

| Chromosome | Position | Accession | Disorder |
|------------|-----------|-----------------|--|
| 1 | 17028628 | RCV000013628.25 | Paragangliomas |
| 1 | 17033078 | RCV000013616.18 | Paragangliomas |
| 1 | 155239934 | RCV000004565.4 | Gaucher's disease, type 1 |
| 1 | 247424041 | RCV000004619.3 | Familial cold urticaria |
| 2 | 166272731 | RCV000006728.2 | Paroxysmal extreme pain disorder |
| 2 | 232540072 | RCV000020009.28 | Multiple pterygium syndrome Escobar type |
| 3 | 184354643 | RCV000201810.1 | Leukoencephalopathy with ataxia |
| 4 | 1001475 | RCV000178733.1 | Hurler syndrome |
| 4 | 6301914 | RCV000023510.4 | Diabetes mellitus AND insipidus with optic atrophy AND deafness |
| 4 | 83267737 | RCV000195593.1 | not provided |
| 5 | 1253798 | RCV000032395.1 | Idiopathic fibrosing alveolitis, chronic form |
| 6 | 32038514 | RCV000012938.3 | 21-hydroxylase deficiency |
| 6 | 32042353 | RCV000133609.3 | Ehlers-Danlos-like syndrome due to tenascin-X deficiency |
| 6 | 43519412 | RCV000186584.2 | Leukodystrophy, hypomyelinating, 11 |
| 7 | 65967863 | RCV000000948.4 | Mucopolysaccharidosis type VII |
| 7 | 66086763 | RCV000194332.1 | Argininosuccinate lyase deficiency |
| 8 | 47931496 | RCV000142391.4 | Immunodeficiency 26 with or without neurologic abnormalities |
| 8 | 116847588 | RCV000198309.1 | Cornelia de Lange syndrome 4 |
| 9 | 105617967 | RCV000003371.3 | Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies, type A4 |
| 9 | 135775346 | RCV000032796.2 | Early infantile epileptic encephalopathy 14 |
| 10 | 47322871 | RCV000074345.2 | Telangiectasia, hereditary hemorrhagic, type 5 |
| 10 | 124401788 | RCV000049547.1 | Ornithine aminotransferase deficiency |
| 11 | 2445479 | RCV000057665.2 | Congenital long QT syndrome |
| 11 | 47332111 | RCV000158263.1 | Cardiomyopathy |
| 11 | 47332133 | RCV000154451.1 | Familial hypertrophic cardiomyopathy 4 |
| 11 | 47332154 | RCV000158255.1 | Cardiomyopathy |
| 11 | 47332173 | RCV000158461.1 | Cardiomyopathy |
| 11 | 47332580 | RCV000151066.1 | Primary familial hypertrophic cardiomyopathy |
| 11 | 47333658 | RCV000158214.1 | Cardiomyopathy |
| 11 | 47335942 | RCV000158181.1 | Cardiomyopathy |
| 13 | 32340392 | RCV000131111.2 | Hereditary cancer-predisposing syndrome |
| 13 | 32379872 | RCV000131038.2 | Hereditary cancer-predisposing syndrome |

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| 15 | 43604408 | RCV000151950.1 | Deafness, autosomal recessive 16 |
| 15 | 68211853 | RCV000023573.4 | Adult neuronal ceroid lipofuscinosis |
| 16 | 88430169 | RCV000114795.1 | Keratoconus 1 |
| 19 | 11123264 | RCV000172965.1 | Familial hypercholesterolemia |
| 19 | 11448917 | RCV000014147.25 | Congenital cystic disease of liver |
| 19 | 12896644 | RCV000207433.1 | Glutaric aciduria, type 1 |
| 19 | 17816945 | RCV000015957.25 | Cryptorchidism, unilateral or bilateral |
| 19 | 34394044 | RCV000014610.26 | Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency |
| 19 | 35842480 | RCV000049885.1 | Finnish congenital nephrotic syndrome |
| 19 | 35848334 | RCV000049845.1 | Finnish congenital nephrotic syndrome |
| 20 | 44623042 | RCV000002044.2 | Partial adenosine deaminase deficiency |
| 20 | 63439610 | RCV000187884.1 | not provided |
| 22 | 18918451 | RCV000004224.5 | Proline dehydrogenase deficiency |