeleton morphology
Abnormality of the pharynx
Abnormality of metabolism/homeostasis
Abnormality of the urinary system physiology
Abnormality of the pancreas
Visceromegaly
Abnormality of the liver
Feeding difficulties
Nausea and vomiting
Abnormal muscle tone
Reduced consciousness/confusion
Cognitive impairment
Behavioural/Psychiatric Abnormality
Abnormality of coordination
Abnormality of pyramidal motor function
Abnormality of the hindbrain
Abnormality of the forebrain
Abnormality of the voice
Abnormality of the skull
Abnormality of fluid regulation
Ketosis
Abnormality of acid-base homeostasis
Abnormality of carboxylic acid metabolism
Abnormality of carbohydrate metabolism/homeostasis
Abnormality of urine homeostasis
Abnormality of pancreas physiology
Hepatomegaly
Decreased liver function
Feeding difficulties in infancy
Vomiting
Muscular hypotonia
Coma
Lethargy
Intellectual disability
Hallucinations
Incoordination
Paralysis due to lesions of the principle motor tracts
Hypertonia
Abnormality of the metencephalon
Aplasia/Hypoplasia involving the central nervous system
Abnormality of the cerebrum
Edema
Acidosis
Abnormality of amino acid metabolism
Abnormal glucose homeostasis
Aciduria
Pancreatitis
Hepatic failure
Hemiplegia/hemiparesis
Abnormality of the cerebellum
Aplasia/Hypoplasia of the cerebrum
Cerebral edema
Abnormality of skull size
Lactic acidosis
Abnormality of branched chain family amino acid metabolism
Aminoaciduria
Abnormality of blood glucose concentration
Ataxia
Microcephaly
Elevated plasma branched chain amino acids
Hypoglycemia