IEMs incidence estimates - beta

generated on 2020-04-01

3-Hydroxy 3-methyl glutaric aciduria

1 gene implicated: HMGCL

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	HMGCL	3.4e-04	1.5e-03	0.0 - 0.6
Latino	HMGCL	3.0e-04	2.5e-03	0.0 - 1.6
Ashkenazi Jewish	HMGCL	1.3e-04	1.4e-03	0.0 - 0.5
East Asian	HMGCL	1.3e-04	1.4e-03	0.0 - 0.5
Finnish	HMGCL	2.4e-04	1.4e-03	0.0 - 0.5
Non-Finnish European	HMGCL	1.3e-04	1.7e-03	0.0 - 0.8
Other	HMGCL	4.8e-04	2.6e-03	0.1 - 1.7
South Asian	HMGCL	1.3e-04	1.4e-03	0.0 - 0.5

3-Methylcrotonyl-CoA carboxylase deficiency

2 genes implicated: MCCC1, MCCC2

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	MCCC1	4.0e-04	5.0e-03	0.0 - 6.2
	MCCC2	1.0e-03	3.6e-03	0.3 - 3.3
	total			0.3 - 9.5
Latino	MCCC1	6.8e-04	3.9e-03	0.1 - 3.8
	MCCC2	5.2e-04	3.0e-03	0.1 - 2.2
	total			0.2 - 6.1
Ashkenazi Jewish	MCCC1	1.0e-03	4.3e-03	0.3 - 4.7
	MCCC2	1.3e-03	3.5e-03	0.4 - 3.0
	total			0.7 - 7.7
East Asian	MCCC1	3.3e-04	3.4e-03	0.0 - 2.9
	MCCC2	5.7e-04	3.0e-03	0.1 - 2.2
	total			0.1 - 5.1
Finnish	MCCC1	1.2e-03	3.7e-03	0.3 - 3.4
	MCCC2	1.0e-03	2.9e-03	0.3 - 2.0
	total			0.6 - 5.4
Non-Finnish European	MCCC1	1.8e-04	3.1e-03	0.0 - 2.4
	MCCC2	2.8e-04	3.0e-03	0.0 - 2.3
	total			0.0 - 4.7
Other	MCCC1	7.1e-04	3.4e-03	0.1 - 2.9
	MCCC2	1.8e-03	3.4e-03	0.8 - 2.9
	total			1.0 - 5.8
South Asian	MCCC1	1.9e-04	3.1e-03	0.0 - 2.4
	MCCC2	2.8e-04	2.5e-03	0.0 - 1.6
	total			0.0 - 4.0

Argininosuccinic aciduria

1 gene implicated: ASL

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	ASL	8.8e-04	2.2e-03	0.2 - 1.2
Latino	ASL	5.6e-04	2.1e-03	0.1 - 1.2
Ashkenazi Jewish	ASL	6.0e-04	2.2e-03	0.1 - 1.2
East Asian	ASL	9.7e-04	2.6e-03	0.2 - 1.7
Finnish	ASL	1.3e-03	2.2e-03	0.4 - 1.2
Non-Finnish European	ASL	2.2e-04	2.5e-03	0.0 - 1.5
Other	ASL	8.3e-04	2.3e-03	0.2 - 1.4
South Asian	ASL	2.3e-04	1.9e-03	0.0 - 0.9

Beta-ketothiolase deficiency; Alpha-methylacetoacetic aciduria

1 gene implicated: ACAT1

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	ACAT1	5.1e-04	1.7e-03	0.1 - 0.7
Latino	ACAT1	3.5e-04	1.8e-03	0.0 - 0.8
Ashkenazi Jewish	ACAT1	5.5e-04	4.3e-03	0.1 - 4.7
East Asian	ACAT1	1.8e-04	1.7e-03	0.0 - 0.7
Finnish	ACAT1	4.3e-04	1.7e-03	0.0 - 0.8
Non-Finnish European	ACAT1	1.8e-04	1.6e-03	0.0 - 0.7
Other	ACAT1	6.5e-04	2.0e-03	0.1 - 1.0
South Asian	ACAT1	1.8e-04	1.6e-03	0.0 - 0.7

CBL A,B, methylmalonic acidemia (cobalamin A and B)

