

## DANH SÁCH CÁC BỆNH ĐƯỢC SÀNG LỌC TRONG GÓI SLSS CAO CẤP BABYGENE

(Danh sách cập nhật tháng 01.12.2018)

STT	Bệnh lý được sàng lọc	Gene liên quan	PP Điều trị
<b>Bệnh do rối loạn chuyển hóa acid hữu cơ</b>			
1	2,Methyl,3 hydroxybutyric acidemia (3,hydroxy,2,methylbutyryl,CoA dehydrogenase deficiency) (2M3HBA <sup>2</sup> )	<i>HSD17B10</i>	1
2	2,Methylbutyryl,CoA dehydrogenase deficiency (2,Methylbutyryl,CoA dehydrogenase deficiency) (2 MBG <sup>2</sup> )	<i>ACADSB</i>	1
3	3,Hydroxy,3,methylglutaric aciduria (3,hydroxy,3,methylglutaryl,CoA lyase deficiency) (HMG <sup>1</sup> )	<i>HMGCL</i>	1
4	3,Methylcrotonyl,CoA carboxylase deficiency (3,MCC Deficiency) (3,MCC <sup>1</sup> )	<i>MCC1, MCCC2</i>	1
5	3,Methylglutaconic aciduria (3,methylglutaconyl CoA hydratase deficiency) (3MGA <sup>2</sup> )	<i>AUH, DNAJC19, OPA3, TAZ</i>	1
6	Beta,ketothiolase deficiency ( $\beta$ KT <sup>1</sup> )	<i>ACAT1</i>	1
7	Glutaric acidemia type I (GA1 <sup>1</sup> )	<i>GCDH</i>	1
8	Holocarboxylase Synthase deficiency (MCD <sup>1</sup> )	<i>HLCS</i>	1
9	Isobutyryl,CoA dehydrogenase (Isobutyryl,CoA dehydrogenase deficiency) (IBG <sup>2</sup> )	<i>ACAD8</i>	1
10	Isovaleric acidemia (IVA <sup>1</sup> )	<i>IVD</i>	1
11	Malonic acidemia (Malonyl,CoA decarboxylase deficiency) (MAL <sup>2</sup> )	<i>MLYCD</i>	1
12	Methylmalonic acidemia (Cobalamin disorders) (Cbl A <sup>1</sup> )	<i>MMAA</i>	1
13	Methylmalonic acidemia (Cobalamin disorders) (Cbl B <sup>1</sup> )	<i>MMAB</i>	1
14	Methylmalonic acidemia (methylmalonyl,CoA mutase deficiency) (MUT <sup>1</sup> )	<i>MUT</i>	1
15	Methylmalonic acidemia with Homocystinuria (Cbl C <sup>2</sup> )	<i>MMACHC</i>	1
16	Methylmalonic acidemia with Homocystinuria (Cbl D <sup>2</sup> )	<i>MMADHC</i>	1
17	Propionic acidemia (PROP <sup>1</sup> )	<i>PCCA, PCCB</i>	1
<b>Bệnh do rối loạn chuyển hóa acid béo</b>			
18	2,4 Dienoyl,CoA reductase deficiency (DE RED <sup>2</sup> )	<i>DECRI</i>	1
19	Carnitine,acylcarnitine translocase deficiency (CACT <sup>2</sup> )	<i>SLC25A20</i>	1
20	Carnitine palmitoyltransferase type I deficiency (CPT IA <sup>2</sup> )	<i>CPT1A</i>	1
21	Carnitine palmitoyltransferase type II deficiency (CPT II <sup>2</sup> )	<i>CPT2</i>	1
22	Carnitine uptake defect (Primary carnitine deficiency) (CUD <sup>1</sup> )	<i>SLC22A5</i>	1
23	Glutaric acidemia type II (GA2 <sup>2</sup> )	<i>ETFA, ETFB, ETFDH</i>	1
24	Long chain L,3 hydroxyacyl,CoA dehydrogenase deficiency (LCHAD <sup>1</sup> )	<i>HADHA</i>	1
25	Medium,chain acyl,CoA dehydrogenase deficiency (MCAD <sup>1</sup> )	<i>ACADM</i>	1
26	Medium/short,chain L,3,hydroxyacyl,CoA dehydrogenase deficiency (M/SCHAD <sup>2</sup> )	<i>HADH</i>	1
27	Short,chain acyl,CoA dehydrogenase deficiency (SCAD <sup>2</sup> )	<i>ACADS</i>	1
28	Trifunctional protein deficiency (Mitochondrial trifunctional protein deficiency) (TFP <sup>1</sup> )	<i>HADHA, HADHB</i>	1
29	Very long,chain acyl,CoA dehydrogenase deficiency (VLCAD <sup>1</sup> )	<i>ACADVL</i>	1
<b>Bệnh do rối loạn chuyển hóa acid amin</b>			
30	(Argininemia) Arginase deficiency (ARG <sup>2</sup> )	<i>ARG1</i>	1
31	Argininosuccinic aciduria (ASA <sup>1</sup> )	<i>ASL</i>	1
32	Biopterin defect in cofactor biosynthesis (BIOPT,BS <sup>2</sup> )	<i>GCH1, PCBD1, PTS, QDPR</i>	1
33	Biopterin defect in cofactor regeneration (BIOPT,REG <sup>2</sup> )	<i>GCH1, PCBD1, PTS, QDPR</i>	1
34	Benign hyperphenylalaninemia (H,PHE <sup>2</sup> )	<i>PAH</i>	1
35	Citrullinemia type I (CIT <sup>1</sup> )	<i>ASS1</i>	1
36	Citrullinemia type II (CIT II <sup>2</sup> )	<i>SLC25A13</i>	1
37	Homocystinuria (HCY <sup>1</sup> )	<i>CBS, MTHFR, MTR, MTRR</i>	1
38	Hypermethioninemia (MET <sup>2</sup> )	<i>AHCY, GNMT, MATIA</i>	1
39	Maple syrup urine disease (MSUD <sup>1</sup> )	<i>BCKDHA, BCKDHB, DBT, DLD</i>	1

