

RD0040	2686	Cyclic neutropenia	0.1ad	0	0	0	0	0	0	0	0	0
RD0050	379	Chronic granulomatous disease	0.29m	0	0	0.57ad	0	0	0	0	0	0
RF0010	726	Alpers syndrome	0.2cd	0	0	0	0	0	0	0	0	2.62cd
RF0030	506	Leigh syndrome	0.2ad	0	0	0.57ad	0	0	0	0	0	0
RF0040	778	Rett syndrome	0.1ad	0	0	0.57ad	0	0	0	0	0	0
RF0060	98261	Progressive myoclonic epilepsy	0.2cd	0	0	0	0	0	0	0	0	0
RF0100	803	Amyotrophic lateral sclerosis	0.2m	0	0	0	0	0	0	0	0	0
RF0130	2382	Lennox-Gastaut syndrome	0.1cd	0	0	0	0	0	0	0	0	1.31cd
RF0140	3451	West syndrome	5.69m	0	0		0	0	4.66cd	21.32cd	9.04cd	6.56cd
RF0200	891	Familial exudative vitreoretinopathy	0.1cd	1.4cd	0	0	0	0	0	0	0	0
RF0280	156071	Keratoconus	0.1cd	0	0	0	0	0	0	0	0	0
RG0040	2331	Kawasaki disease	4.31m	4.19ad	0	3.97ad	0	0	4.66cd	5.33cd	4.52cd	
RG0100	774	Hereditary hemorrhagic telangiectasia	0.1cd	0	0	0	0	0	0	0	0	0
RI0030	2070	Eosinophilic gastroenteritis	0.29cd	0	0	0	0	0	0	5.33cd	0	0
RI0040		Chronic intestinal pseudoobstruction	0.1cd	0	0	0.57cd	0	0	0	0	0	0
RI0070	2290	Microvillus inclusion disease (MVID)	0.1ad	1.4ad	0	0	0	0	0	0	0	0
RI0080	36204	Intestinal lymphangiectasia	0.2cd	0	0	0	9.35cd	0	1.17cd	0	0	0
RL0040		Bullous pemphigoid	0.1ad	0	0	0	0	0	0	0	0	0
RN0010		Arnold-Chiari malformation	1.86m	4.19ad	0	0.57ad	0	0	2.33cd	0	0	1.31cd
RN0020	199642	Microcephaly	0.29m	0	0	0	0	0	0	0	0	1.31cd
RN0040	475	Joubert syndrome	0.2m	0	0	0	0	0	1.17cd	0	0	0
RN0050		Lissencephaly	1.08m	0	0	1.13ad	0	0	1.17cd	0	0	1.31cd
RN0060	2162	Holoprosencephaly	0.59m	1.4ad	0	0	0	0	0	0	0	0
RN0090		Axenfeld-Rieger anomaly	0.1ad	0	0	0	0	0	0	0	0	0
RN0100	708	Peters anomaly	0.1cd	0	0	0	0	0	1.17cd	0	0	0
RN0110	77	Aniridia	0.49m	0	0	0	0	0	0	0	0	2.62cd
RN0120		Congenital colobomatous optic disc	0.69m	0	0	0	9.35cd	9.74cd		0	0	1.31cd
RN0140		Pupillar membrane persistence	0.1cd	0	0	0	0	0	0	0	0	1.31cd
RN0160		Esophageal atresia and/or Isolated tracheo-esophageal fistula	6.67m	20.95ad	46.17ad	3.4ad	0	0	12.82cd	15.99cd	4.52cd	7.87cd
RN0170	1201	Atresia of small intestine	1.67m	8.38ad	0	0.57ad	0	0	1.17cd	5.33cd	13.55cd	0
RN0180	1203	Duodenal atresia	1.86m	12.57ad	0	2.27ad	0	0	2.33cd	0	4.52cd	2.62cd
RN0190	96346	Anorectal malformation	6.37m	15.36ad	46.17ad	1.7ad	9.35cd	9.74cd	18.65cd	10.66cd	27.11cd	13.12cd

RN0200	388	Hirschsprung disease	4.22m	8.38ad	0		0	29.23cd	9.32cd	15.99cd	9.04cd	7.87cd
RN0210		Biliary atresia	3.14m	1.4ad	0	1.7ad	0	0		5.33cd	9.04cd	2.62ad
RN0310	2345	Isolated Klippel-Feil syndrome	0.2cd	0	0	0	0	9.74cd	1.17cd	0	0	0
RN0320	2368	Gastroschisis	1.18m	0	0		0	0	1.17cd	0	9.04cd	2.62cd
