첨부파일

Tandem mass 검사법을 이용한 선천성 대사이상 검사 분석항목 55종

삼광의료재단

변경 전

질환분류	No.	내용		
유기산 대사이상질환	1	2-Methyl-3-Hydroxybutyric Aciduria	2-메틸-3-하이드록시뷰티릭산뇨증	
	2	2-Methylbutyrylglycinuria	2-메틸뷰티릴글라이신뇨증	
	3	3-Methylglutaconic Aciduria	3-메틸글루타코닉산뇨증	
	4	3-Methylcrotonylglycinuria	3-메틸크로토닐글라이신뇨증	
	5	Glutaric Acidemia Type I	글루타릭산혈증 Type I	
	6	Methylmalonic Acidemia, Vit. B12 responsive	비타민 B12 반응성 메틸말론산혈증	
	7	Multiple Carboxylase Deficiency	복합탈탄산효소결핍증	
	8	Beta Ketothiolase Deficiency	베타케토티올분해효소결핍증	
	9	Methylmalonic Acidemia	메틸말로닉산혈증	
	10	Isovaleric Acidemia	이소발레릭산혈증	
	11	Propionic Acidemia	프로피오닉산혈증	
	12	Malonic Acidemia	말로닉산혈증	
	13	Isobutyryl-CoA Dehydrogenase Deficiency	이소부티릴코에이탈수소효소결핍증	
	14	Methylmalonic Aciduria with Homocystinuria	메틸말론산 및 호모시스틴뇨증	
	15	Ethylmalonic Encephalopathy	에틸말로닉 뇌증	
아미노산 대사이상질환	16	Phenylketonuria	페닐케톤뇨증	
	17	Defect of Biopterin Cofactor Biosynthesis	비옵테린 조효소 생합성 결핍증	
	18	Homocystinuria	호모시스틴뇨증	
	19	Hypermethioninemia	고메티오닌혈증	
	20	Maple Syrup Urine Disease	단풍당뇨증	
	21	Hypervalinemia	고발린혈증	
	22	Ornithine Transcarbamylase Deficiency	오르니틴트랜스카바밀효소결핍증	
	23	Defect of Biopterin Cofactor Regeneration	비옵테린 조효소 재합성 결핍증	
	24	Citrullinemia	시트룰린혈증	
	25	Citrullinemia Type Ⅱ	시트룰린혈증 Type Ⅱ	
	26	Argininosuccinic Acidemia	알지노석시닉산혈증	
	27	HHH syndrome	HHH신드롬	
	28	Argininemia	고알리진혈증	
	29	Tyrosinemia Type Ⅰ	고타이로신혈증 Ⅰ형	
	30	Tyrosinemia Type Ⅱ	고타이로신혈증 Ⅱ형	
	31	Tyrosinemia TypeⅢ	고타이로신혈증 Ⅲ형	
	32	Neonatal Tyrosinemia	신생아기 고티로신혈증	
	33	Carbamylphosphate I Synthetase deficiency	카바밀합성효소결핍증	
	34	Hyperornithinemia	고오르니틴혈증	
	35	Glycine encephalopathy (nonketotic hyperglycinemia)	글리신뇌증	
	36	N-Acetylglutamate synthase deficiency	N-아세틸글루타메이트 합성결핍증	▶삭제
	37	S-Adenosylhomocysteine hydrolase deficiency	S-아데노실호모시스틴 분해효소결핍증	▶삭제
	38	Histidinemia	히스티딘혈증	▶삭제
	39	Hyperprolinemia type Ⅰ	고프롤린혈증 Ⅰ형	
	40	Hyperprolinemia type Ⅱ	고프롤린혈증 Ⅱ형	
	41	E3 Deficiency	E3 결핍증	▶삭제
지방산 대사이상질환	42	Glutaric Acidemia Type Ⅱ	글루타릭산혈증 Type Ⅱ	
	43	Short-Chain Acyl CoA Dehydrogenase Deficiency	단쇄아실코에이탈수소효소 결핍증	
	44	Multiple Acyl-CoA Dehydrogenase Deficiency	복합아실코에이탈수소효소 결핍증	▶삭제
	45	Long-Chain 3-HydroxyAcyl-CoA Dehydrogenase Deficiency	장쇄하이드록시아실코에이탈수소효소 결핍증	
	46	Medium Chain Acyl-CoA Dehydrogenase Deficiency	중쇄아실코에이탈수소효소 결핍증	
	47	Very long Chain Acyl-CoA Dehydrogenase Deficiency	초장쇄아실코에이탈수소효소 결핍증	
	48	Carnitine-Acylcarnitine Translocase Deficiency	카르니틴-아실카르니틴전이효소 결핍증	
	49	Carnitine uptake defect	카르니틴 업테이크 결핍증	
	50	Carnitine Palmitoyl Transferase Ⅰ Deficiency	카르니틴팔미토일전이효소 Ⅰ 결핍증	
	51	Carnitine Palmitoyl Transferase Ⅱ Deficiency	카르니틴팔미토일전이효소 Ⅱ 결핍증	
	52	Medium/short-Chain 3-Hydroxy Acyl CoA Dehydrogenase Deficiency	중단쇄하이드록시아실코에이탈수소효소 결핍증	
	53	2,4-Dienoyl CoA Reductase Deficiency	2,4-디에노일코에이환원효소 결핍증	
	54	Mitochondrial Trifunctional Protein Deficiency	미토콘드리아삼중기능성단백질 결핍증	
케톤대사이상질환	55	3-Hydroxy-3-Methylglutaric Aciduria	3-하이드록시-3-메틸글루타릭산뇨증	

