

# Disorders Screened and Test Accuracy

## 1. What disorders does the newborn screen detect?

- BC's Newborn Screening Program screens for 22 disorders.

**Table 1: Disorders Screened**

Grouping	Metabolites Measured	Disorder	Abbrev.
Metabolic Disorders			
Amino Acid Disorders	Amino Acids	Phenylketonuria	PKU
		Maple Syrup Urine Disease	MSUD
		Citrullinemia	CIT
		Argininosuccinic Acidemia	ASA
		Homocystinuria	Hcy
		Tyrosinemia, Type I	Tyr I
Fatty Acid Oxidation Disorders	Acylcarnitines	Medium-chain Acyl-CoA Dehydrogenase Deficiency	MCAD
		Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
		Trifunctional Protein Deficiency	TFP
		Very-long Chain AcylCoA Dehydrogenase Deficiency	VLCAD
Organic Acid Disorders	Acylcarnitines	Propionic Acidemia	PROP
		Methylmalonic Acidemia	MUT
		Cobalamin Disorders	Cbl A, B
		Glutaric Aciduria, Type I	GA I
		Isovaleric Acidemia	IVA
Galactosemia	GALT enzyme activity	Galactosemia	GALT
Endocrine Disorders	Thyroid stimulating hormone (TSH)	Congenital Hypothyroidism	CH
	17OH-progesterone (1 <sup>st</sup> tier) Steroid panel (2 <sup>nd</sup> tier)	Congenital Adrenal Hyperplasia	CAH
Hemoglobinopathies	Hemoglobin HPLC	Sickle Cell Disease	HbSS
		Sickle Cell/Hemoglobin C	HSC
		Sickle Cell/ $\beta$ -thalassemia	HbS/ $\beta$ -thal
Cystic Fibrosis	Immunoreactive trypsinogen (IRT) (1 <sup>st</sup> tier) CFTR mutation panel (2 <sup>nd</sup> tier)	Cystic Fibrosis	CF