

## C 86 Inborn Errors of Metabolism

Inborn errors of metabolism generally refer to gene mutations or gene deletions that alter metabolism in the body, including but not limited to:

### **Amino Acid Disorders**

- Phenylketonuria
- Maple Syrup Urine Disease
- Homocystinuria
- Tyrosinemia

### **Carbohydrate Disorders**

- Galactosemia
- Glycogen Storage Disease I
- Glycogen Storage Disease II
- Glycogen Storage Disease III
- Glycogen Storage Disease IV
- Glycogen Storage Disease V
- Glycogen Storage Disease VI
- Hereditary Fructose Intolerance (Fructose 1-phosphate aldolase deficiency, Fructose 1,6,biphosphate deficiency, fructose kinase deficiency)

### **Fatty Acid Oxidation Defects**

- Medium chain acyl-CoA dehydrogenase deficiency
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency type 1
- Trifunctional protein deficiency type 2
- Carnitine uptake defect
- Very long chain acyl-CoA dehydrogenase deficiency

### **Organic Acid disorders (AKA organic aciduria or organic acidemia)**

- Isovaleric acidemia
- 3-Methylcrotonyl-CoA-carboxylase deficiency
- Glutaric acidemia type I
- Glutaric acidemia type II
- 3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency
- Multiple carboxylase deficiency (Biotinidase deficiency, Holocarboxylase synthetase deficiency)
- Methylmalonic acidemia
- Propionic acidemia
- Beta-ketothiolase deficiency

## **Lysosomal Storage Disease**

- Fabry disease ( $\alpha$ -galactosidase A deficiency)
- Gauchers disease (glucocerebrosidase deficiency)
- Pompe disease (glycogen storage disease Type II, or acid  $\alpha$ -glucosidase deficiency)

## **Mitochondrial Disorders**

- Leber hereditary optic neuropathy
- Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)
- Mitochondrial neurogastrointestinal encephalopathy disease (MNGIE)
- Myoclonic epilepsy with ragged-red fibers (MERRF)
- Neuropathy, ataxia, and retinitis pigmentosa (NARP)
- Pyruvate carboxylase deficiency

## **Peroxisomal Disorders**

- Zellweger Syndrome Spectrum
- Adrenoleukodystrophy (x-ALD)

## **Urea Cycle Disorders**

- Citrullinemia
- Argininosuccinic aciduria
- Carbamoyl phosphate synthetase I deficiency