별 첨 1

GC Labs

신생아 선천성 대사이상 선별검사 세부항목 안내

분류		질환명
Core panel	내분비 대사이상	선천성 갑상선기능저하증(Congenital Hypothyroidism)
		선천성 부신과형성증(Congenital Adrenal Hyperplasia)
	기타 대사이상	갈락토스혈증(Galactosemia)
	아미노산 대사이상	Classic Phenylketonuria (PKU)
		Maple Syrup Urine Disease (MSUD)
		Classic Homocystinuria with Hypermethioninemia
		Citrullinemia, Type Ⅰ
		Argininosuccinic Aciduria
		Tyrosinemia, Type Ⅰ
	유기산 대사이상	Isovaleric Acidemia
		Propionic Acidemia
		Glutaric Acidemia, Type Ⅰ(Glutaryl-CoA Dehydrogenase Deficiency)
		Holocarboxylase Synthetase Deficiency
		Biotinidase Deficiency
		Methylmalonic Acidemia without Homocystinuria, Vitamin B12-Unresponsive (due to Methylmalonyl-CoA Mutase Deficiency)
		Methylmalonic Acidemia without Homocystinuria, Vitamin B12-Responsive (Cbl A, B, & Dv2)
		3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency (3-Methylcrotonylglycinuria)
		3-Hydroxy-3-Methylglutaric Aciduria (3-Hydroxy-3-Methylglutaryl-CoA-Lyase Deficiency = HMG-CoA Lyase Deficiency)
		β-Ketothiolase Deficiency
	지질 대사이상	Systemic Primary Carnitine Deficiency (Carnitine Uptake Deficiency = Carnitine Transporter Defect)
		Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
		Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
		Long-Chain Hydroxy Acyl-CoA Dehydrogenase (LCHAD) Deficiency
		Trifunctional Protein Deficiency
Secondary panel	아미노산 대사이상	Argininemia
		Citrullinemia, Type Ⅱ (Citrin Deficiency)
		Benign Hyperphenylalaninemia
		Hyperphenylalaninemia due to Impaired Regeneration of BH4 (DHPR, PCD Deficiency)
		Hyperphenylalaninemia due to Impaired Biosynthesis of BH4 (GTPCH, PTPS Deficiency)

분류		질환명
Secondary panel	아미노산 대사이상	Tyrosinemia, Type Ⅱ
		Tyrosinemia, Type Ⅲ
		Transient Neonatal Tyrosinemia
		Hypermethioninemia
		Homocystinuria without Hypermethioninemia & Methylmalonic Aciduria (Cbl E, G, & Dv1 / MTHFR Deficiency)
		Carbamoyl Phosphate Synthetase Ⅰ (CPS Ⅰ) Deficiency
		Ornithine Transcarbamylase (OTC) Deficiency
		Ornithine Aminotransferase (OAT) Deficiency (Gyrate Atrophy of Choroid and Retina)
		Glycine Encephalopathy (Non-Ketotic Hyperglycinemia)
		Hyperleucine-Isoleucinemia
		Hypervalinemia
		Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome (Ornithine Translocase Deficiency)
		Histidinemia
		Hyperprolinemia, Type Ⅰ
		Hyperprolinemia, Type Ⅱ
	유기산 대사이상	Methylmalonic Acidemia with Homocystinuria (Cbl C, D, F, & J)
		Succinyl-CoA Ligase Deficiency
		Ethylmalonic Encephalopathy
		Malonic Aciduria (Malonyl-CoA Decarboxylase Deficiency)
		Isobutyryl-CoA Dehydrogenase (IBD) Deficiency (Isobutyrylglycinuria)
		2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase (MHBD) Deficiency (HSD10 Disease = 2-Methyl-3-Hydroxybutyric Aciduria)
		2-Methylbutyryl-CoA Dehydrogenase (2-MBCD) Deficiency (2-Methylbutyryl Aciduria)
		3-Methylglutaconic Aciduria (3-Methylglutaconyl-CoA Hydratase Deficiency)
	지질 대사이상	Carnitine Palmitoyltransferase Ⅰ (CPT Ⅰ) Deficiency
		Carnitine Palmitoyltransferase Ⅱ (CPT Ⅱ) Deficiency
		Carnitine-Acylcarnitine Translocase (CACT) Deficiency
		Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
		Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) (Glutaric Acidemia Type Ⅱ)
		Short-Chain Hydroxy Acyl-CoA Dehydrogenase (SCHAD) Deficiency
		2,4 Dienoyl-CoA Reductase Deficiency (Progressive Encephalopathy with Leukodystrophy due to DECR Deficiency)
		Medium-Chain Ketoacyl-CoA Thiolase (MCAT) Deficiency