RN0330	98249	Ehlers-Danlos syndrome	0.1ad	1.4ad	0	0	0	0	0	0	0	0
RN0340	974	Adams-Oliver syndrome	0.2m	0	0	0	0	0	1.17cd	0	0	0
RN0380	3255	Filippi syndrome	0.1ad	0	0	0	0	0	0	0	0	0
RN0390	380	Greig cephalopolysyndactyly syndrome	0.2ad	0	0	0	0	0	0	0	0	0
RN0400	1540	Jackson-Weiss syndrome	0.1ad	0	0	0	0	0	1.17ad	0	0	0
RN0410	2311	Jarcho-Levin syndrome	0.1cd	1.4cd	0	0	0	0	0	0	0	0
RN0430	2911	Poland syndrome	1.27m		0	1.13cd	0	0	0	5.33cd	0	0
RN0470	669	Otopalatodigital syndrome	0.1ad	0	0	0	0	0	0	0	0	0
RN0510	464	Incontinentia pigmenti	0.78m	2.79ad	0	0	0	0	0	0	0	3.93cd
RN0540	1556	Cutis marmorata telangiectatica congenita	0.1cd	0	0	0	0	0	0	0	0	1.31cd
RN0570		Epidermolysis bullosa	1.37m	0	0	0	0	0	1.17cd	5.33cd	0	1.31cd
RN0640	1114	Aplasia cutis congenita	0.1ad	0	0	0	0	0	0	0	0	0
RN0660	870	Down syndrome	35m	60.05ad	0		168.3cd	19.48cd	60.61cd	10.66cd	40.66cd	17.05cd
RN0670	281	Cri du chat syndrome	0.29m	0	0	0.57ad	0	0	0	0	0	0
RN0680	881	Turner syndrome	1.96m	0	0	0.57ad	0	9.74cd	2.33cd	0	4.52cd	1.31cd
RN0690		Klinefelter syndrome	2.65m	5.59ad	0	0.57ad	9.35cd	0	5.83cd	5.33cd	4.52cd	6.56cd
RN0700	280	Wolf-Hirschhorn syndrome	0.2m	0	0	0	0	0	0	0	0	0
RN0710	550	MELAS	0.59ad	0	0	0	0	0	0	0	0	1.31ad
RN0740		Ivemark syndrome	0.1cd	0	0	0	0	0	1.17cd	0	0	0
RN0750	805	Tuberous sclerosis	2.84m	5.59ad	0	1.7ad	0	9.74cd		0	9.04cd	6.56cd
RN0770	3205	Sturge-Weber syndrome	0.69m	1.4ad	0	0	0	0	2.33cd	0	0	1.31cd
RN0790	915	Aarskog-Scott syndrome	0.1ad	0	0	0	0	0	0	0	0	0
RN0810	1225	Baller-Gerold syndrome	0.1ad	0	0	0	0	0	0	0	0	0
RN0820	116?	Beckwith-Wiedemann syndrome	2.65m	6.98ad	0	0	9.35cd	0	4.66cd	0	0	2.62cd
RN0850	138	CHARGE syndrome	1.18m	1.4ad	0	1.13ad	0	0	1.17cd	5.33ad	9.04cd	1.31cd
RN0860	3157	De Morsier syndrome	0.2ad	1.4ad	0	0	0	0	0	0	0	0
RN0880	1896	EEC syndrome	0.59m	2.79ad	0	0	0	0	0	0	0	0
RN0890	2053	Freeman-Sheldon syndrome	0.1cd	0	0	0	0	0	1.17cd	0	0	0
RN0900	2059	Fryns syndrome	0.1cd	0	0	0	0	0	1.17cd	0	0	0

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RBG010	Neurofibromatoses	3.92m	8.38ad	0	3.4ad	0	0	3.5cd	10.66cd	4.52cd	7.87cd
RCG010	Primary hyperaldosteronisms	0.2m	0	0	0	0	0	0	0	4.52cd	0
RCG020	Congenital adrenogenital syndromes	3.14m		0	0.57ad	0	0	4.66cd	5.33cd	0	2.62cd
	Disturbances of aminoacid transport and metabolism										
RCG040		30.1m	27.93ad	0	31.74ad	18.7cd	0	13.99cd	58.63cd		20.98cd
RCG050	Urea cycle disturbances	1.18m	0	0	0	0	0	3.5cd	0	0	1.31ad
	Disturbances of carbohydrate transport and metabolism, excluded diabetes mellitus										
