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삼성랩 선천성 대사질환 검사 의뢰서

의뢰병원*		진료과*	
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진단명*			
임상증상			
General physical abnormalities Height ____ cm Weight ____ kg ☐ Megalocephaly ☐ Microcephaly ☐ Congenital anomalies ☐ Splenomegaly ☐ Cardiomegaly ☐ Skin lesion ☐ Strange smell		Gastrointestinal abnormalities ☐ Vomiting ☐ Diarrhea ☐ GI reflux ☐ Constipation Neuromuscular abnormalities ☐ Developmental delay ☐ Mental retardation ☐ Muscle weakness ☐ Hypertonia ☐ Hypotonia ☐ Seizure ☐ Ataxia ☐ Lethargy/coma	
		Hematologic abnormalities ☐ Anemia ☐ Anemia ☐ Neutropenia ☐ Thrombocytopenia ☐ Thrombo-embolic abnormalities ☐ Bleeding tendency Others ☐ Delayed bone age ☐ Hearing loss ☐ Corneal clouding ☐ Recurrent infection ☐ Family history	
관련검사 결과* ☐ Acidosis ☐ Hyperlactic acidemia ☐ Hypoglycemia ☐ Other abnormal findings ☐ Abnormal liver function _____ ☐ Ketosis ☐ Hyperammonemia ☐ Hyperlipidemia		Pedigree	

● 검체 종류

- ☐ Whole blood ☐ Urine ☐ Liver ☐ Muscle ☐ Fibroblast
☐ Others _____

● 의뢰 검사

I. ENZYME TEST

MUCOPOLYSACCHARIDOSIS (MPS)

- ☐ BL3A12 MPS screening
 - ☐ Toluidine blue spot test / CPC quantitation
 - ☐ Thin layer chromatography
- ☐ BL3A1601 MPS I Alpha-L-Iduronidase
- ☐ BL3A1602 MPS II Iduronate sulfatase
- ☐ BL3A1603 MPS IIIa
- ☐ BL3A1604 MPS IIIb N-acetyl-a-D-glucosaminidase
- ☐ BL3A1605 MPS IIIc
- ☐ BL3A1606 MPS IIId
- ☐ BL3A1607 MPS IVa N-acetylgalactosamine-6-sulfatase
- ☐ BL3A1608 MPS IVb Beta-D-galactosidase
- ☐ BL3A1609 MPS VI N-acetylgalactosamine -4-sulfatase
- ☐ BL3A1610 MPS VII Beta-D-glucuronidase
- ☐ BL3A1611 MPS, Others

GLYCOGEN STORAGE DISEASE (GSD)

- ☐ BL3A1501 GSD Ia Glucose-6-phosphatase
- ☐ BL3A1502 GSD Ib Glucose-6-phosphate translocase
- ☐ BL3A1503 GSD II Acid a-glucosidase
- ☐ BL3A1505 GSD IV Branching enzyme
- ☐ BL3A1507 GSD VI Phosphorylase B

METACHROMATIC LEUKODYSTROPHY (MLD)

- ☐ BL3A18 MLD enzyme assay
 - ☐ Arylsulfatase A
 - ☐ Arylsulfatase B

LYSOSOMAL STORAGE DISEASE (LSD)

- ☐ BL3A19 LSD enzyme assay
 - ☐ β -Glucosidase
 - ☐ β - Hexosaminidase

II. GENETIC TEST