The report below is a scientific interpretation of the DNA variants (referred as “RS” numbers) provided to the customer by a third party (for example 23andMe). The report should not be considered exclusive, because of several reasons: (a) typically the customer provides limited number of DNA variants since the third party company typically does not do whole DNA sequencing, but rather targets particular positions of the customer DNA; (b) some variants may not be interpretable due to lack of clinical data. In addition, it should be stated again that due to the complexity of human genomic data, the observation that the customer has DNA variant which is found to be disease-causing in another individual does not necessarily indicate that the customer has the same high probability to develop the disease. Finally, due to complexity of matching fussy language expressions, a particular RS may be reported to be associated with more than one disease.

PREDICAGEN LLC REPORT

Case: 164

Below we describe the meanings of the symbols/abbreviations used in the report.

ICDM name: Standardized disease names taken in accordance with International Statistical Classifcation of Diseases and Related Health Problems (ICD). This is the name of disease that your primary physician will recognize and use to make decisions about your health.

ICDM ID: Corresponding disease names ID taken from International Statistical Classifcation of Diseases and Related Health Problems (ICD). This is the number (the tag) of disease listed in your primary physician booklet and used by your health insurance provider.

ICDM score: Disease name matching score using ICDM (maximum 100). This is a measure of confidence of matching scientific descriptors and ICD names. The large number(but smaller than 100) indicates high confidence.

Phenotype: The set of observable characteristics of corresponding disease. This is scientific description of effect of the DNA variant(s) on functionality of the corresponding biological macromolecules. If you want to know more how your DNA variants affect the corresponding macromolecules functionality at molecular level, you should contact us at support@predicagen.com (mailto: support@predicagen.com).

RS ID: The genetic variants RS ID. This is a list of DNA variants associated with the corresponding disease. More RS numbers associated with a disease indicate higher probability that you are at risk.

High and medium confidence: The first table "high confidence predictions" list cases for which your DNA variant exactly matches the variant in ClinVar database; the second table "medium confidence predictions" list cases for which you do not have the wild type nucleotide.

High Confidence Predictions

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Other disorders of peroxisome biogenesis	E71518	76	Peroxisome biogenesis disorder 6B	61752095, 28936698
Neuronal ceroid lipofuscinosis	E754	76	Ceroid lipofuscinosis neuronal 1;not provided	137852695
Immunodeficiency, unspecified	D849	82	Immunodeficiency 20;not specified	10127939
Primary open-angle glaucoma, left eye, stage unspecified	H401120	73	Coloboma;Primary open angle glaucoma;not specified	9282671
Choroidal dystrophy (central areolar) (generalized) (peripapillary)	H3122	71	Choroidal dystrophy, central areolar 2;not provided	61755792

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Beta thalassemia	D561	86	beta Thalassemia	34483965, 34809925, 34135787
Delta-beta thalassemia	D562	82	delta Thalassemia	35654785, 63750345
Smith-Lemli-Opitz syndrome	E7872	74	Smith-Lemli-Opitz syndrome;not provided	28938174, 138659167
Mucopolysaccharidosis, type II	E761	74	Mucopolysaccharidosis, MPS-III-A	34946266
Classical phenylketonuria	E700	71	Phenylketonuria	62516142
Tay-Sachs disease	E7502	87	Tay-Sachs disease	28940871
Other GM2 gangliosidosis	E7509	74	Gm2-gangliosidosis, subacute	28942072
Refractory anemia, unspecified	D464	71	Fanconi anemia;not specified	17227403
Congenital bullous ichthyosiform erythroderma	Q803	70	Bullous ichthyosiform erythroderma;not provided	58026994
Osteogenesis imperfecta	Q780	79	Osteogenesis imperfecta type I	67543897, 72645337
Glutaric aciduria type II	E71313	80	Glutaric aciduria, type 1	11085825
Metachromatic leukodystrophy	E7525	76	Metachromatic leukodystrophy;not provided	80338819

Medium Confidence Predictions

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Other disorders of peroxisome biogenesis	E71518	76	Peroxisome biogenesis disorder 6B	61750434, 61752092, 61752119, 61752127, 61752117, 28936697, 61752112
Congenital night blindness	H5363	75	Congenital stationary night blindness	61751398

