[첨부1] 선천성 대사이상 선별검사(검사코드:13071) 보고항목

항 목			
갈락토즈혈증	Galactose	HHH syndrome	HHH 신드롬
갑상선기능저하증	Neonatal-TSH	Argininemia	고알리진혈증
선천성부신과형성증	17a-OH Progesterone	Tyrosinemia Type Ⅰ	고타이로신혈증 I형
유기산대사이상 15종		Tyrosinemia Type Ⅱ	고타이로신혈증 II형
2-Methyl-3-Hydroxybutyric Aciduria	2-메틸-3-하이드록시뷰티릭산뇨증	Tyrosinemia Type Ⅲ	고타이로신혈증 III형
2-Methylbutyrylglycinuria	2-메틸뷰티릴글라이신뇨증	Neonatal Tyrosinemia	신생아기 고티로신혈증
3-Methylglutaconic Aciduria	3-메틸글루타코닉산뇨증	Carbamylphosphate I Synthetase deficiency	카바밀합성효소결핍증
3-Methylcrotonylglycinuria	3-메틸크로토닐글라이신뇨증	Hyperornithinemia	고오르니틴혈증
Glutaric Acidemia Type I	글루타릭산혈증 I형	Glycine encephalopathy (nonketotic hyperglycinemia)	글리신뇌증
Methylmalonic Acidemia, Vit. B12 responsive	비타민 B12 반응성 메틸말론산혈증	N-Acetylglutamate synthase deficiency	N-아세틸글루타메이트 합성결핍증
Multiple Carboxylase Deficiency	복합탈탄산효소결핍증	S-Adenosylhomocysteine hydrolase deficiency	S-아데노실호모시스틴 분해효소결핍증
Beta Ketothiolase Deficiency	베타케토티올분해효소결핍증	Histidinemia	히스티딘혈증
Methylmalonic Acidemia	메틸말로닉산혈증	Hyperprolinemia Type Ⅰ	고프롤린혈증 I형
Isovaleric Acidemia	이소발레릭산혈증	Hyperprolinemia Type Ⅱ	고프롤린혈증 II형
Propionic Acidemia	프로피오닉산혈증	E3 Deficiency	E3 결핍증
Malonic Acidemia	말로닉산혈증	**지방산대사이상 13종**	
Isobutyryl-CoA Dehydrogenase Deficier	이소부티릴코에이탈수소효소결핍증	Glutaric Acidemia Type Ⅱ	글루타릭산혈증 II형
Methylmalonic Aciduria with Homocystinuria	메틸말론산 및 호모시스틴뇨증	Short-Chain Acyl CoA Dehydrogenase Deficiency	단쇄아실코에이탈수소효소 결핍증
Ethylmalonic Encephalopathy	에틸말로닉 뇌증	Multiple Acyl-CoA Dehydrogenase Deficiency	복합아실코에이탈수소효소 결핍증
아미노산대사이상 26종		Long-Chain 3-HydroxyAcyl-CoA Dehydrogenase Deficiency	장쇄하이드록시아실코에이탈수소효소 결핍증
Phenylketonuria	페닐케톤뇨증	Medium Chain Acyl-CoA Dehydrogenase Deficiency	중쇄아실코에이탈수소효소 결핍증
Defect of Biopterin Cofactor Biosynthes	비옵테린 조효소 생합성 결핍증	Very long Chain Acyl-CoA Dehydro genase Deficiency	초장쇄아실코에이탈수소효소 결핍증
Homocystinuria	호모시스틴뇨증	Carnitine-Acylcarnitine Translocase Deficiency	카르니틴-아실카르니틴전이효소 결핍증
Hypermethioninemia	고메티오닌혈증	Carnitine uptake defect	카르니틴 업테이크 결핍증
Maple Syrup Urine Disease	단풍당뇨증	Carnitine Palmitoyl Transferase Ⅰ Deficiency	카르니틴팔미토일전이효소 I 결핍증
Hypervalinemia	고발린혈증	Carnitine Palmitoyl Transferase Ⅱ Deficiency	카르니틴팔미토일전이효소 II 결핍증
Ornithine Transcarbamylase Deficiency	오르니틴트랜스카바밀효소결핍증	Medium/Short-Chain 3-Hydroxy Acyl CoA Dehydrogenase Deficiency	중단쇄하이드록시아실코에이탈수소효소 결핍증
Defect of Biopterin Cofactor Regenerat	비옵테린 조효소 재합성 결핍증	2,4-Dienoyl CoA Reductase Deficiency	2,4-디에노일코에이환원효소 결핍증
Citrullinemia	시트룰린혈증	Mitochondrial Trifunctional Protein Deficiency	미토콘드리아삼중기능성단백질 결핍증
Citrullinemia Type Ⅱ	시트룰린혈증 II형	**케톤대사이상 1종**	
Argininosuccinic Acidemia	알지노석시닉산혈증	3-Hydroxy-3-Methylglutaric Aciduria	3-하이드록시-3-메틸클루타릭산뇨증

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