Neurometabolic conditions refer to metabolic disorders that impact brain development or function. Individuals with these disorders are either not able to produce certain enzymes, proteins, and/or other substances needed by the brain, or they are unable to break down these substances, which can cause them to build up and can be toxic to the brain.

As a whole, neurometabolic disorders range both in age of onset and severity, even within the same specific disorder. People with neurometabolic disorders can show symptoms anywhere from birth all the way through adulthood. Individuals who have symptoms at birth or in infancy tend to be more severely affected, but there is wide variability in the number and severity of symptoms. Some symptoms that can be seen with neurometabolic disorders include:

- abnormal brain structures
- seizures
- poor development (also called neurodevelopmental delay)
- neurological regression (e.g. loss of skills)
- movement disorders
- abnormal behaviors
- sleep disorders
- psychiatric disorders
- other body system involvement (also called multi-systemic involvement).

Diagnosis of a neurometabolic disorder is similar to how metabolic disorders in general are diagnosed, which is:

- through careful evaluation of an individual's medical and family history
- physical examination
- measurement of specific levels of certain substances in the blood, urine, cerebrospinal fluid (CSF), or other body tissues (e.g. skin or muscle)
- neuroimaging (MRI, functional MRI, MRS)
- other studies, and/or genetic testing.

While many neurometabolic disorders do not have known treatments, early diagnosis is crucial so those with an available treatment can be started early. If these conditions lead to any sort of brain damage, the brain is often not able to regain full function. Thus, treatment is focused on reducing and eliminating the buildup of the toxic substances in the brain to prevent the damage from occurring in the first place. This may be done through a specialized diet, taking supplements, or through replacing the missing enzymes. Gene therapy is also being assessed for a handful of these disorders through clinical trials.

Starting disease-specific treatments during this crucial early period is dependent on having an accurate diagnosis.

Examples of neurometabolic disorders include:

- Amino acid disorders (e.g. phenylketonuria, PKU)
- Peroxisomal disorders (e.g., adrenoleukodystrophy, ALD or Zellweger or Neuronal Ceroid-lipofuscinosis, NCL)
- Lysosomal storage disorders (e.g. Tay-Sachs disease or mucopolysaccharidoses, MPS)
- Mitochondrial disorders (e.g. Leigh syndrome)
- Neurotransmitter disorders (e.g. PTPS deficiency)
- Other (e.g. GLUT1 deficiency)