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Folate deficiency anemia, unspecified	D529	70	Muscle AMP deaminase deficiency;not specified	17602729
Ichthyosis vulgaris	Q800	86	Ichthyosis vulgaris	141784184
Familial hypercholesterolemia	E7801	92	Familial hypercholesterolemia	5742904, 28937579
Hyperstimulation of ovaries	N981	71	Ovarian hyperstimulation syndrome	28928870, 28928871
MERRF syndrome	E8842	75	Perry syndrome	67586389, 72466486, 72466487
Other hypertrophic cardiomyopathy	I422	71	Familial hypertrophic cardiomyopathy 9	28933405
Other hemochromatosis	E83118	73	Hemochromatosis type 4	28939076
Long chain/very long chain acyl CoA dehydrogenase deficiency	E71310	80	Very long chain acyl-CoA dehydrogenase deficiency;not provided	2286963
GM2 gangliosidosis, unspecified	E7500	72	Infantile GM1 gangliosidosis;not specified	72555358, 72555359, 72555361
Morquio B mucopolysaccharidoses	E76211	72	Mucopolysaccharidosis, MPS-IV-B	72555362, 72555364, 72555365, 72555367, 72555368, 72555369
Choroidal dystrophy (central areolar) (generalized) (peripapillary)	H3122	71	Choroidal dystrophy, central areolar 2;not provided	61755793
Disease of pancreas, unspecified	K869	70	Parkinson disease 2;not specified	55774500
Malignant carcinoid tumor of the stomach	C7A092	70	Malignant tumor of prostate	72552387
Achromatopsia	H5351	76	Achromatopsia 3	6471482

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Non-ketotic hyperglycinemia	E7251	92	Non-ketotic hyperglycinemia	121964974
Familial dysautonomia [Riley-Day]	G901	73	Familial dysautonomia;not provided	111033171
Hemophagocytic lymphohistiocytosis	D761	81	Hemophagocytic lymphohistiocytosis, familial, 2	28933374, 28933376
Beta thalassemia	D561	86	beta Thalassemia	35703285, 63750954, 34500389
Neuronal ceroid lipofuscinosis	E754	90	Ceroid lipofuscinosis neuronal 2	28940573
Transient neonatal myasthenia gravis	P940	71	Cyanosis, transient neonatal	34878913
Delta-beta thalassemia	D562	82	delta Thalassemia	34975911, 35324967, 35518301, 35887507
Congenital central alveolar hypoventilation syndrome	G4735	77	Congenital central hypoventilation	8192466
Smith-Lemli-Opitz syndrome	E7872	91	Smith-Lemli-Opitz syndrome	61757582, 80338859, 80338862
Aggressive periodontitis, unspecified	K0520	71	Periodontitis, aggressive, 1	28937571
Dystonia, unspecified	G249	79	Dystonia;not specified	1800496
Hereditary vitamin D-dependent rickets (type 1) (type 2)	E8332	78	Vitamin D-dependent rickets, type 1	28934605, 28934607
Congenital bullous ichthyosiform erythroderma	Q803	70	Bullous ichthyosiform erythroderma;not provided	61616632, 57784225, 58075662, 58414354, 60035576, 60118264
Classical phenylketonuria	E700	71	Phenylketonuria	62508752
Wilson's disease	E8301	80	Wilson disease	28942076

ICDM Name	ICDM ID	ICDM Score	Phenotype	RS IDs
Other specified viral diseases	B338	73	Wilson disease;not specified	61733679, 137853283
Obstructive hypertrophic cardiomyopathy	I421	79	Hypertrophic cardiomyopathy;not provided	45442096
Alpha-1-antitrypsin deficiency	E8801	91	Alpha-1-antitrypsin deficiency	61761869
Keratosis punctata (palmaris et plantaris)	L852	72	Keratosis palmoplantaris papulosa	200564757
Other GM2 gangliosidosis	E7509	71	Gm2-gangliosidosis, late onset	28941771
Medullary cystic kidney	Q615	75	Medullary cystic kidney disease 2	28934584
Macular corneal dystrophy	H1855	79	Macular corneal dystrophy Type I	72547544
Osteogenesis imperfecta	Q780	79	Osteogenesis imperfecta type I	8179178, 66523073, 67394386, 67445413, 67815019, 67828806, 72645323, 72645328, 72645331, 72645334, 72645341, 72645356, 72648320, 72651614, 72651635, 72656314, 72656340, 72667031, 72667037
Niemann-Pick disease type C	E75242	90	Niemann-Pick disease type C1	28940897, 28942106
3-methylglutaconic aciduria	E71111	82	3-Methylglutaconic aciduria type 3	28937899
Dressler's syndrome	I241	70	Kindler's syndrome	121918293, 146180696
Hermansky-Pudlak syndrome	E70331	88	Hermansky-Pudlak syndrome 4	119471023
Metachromatic leukodystrophy	E7525	77	Metachromatic leukodystrophy, adult type	28940894

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