2 genes implicated: MMAA, MMAB

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	MMAA	2.4e-04	5.1e-04	0.0 - 0.1
	MMAB	6.3e-04	1.5e-03	0.1 - 0.6
	total			0.1 - 0.6
Latino	MMAA	2.1e-04	5.1e-04	0.0 - 0.1
	MMAB	1.7e-04	1.1e-03	0.0 - 0.3
	total			0.0 - 0.3
Ashkenazi Jewish	MMAA	2.4e-04	5.1e-04	0.0 - 0.1
	MMAB	8.9e-05	1.3e-03	0.0 - 0.5
	total			0.0 - 0.5
East Asian	MMAA	2.7e-04	5.1e-04	0.0 - 0.1
	MMAB	2.4e-04	1.1e-03	0.0 - 0.3
	total			0.0 - 0.4
Finnish	MMAA	6.7e-04	5.1e-04	0.1 - 0.1
	MMAB	5.2e-04	1.3e-03	0.1 - 0.4
	total			0.2 - 0.5
Non-Finnish European	MMAA	1.3e-04	5.1e-04	0.0 - 0.1
	MMAB	9.3e-05	1.0e-03	0.0 - 0.3
	total			0.0 - 0.3
Other	MMAA	5.5e-04	5.1e-04	0.1 - 0.1
	MMAB	6.9e-04	1.2e-03	0.1 - 0.4
	total			0.2 - 0.5
South Asian	MMAA	1.3e-04	5.1e-04	0.0 - 0.1
	MMAB	9.3e-05	1.0e-03	0.0 - 0.3
	total			0.0 - 0.3

Carnitine uptake defect

1 gene implicated: SLC22A5

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	SLC22A5	2.5e-03	6.8e-04	1.5 - 0.1
Latino	SLC22A5	2.0e-03	6.8e-04	1.0 - 0.1
Ashkenazi Jewish	SLC22A5	1.4e-03	6.8e-04	0.5 - 0.1
East Asian	SLC22A5	8.8e-04	6.8e-04	0.2 - 0.1
Finnish	SLC22A5	1.4e-03	6.8e-04	0.5 - 0.1
Non-Finnish European	SLC22A5	2.8e-04	6.8e-04	0.0 - 0.1
Other	SLC22A5	1.6e-03	6.8e-04	0.6 - 0.1
South Asian	SLC22A5	3.0e-04	6.8e-04	0.0 - 0.1

Citrullinemia

1 gene implicated: ASS1

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	ASS1	9.6e-04	2.8e-03	0.2 - 2.0
Latino	ASS1	2.1e-03	3.4e-03	1.1 - 2.9
Ashkenazi Jewish	ASS1	1.6e-03	2.8e-03	0.6 - 2.0
East Asian	ASS1	9.4e-04	2.7e-03	0.2 - 1.8
Finnish	ASS1	1.6e-03	2.9e-03	0.7 - 2.1
Non-Finnish European	ASS1	3.0e-04	2.5e-03	0.0 - 1.6
Other	ASS1	2.0e-03	3.5e-03	1.0 - 3.0
South Asian	ASS1	3.1e-04	2.5e-03	0.0 - 1.6

Galactosemia

1 gene implicated: GALT

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	GALT	1.3e-03	6.7e-03	0.4 - 11.3
Latino	GALT	1.8e-03	3.4e-03	0.9 - 2.9
Ashkenazi Jewish	GALT	2.0e-03	2.8e-03	1.0 - 2.0
East Asian	GALT	1.0e-03	2.6e-03	0.3 - 1.7
Finnish	GALT	2.3e-03	5.1e-03	1.3 - 6.4
Non-Finnish European	GALT	9.0e-04	2.6e-03	0.2 - 1.7
Other	GALT	2.5e-03	3.3e-03	1.6 - 2.8
South Asian	GALT	9.1e-04	2.6e-03	0.2 - 1.7

Glutaric acidemia type I

1 gene implicated: GCDH

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	GCDH	2.2e-03	3.5e-03	1.2 - 3.0
Latino	GCDH	1.4e-03	3.1e-03	0.5 - 2.3
Ashkenazi Jewish	GCDH	1.2e-03	3.2e-03	0.4 - 2.5
East Asian	GCDH	1.1e-03	2.9e-03	0.3 - 2.1
Finnish	GCDH	2.1e-03	3.2e-03	1.1 - 2.6
Non-Finnish European	GCDH	5.0e-04	2.8e-03	0.1 - 2.0
Other	GCDH	1.6e-03	3.5e-03	0.6 - 3.1
South Asian	GCDH	5.1e-04	2.7e-03	0.1 - 1.8