40	Phenylketonuria (PKU <sup>1</sup> )	<i>PAH</i>	1
41	Tyrosinemia Type I (TYR I <sup>1</sup> )	<i>FAH</i>	1
42	Tyrosinemia Type II (TYR II <sup>2</sup> )	<i>TAT</i>	1
43	Tyrosinemia Type III (TYR III <sup>2</sup> )	<i>HPD</i>	1
44	Ornithine Transcarbamylase Deficiency (OTC)	<i>OTC</i>	2
<b>Bệnh lý rối loạn nội tiết và các bệnh Hemoglobin</b>			
45	Congenital adrenal hyperplasia (21,hydroxylase deficiency) (CAH <sup>1</sup> )	<i>CYP21A2, ADA</i>	1
46	Primary congenital hypothyroidism (CH <sup>1</sup> )	<i>DUOX2, PAX8, SLC5A5, TG, TPO, TSHB, TSHR</i>	1
47	S, Beta thalassemia (Hb S/βTh <sup>1</sup> )	<i>HBB</i>	2
48	Sickle cell anemia (S,S Disease) (Hb SS <sup>1</sup> )	<i>HBB</i>	2
49	Sickle cell disease (S,C Disease) (Hb S/C <sup>1</sup> )	<i>HBB</i>	2
50	Alpha thalassemia	<i>HBA1, HBA2</i>	2
51	Structural abnormalities off Hemoglobin (HbD)	<i>HBB</i>	2
52	Structural abnormalities off Hemoglobin (HbD)	<i>HBB</i>	2
53	Various other hemoglobinopathies (Var Hb <sup>2</sup> )	<i>HBA1, HBA2, HBB</i>	2
<b>Một số bệnh lý khác</b>			
54	Biotinidase deficiency (BIOT <sup>1</sup> )	<i>BTD</i>	1
55	Classic Galactosemia (GALT <sup>1</sup> )	<i>GALT</i>	1
56	Cystic Fibrosis (CF <sup>1</sup> )	<i>CFTR</i>	1
57	Galactosepimerase deficiency (Galactosemia type III) (GALE <sup>2</sup> )	<i>GALE</i>	1
58	Galactokinase deficiency (Galactosemia type II) (GALK <sup>2</sup> )	<i>GALK1</i>	1
59	Glycogen Storage Disease Type II (Pompe disease) (GSD II <sup>1</sup> )	<i>GAA</i>	2
60	Nonsyndromic deafness (Hearing loss) (HEAR <sup>1</sup> )	<i>GJB2, GJB3, GJB6</i>	3
61	X,linked Severe combined immunodeficiency (SCID <sup>1</sup> )	<i>IL2RG</i>	2
62	Fabry disease (GLA deficiency) (GLA <sup>R</sup> )	<i>GLA</i>	2
63	Gaucher disease (GBA)	<i>GBA</i>	2
64	Glucose,6,Phosphatase Dehydrogenase Deficiency	<i>G6PD</i>	1
65	Krabbe disease (GALC <sup>R</sup> )	<i>GALC</i>	3
66	Methylmalonyl CoA epimerase deficiency	<i>MCEE</i>	1
67	Mucopolysaccharidosis type I (MPS1) <sup>R</sup>	<i>IDUA</i>	2
68	Niemann,Pick disease <sup>R</sup> Types A	<i>SMPD1</i>	2
69	Niemann,Pick disease <sup>R</sup> Types B	<i>SMPD1</i>	2
70	Niemann,Pick disease <sup>R</sup> Types C1	<i>NPC1</i>	2
71	Niemann,Pick disease <sup>R</sup> Types C2	<i>NPC2</i>	2
72	X,linked adrenoleukodystrophy (ALD <sup>R</sup> )	<i>ABCD1</i>	2

#### A – Danh sách bệnh được sàng lọc:

<sup>1</sup>. Các bệnh lý cơ bản

<sup>2,R</sup>. Các bệnh lý được bổ sung

#### B – Các phương pháp điều trị nhóm bệnh phát hiện bằng Babygene:

1. Bệnh lý được điều trị tốt bằng việc kiểm soát chế độ ăn uống, sinh hoạt hoặc sử dụng các loại thuốc hỗ trợ bổ sung khác.
2. Điều trị bằng các phương pháp xâm lấn (Ghép tủy xương, tế bào gốc, truyền máu, ...).
3. Không có phương pháp điều trị nhưng chất lượng cuộc sống được cải thiện nhờ phát hiện sớm.