

term	p-value	q-value
Non-epidermolytic palmoplantar keratoderma ORPHA:2337	4,87E-05	0,031062
Distal 17p13.3 microdeletion syndrome ORPHA:261257	0,001078	0,171985
Sjogren-Larsson syndrome ORPHA:816	0,001078	0,171985
Bamforth-Lazarus syndrome ORPHA:1226	0,001078	0,171985
Amyotrophic lateral sclerosis ORPHA:803	0,002581	0,199151
White sponge nevus ORPHA:171723	0,017063	0,199151
Giant axonal neuropathy ORPHA:643	0,017386	0,199151
Glutamate-cysteine ligase deficiency ORPHA:33574	0,017386	0,199151
Wolman disease ORPHA:75233	0,017386	0,199151
X-linked Ehlers-Danlos syndrome ORPHA:75497	0,017386	0,199151
Neuronal intestinal pseudoobstruction ORPHA:99811	0,017386	0,199151
Neuropathy with hearing impairment ORPHA:139512	0,017386	0,199151
X-linked hypophosphatemia ORPHA:89936	0,017386	0,199151
OBSOLETE: Alpha-1-antichymotrypsin deficiency ORPHA:93594	0,017386	0,199151
X-linked keloid scarring-reduced joint mobility-increased optic cup-to-disc ratio syndrome ORPHA:482606	0,017386	0,199151
Oculocutaneous albinism type 3 ORPHA:79433	0,017386	0,199151
Otopalatodigital syndrome type 2 ORPHA:90652	0,017386	0,199151
Otopalatodigital syndrome type 1 ORPHA:90650	0,017386	0,199151
NON RARE IN EUROPE: Glucose-6-phosphate-dehydrogenase deficiency ORPHA:362	0,017386	0,199151
PLIN1-related familial partial lipodystrophy ORPHA:280356	0,017386	0,199151
Pancreatic insufficiency-anemia-hyperostosis syndrome ORPHA:199337	0,017386	0,199151
Cerebellar-facial-dental syndrome ORPHA:444072	0,017386	0,199151
Partial corpus callosum agenesis-cerebellar vermis hypoplasia with posterior fossa cysts syndrome ORPHA:401959	0,017386	0,199151
Hyperlipidemia due to hepatic triacylglycerol lipase deficiency ORPHA:140905	0,017386	0,199151
Cholesteryl ester storage disease ORPHA:75234	0,017386	0,199151
Class I glucose-6-phosphate dehydrogenase deficiency ORPHA:466026	0,017386	0,199151
Infantile-onset X-linked spinal muscular atrophy ORPHA:1145	0,017386	0,199151
Odonto-onycho-dermal dysplasia ORPHA:2721	0,017386	0,199151
Autosomal recessive extra-oral halitosis ORPHA:562538	0,017386	0,199151
NON RARE IN EUROPE: Wolff-Parkinson-White syndrome ORPHA:907	0,017386	0,199151
Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease ORPHA:439854	0,017386	0,199151
Pyruvate dehydrogenase phosphatase deficiency ORPHA:79246	0,017386	0,199151
19p13.3 microduplication syndrome ORPHA:447980	0,017386	0,199151
1p31p32 microdeletion syndrome ORPHA:401986	0,017386	0,199151
3-hydroxy-3-methylglutaryl-CoA synthase deficiency ORPHA:35701	0,017386	0,199151
KLHL9-related early-onset distal myopathy ORPHA:399081	0,017386	0,199151
Terminal osseous dysplasia-pigmentary defects syndrome ORPHA:88630	0,017386	0,199151
AA amyloidosis ORPHA:85445	0,017386	0,199151
Schopf-Schulz-Passarge syndrome ORPHA:50944	0,017386	0,199151
Sebocystomatosis ORPHA:841	0,017386	0,199151
MUC1-related autosomal dominant tubulointerstitial kidney disease ORPHA:88949	0,017386	0,199151
Malan overgrowth syndrome ORPHA:420179	0,017386	0,199151
Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta ORPHA:566243	0,017386	0,199151
Melnick-Needles syndrome ORPHA:2484	0,017386	0,199151
Spinocerebellar ataxia type 10 ORPHA:98761	0,017386	0,199151
Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency ORPHA:319595	0,017386	0,199151
FLNA-related X-linked myxomatous valvular dysplasia ORPHA:555877	0,017386	0,199151
Atypical dentin dysplasia due to SMOC2 deficiency ORPHA:314721	0,017386	0,199151
Spondylo-ocular syndrome ORPHA:85194	0,017386	0,199151
Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome ORPHA:391487	0,017386	0,199151
Familial benign flecked retina ORPHA:363989	0,017386	0,199151
Marshall-Smith syndrome ORPHA:561	0,017386	0,199151
Familial hyperprolactinemia ORPHA:397685	0,017386	0,199151
Susceptibility to viral and mycobacterial infections due to STAT1 deficiency ORPHA:391311	0,017386	0,199151
Bladder exstrophy ORPHA:93930	0,017712	0,199151
Autosomal recessive infantile hypercalcemia ORPHA:300547	0,017712	0,199151
Familial papillary or follicular thyroid carcinoma ORPHA:319487	0,01804	0,199151
Renal tubular dysgenesis of genetic origin ORPHA:97369	0,01837	0,199151
Athyreosis ORPHA:95713	0,018704	0,199151