Homocystinuria, due to cystathionine beta-synthase deficiency

1 gene implicated: CBS

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	CBS	2.9e-03	2.6e-03	2.2 - 1.7
Latino	CBS	1.4e-03	3.1e-03	0.5 - 2.4
Ashkenazi Jewish	CBS	1.3e-03	2.7e-03	0.4 - 1.8
East Asian	CBS	6.5e-04	2.5e-03	0.1 - 1.6
Finnish	CBS	3.0e-03	3.1e-03	2.2 - 2.4
Non-Finnish European	CBS	5.0e-04	2.7e-03	0.1 - 1.8
Other	CBS	1.4e-03	3.3e-03	0.5 - 2.7
South Asian	CBS	5.1e-04	2.2e-03	0.1 - 1.2

Isovaleric acidemia

1 gene implicated: IVD

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	IVD	9.4e-04	2.4e-03	0.2 - 1.4
Latino	IVD	6.9e-04	2.7e-03	0.1 - 1.8
Ashkenazi Jewish	IVD	9.2e-04	3.7e-03	0.2 - 3.4
East Asian	IVD	1.7e-04	2.2e-03	0.0 - 1.2
Finnish	IVD	4.4e-04	3.1e-03	0.0 - 2.4
Non-Finnish European	IVD	1.7e-04	2.3e-03	0.0 - 1.4
Other	IVD	3.4e-04	3.8e-03	0.0 - 3.7
South Asian	IVD	1.7e-04	2.2e-03	0.0 - 1.2

Long-chain L-3- hydroxyacyl-CoA dehydrogenase deficiency

1 gene implicated: HADHA

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	HADHA	7.1e-04	9.3e-04	0.1 - 0.2
Latino	HADHA	7.8e-04	9.3e-04	0.2 - 0.2
Ashkenazi Jewish	HADHA	4.2e-04	9.3e-04	0.0 - 0.2
East Asian	HADHA	4.6e-04	9.3e-04	0.1 - 0.2
Finnish	HADHA	1.0e-03	9.3e-04	0.3 - 0.2
Non-Finnish European	HADHA	1.7e-04	9.3e-04	0.0 - 0.2
Other	HADHA	5.2e-04	9.3e-04	0.1 - 0.2
South Asian	HADHA	1.7e-04	9.3e-04	0.0 - 0.2

Maple syrup urine disease

3 genes implicated: DBT, BCKDHB, BCKDHA

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	DBT	3.4e-04	1.6e-03	0.0 - 0.7
	BCKDHB	4.1e-04	1.3e-03	0.0 - 0.4
	BCKDHA	1.1e-03	1.6e-03	0.3 - 0.6
	total			0.4 - 1.7
Latino	DBT	5.7e-04	1.7e-03	0.1 - 0.7
	BCKDHB	8.2e-04	1.1e-03	0.2 - 0.3
	BCKDHA	1.0e-03	1.5e-03	0.3 - 0.5
	total			0.5 - 1.5
Ashkenazi Jewish	DBT	2.5e-04	1.8e-03	0.0 - 0.8
	BCKDHB	2.0e-04	1.0e-03	0.0 - 0.3
	BCKDHA	9.5e-04	1.6e-03	0.2 - 0.7
	total			0.3 - 1.8
East Asian	DBT	5.8e-04	1.5e-03	0.1 - 0.6
	BCKDHB	5.0e-04	1.1e-03	0.1 - 0.3
	BCKDHA	2.0e-04	1.4e-03	0.0 - 0.5
	total			0.2 - 1.4
Finnish	DBT	3.7e-04	1.8e-03	0.0 - 0.8
	BCKDHB	8.0e-04	1.1e-03	0.2 - 0.3
	BCKDHA	5.5e-04	1.6e-03	0.1 - 0.6
	total			0.3 - 1.7
Non-Finnish European	DBT	1.3e-04	1.6e-03	0.0 - 0.6
	BCKDHB	2.0e-04	1.0e-03	0.0 - 0.3
	BCKDHA	2.0e-04	1.4e-03	0.0 - 0.5
	total			0.0 - 1.4
Other	DBT	3.7e-04	1.7e-03	0.0 - 0.7
	BCKDHB	3.7e-04	1.1e-03	0.0 - 0.3
	BCKDHA	4.4e-04	1.5e-03	0.0 - 0.6
	total			0.1 - 1.6
South Asian	DBT	1.3e-04	1.5e-03	0.0 - 0.5
	BCKDHB	2.0e-04	1.0e-03	0.0 - 0.3
	BCKDHA	2.1e-04	1.4e-03	0.0 - 0.5
	total			0.0 - 1.3