변경 후

질환분류	No.	내용		
아미노산 대사이상질환	1	Argininemia (ARG)	알지닌혈증	
	2	Citrullinemia type I (CIT-I)	시트룰린혈증 I형	
	3	Citrullinemia type II (CIT-II)	시트룰린혈증 II형	
	4	Arginosuccinic aciduria (ASA)	알지노석시닌산뇨증	
	5	Pyruvate carboxylase deficiency (PC)	피루브산 카복실레이즈 결핍증	▶추가
	6	Ornithine transcarbamylase deficiency (OTC)	오르니틴트랜스카바밀효소결핍증	
	7	Carbamoyl-phosphate synthase deficiency (CPS)	카바밀합성효소결핍증	
	8	Homocystinuria (HCY)	호모시스틴뇨증	
	9	Hypermethioninemia (MET)	고메티오닌혈증	
	10	CBL E, CBL G, MTHFR deficiency	CBL E, CBL G, MTHFR 결핍증	▶추가
	11	Maple syrup urine disease (MSUD)	단풍당뇨증	
	12	Valinemia (Hyper VAL)	고발린혈증	
	13	Phenylketonuria (PKU)	페닐케톤뇨증	
	14	Disorders of biopterin biosynthesis (BIOPT-BS)	비옵테린 조효소 생합성 결핍증	
	15	Disorders of biopterin regeneration (BIOPT-REG)	비옵테린 조효소 재합성 결핍증	
	16	Tyrosinemia type I (TYR-1)	고타이로신혈증 I형	
	17	Tyrosinemia type II (TYR-II)	고타이로신혈증 II형	
	18	Tyrosinemia type III (TYR-III)	고타이로신혈증 III형	
	19	Transient tyrosinemia of the neonate (TTN)	신생아기 고타이로신혈증	
	20	Nonketotic hyperglycinemia (glycine encephalopathy) (NKHG)	글리신뇌증	
	21	~~Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (HHH)~~	HHH신드롬	
	22	Girate atrophy of the retina (Hyper ORN)	고오르니틴혈증	
	23	Hyperprolinemia type I (PRO I)	고프롤린혈증 I형	
	24	Hyperprolinemia type II (PRO II)	고프롤린혈증 II형	
지방산 대사이상질환	25	Carnitine uptake defect (CUD)	카르니틴 업테이크 결핍증	
	26	Carnitine palmitoyltransferase I deficiency (CPT-Ia)	카르니틴 팔미토일 전이효소 I 결핍증	
	27	Carnitine palmitoyltransferase II deficiency (CPT-II)	카르니틴 팔미토일 전이효소 II 결핍증	
	28	Carnitine-acylcarnitine translocase deficiency (CACT)	카르니틴-아실카르니틴 전이효소 결핍증	
	29	Glutaric acidemia type II (GA-2)	글루타릭산혈증 II형	
	30	Long-chain L-3-Hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)	장쇄하이드록시아실코에이탈수소효소 결핍증	
	31	Trifunctional protein deficiency (TFP)	삼중기능성단백질 결핍증	
	32	Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	중쇄아실코에이탈수소효소 결핍증	
	33	Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)	중쇄케토아실코에이 티올라제결핍증	▶추가
	34	Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	초장쇄아실코에이탈수소효소 결핍증	
	35	2,4-Dienoyl-CoA reductase deficiency (De-Red)	2,4-디에노일코에이환원효소 결핍증	
	36	Short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (SCHAD)	단쇄하이드록시아실코에이탈수소효소 결핍증	
	37	Short-chain acyl-CoA dehydrogenase deficiency (SCAD)	단쇄아실코에이탈수소효소 결핍증	
유기산 대사이상질환	38	Isobutyrylglycinuria (IBG)	이소부티릴글리신뇨증	
	39	Ethylmalonic encephalopathy (EMA)	에틸말로닉 뇌증	
	40	3-Methylglutaconic aciduria (3MGA)	3-메틸글루타코닉산뇨증	
	41	2-Methyl-3-hydroxybutyric aciduria (2M3HBA)	2-메틸-3-하이드록시뷰티릭산뇨증	
	42	3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)	3-메틸크로토닐코에이 탈탄산효소 결핍증	
	43	beta-Ketothiolase deficiency (BKT)	베타케토티올분해효소결핍증	
	44	3-Hydroxy-3-methylglutaric aciduria (HMG)	3-하이드록시-3-메틸글루타릭산뇨증	
	45	Multiple carboxylase deficiency (MCD)	복합탈탄산효소결핍증	
	46	Glutaric acidemia type I (GA-1)	글루타릭산혈증 I형	
	47	Isovaleric acidemia (IVA)	이소발레릭산혈증	
	48	2-Methylbutyrylglycinuria (2MBG)	2-메틸뷰티릴글라이신뇨증	
	49	Malonic acidemia (MAL)	말로닉산혈증	
	50	Propionic acidemia (PROP)	프로피오닉산혈증	
	51	Methylmalonic acidemia (MUT)	메틸말로닉산혈증 (MUT)	
	52	Methylmalonic acidemia (CBL A, CBL B)	메틸말로닉산혈증 (CBL A, CBL B)	
	53	Methylmalonic aciduria and homocystinuria (CBL C, CBL D)	메틸말론산 및 호모시스틴뇨증 (CBL C, CBL D)	
퍼옥시좀질환	54	X-linked adrenoleukodystrophy (X-ALD)	X-관련 부신백질이영양증	▶추가
퓨린대사이상질환	55	Adenosine deaminase-severe combined immunodeficiency (ADA-SCID)	아데노신 디아미나아제 결핍 중증 복합면역결핍증	▶추가