Medium-chain acyl-CoA dehydrogenase deficiency

1 gene implicated: ACADM

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	ACADM	2.2e-03	3.8e-03	1.3 - 3.7
Latino	ACADM	1.5e-03	3.7e-03	0.5 - 3.3
Ashkenazi Jewish	ACADM	1.6e-03	3.7e-03	0.6 - 3.3
East Asian	ACADM	7.3e-04	4.4e-03	0.1 - 4.9
Finnish	ACADM	1.9e-03	5.0e-03	0.9 - 6.1
Non-Finnish European	ACADM	4.3e-04	4.0e-03	0.0 - 3.9
Other	ACADM	1.6e-03	3.6e-03	0.6 - 3.3
South Asian	ACADM	4.5e-04	3.0e-03	0.1 - 2.3

Methylmalonyl-CoA mutase deficiency

1 gene implicated: MUT

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	MUT	2.2e-03	4.3e-03	1.2 - 4.6
Latino	MUT	2.4e-03	4.5e-03	1.4 - 5.1
Ashkenazi Jewish	MUT	2.0e-03	5.4e-03	1.0 - 7.2
East Asian	MUT	1.2e-03	3.5e-03	0.3 - 3.1
Finnish	MUT	1.9e-03	3.9e-03	0.9 - 3.8
Non-Finnish European	MUT	7.2e-04	3.3e-03	0.1 - 2.8
Other	MUT	1.6e-03	3.7e-03	0.6 - 3.4
South Asian	MUT	7.2e-04	3.2e-03	0.1 - 2.6

Multiple carboxylase deficiency

1 gene implicated: BTD

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	BTD	1.7e-03	1.4e-03	0.7 - 0.5
Latino	BTD	2.8e-03	1.3e-03	1.9 - 0.4
Ashkenazi Jewish	BTD	1.4e-03	1.7e-03	0.5 - 0.7
East Asian	BTD	6.2e-04	1.2e-03	0.1 - 0.4
Finnish	BTD	1.8e-03	1.3e-03	0.8 - 0.4
Non-Finnish European	BTD	4.5e-04	1.2e-03	0.1 - 0.4
Other	BTD	2.0e-03	1.2e-03	1.0 - 0.4
South Asian	BTD	4.8e-04	1.2e-03	0.1 - 0.4

Phenylketonuria

1 gene implicated: PAH

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	PAH	6.5e-03	3.6e-03	10.6 - 3.2
Latino	PAH	7.9e-03	3.9e-03	15.6 - 3.8
Ashkenazi Jewish	PAH	4.2e-03	2.9e-03	4.5 - 2.1
East Asian	PAH	4.4e-03	2.8e-03	4.9 - 1.9
Finnish	PAH	8.5e-03	5.3e-03	18.1 - 7.0
Non-Finnish European	PAH	1.9e-03	4.0e-03	0.9 - 3.9
Other	PAH	4.6e-03	2.8e-03	5.3 - 2.0
South Asian	PAH	1.9e-03	2.6e-03	0.9 - 1.7

Propionic acidemia

2 genes implicated: PCCB, PCCA

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	PCCB	1.4e-03	7.9e-03	0.5 - 15.7
	PCCA	6.5e-04	3.2e-03	0.1 - 2.5
	total			0.6 - 18.2
Latino	PCCB	8.8e-04	5.6e-03	0.2 - 7.9
	PCCA	3.7e-04	4.7e-03	0.0 - 5.5
	total			0.2 - 13.4
Ashkenazi Jewish	PCCB	1.4e-03	5.9e-03	0.5 - 8.7
	PCCA	8.3e-04	2.9e-03	0.2 - 2.2
	total			0.7 - 10.8
East Asian	PCCB	5.8e-04	4.1e-03	0.1 - 4.1
	PCCA	3.5e-04	3.3e-03	0.0 - 2.8
	total			0.1 - 6.9
Finnish	PCCB	7.9e-04	5.0e-03	0.2 - 6.2
	PCCA	1.1e-03	6.1e-03	0.3 - 9.4
	total			0.4 - 15.5
Non-Finnish European	PCCB	2.8e-04	9.2e-03	0.0 - 21.0
	PCCA	2.0e-04	6.3e-03	0.0 - 9.9
	total			0.0 - 30.9
Other	PCCB	5.1e-04	1.1e-02	0.1 - 32.8
	PCCA	1.1e-03	5.0e-03	0.3 - 6.2
	total			0.4 - 39.0
South Asian	PCCB	2.9e-04	3.8e-03	0.0 - 3.6
	PCCA	2.1e-04	2.5e-03	0.0 - 1.6
	total			0.0 - 5.2

Trifunctional protein deficiency

2 genes implicated: HADHA, HADHB

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	HADHA	7.1e-04	9.3e-04	0.1 - 0.2
	HADHB	3.5e-04	5.8e-04	0.0 - 0.1
	total			0.2 - 0.3
Latino	HADHA	7.8e-04	9.3e-04	0.2 - 0.2
	HADHB	5.7e-04	5.8e-04	0.1 - 0.1
	total			0.2 - 0.3
Ashkenazi Jewish	HADHA	4.2e-04	9.3e-04	0.0 - 0.2
	HADHB	1.0e-03	5.8e-04	0.3 - 0.1
	total			0.3 - 0.3
East Asian	HADHA	4.6e-04	9.3e-04	0.1 - 0.2
	HADHB	1.4e-04	5.8e-04	0.0 - 0.1
	total			0.1 - 0.3
Finnish	HADHA	1.0e-03	9.3e-04	0.3 - 0.2
	HADHB	5.0e-04	5.8e-04	0.1 - 0.1
	total			0.3 - 0.3
Non-Finnish European	HADHA	1.7e-04	9.3e-04	0.0 - 0.2
	HADHB	1.3e-04	5.8e-04	0.0 - 0.1
	total			0.0 - 0.3
Other	HADHA	5.2e-04	9.3e-04	0.1 - 0.2
	HADHB	2.5e-04	5.8e-04	0.0 - 0.1
	total			0.1 - 0.3
South Asian	HADHA	1.7e-04	9.3e-04	0.0 - 0.2
	HADHB	1.4e-04	5.8e-04	0.0 - 0.1
	total			0.0 - 0.3

Tyrosinemia type I

1 gene implicated: FAH

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	FAH	1.3e-03	2.6e-03	0.4 - 1.8
Latino	FAH	8.6e-04	3.7e-03	0.2 - 3.4
Ashkenazi Jewish	FAH	4.9e-04	1.9e-03	0.1 - 0.9
East Asian	FAH	5.3e-04	3.9e-03	0.1 - 3.7
Finnish	FAH	1.5e-03	3.9e-03	0.6 - 3.8
Non-Finnish European	FAH	2.2e-04	4.5e-03	0.0 - 5.0
Other	FAH	1.4e-03	2.6e-03	0.5 - 1.6
South Asian	FAH	2.4e-04	1.8e-03	0.0 - 0.8

Very long-chain acyl-CoA dehydrogenase deficiency

1 gene implicated: ACADVL

Population	Gene	Frequency of known disease variants	Frequency of all (putative) disease related SNVs	Incidence range (per million)
African	ACADVL	1.7e-03	6.9e-03	0.7 - 12.0
Latino	ACADVL	1.7e-03	3.9e-03	0.7 - 3.9
Ashkenazi Jewish	ACADVL	1.4e-03	3.3e-03	0.5 - 2.8
East Asian	ACADVL	6.8e-04	2.9e-03	0.1 - 2.0
Finnish	ACADVL	1.6e-03	3.4e-03	0.6 - 2.9
Non-Finnish European	ACADVL	3.9e-04	2.9e-03	0.0 - 2.0
Other	ACADVL	1.8e-03	3.4e-03	0.8 - 2.9
South Asian	ACADVL	3.9e-04	2.9e-03	0.0 - 2.0