RCG060		5.39m	6.98ad	0	4.53ad	0	0		0	0	6.56cd
	Congenital alterations of lipoprotein metabolism, excluded: heterozygous familial hypercholesterolhaemia type IIa and IIb; polygenic primary hypercholesterolhaemia; combined familial hypercholesterolhaemia;										
RCG070	Hyperlipoproteinemia type III	3.82m	2.79ad	0	2.27ad	9.35cd	0	0	5.33cd	0	15.74cd
RCG080	Lipid storage disturbances	0.98m	1.4ad	0	1.13ad	0	0	0	5.33cd	0	0
RCG090	Mucopolysaccharidoses	0.1ad	0	0	0	0	0	0	0	0	0
RCG100	Congenital alterations of iron metabolism	0.1cd	0	0	0	0	0	0	0	0	0
RCG140	Mucopolysaccharidoses	0.2m	0	0	0	0	0	1.17cd	0	0	0
RCG150	Chronic histiocytoses	0.59m	2.79ad	0	0	0	0	0	0	0	0
RCG160	Primary immunodeficiencies	4.9m	4.19ad	0	3.97ad	9.35cd	0		5.33cd	0	2.62cd
RDG010	Hereditary anemias	20.69m	30.72ad	0	2.27ad	28.05cd	19.48cd	103.74cc	5.33cd	27.11cd	38.03cd
RDG020	Hereditary coagulation defects	5.49m	11.17ad	0	3.97ad	9.35cd	0	5.83cd	0	27.11cd	6.56cd
RDG030	Hereditary thrombocytopathies	0.1ad	1.4ad	0	0	0	0	0	0	0	0
RDG040	Primary hereditary thrombocytopenias	0.2m	1.4ad	0	0.57cd	0	0	0	0	0	0
RFG010	Leucodystrophies	0.39m	0	0	0	0	0	1.17cd	0	0	0
RFG030	Gangliosidoses	0.78m	0	0	0.57ad	0	0	1.17cd	0	0	1.31cd
RFG040	Spinocerebellar diseases	0.29ad	0	0	0	0	0	0	0	0	0
RFG050	Spinal muscular atrophies	3.24m	1.4ad	0	1.13ad	0	0	5.83cd	0	4.52cd	2.62cd
RFG060	Hereditary neuropathies	0.59m	1.4ad	0	0	0	0	0	0	0	1.31cd
RFG070	Hereditary congenital miopathies	0.49m	0	0	0	0	0	1.17cd	0	0	0
RFG080	Muscular dystrophies	1.57m	0	0	0.57ad	0	0	0	5.33cd	4.52cd	1.31ad

RFG090	Miotonic dystrophies	0.69m	0	0	0.57ad	0	0	1.17cd	0	4.52cd	1.31cd
RFG110	Hereditary retinic dystrophies	1.08m	0	0	0	0	0	2.33cd	0	0	
	Undifferentiated connective tissue										
RMG010	syndromes	0.1ad	0	0	0	0	0	0	0	0	0
RNG010	Pseudohermaphroditisms	1.08m	4.19ad	0	0.57ad	0	0	1.17cd	0	0	1.31cd
RNG020	Multiple congenital arthrogryposes	0.69m	0	0	0	0	0	0	0	0	1.31cd
RNG030	Acrocephalosyndactyly	0.1ad	0	0	0	0	0	0	0	0	0
RNG040	Congenital craniofacial anomalies	21.57m	19.55ad	0	13.6ad	74.8cd		72.27cd	10.66cd		41.97cd
RNG050	Congenital chondrodystrophies	3.53m	12.57ad	0	2.27ad	9.35cd	0	2.33cd	0	4.52cd	1.31cd
RNG060	Congenital osteodystrophies	2.84m	5.59ad	0	0	9.35cd	0	0	0	0	3.93cd
RNG070	Congenital ichthyoses	2.94m	11.17ad	0	0	0	9.74ad	3.5cd	0	9.04cd	1.31cd
RNG080	Chromosomal aneuploidy syndromes	2.55m	4.19ad	0	2.27ad	9.35cd	0	1.17cd	0	0	0
	Chromosomal duplication/deficiency										
RNG090	syndromes	8.73m	6.98ad	0	3.4ad	18.7cd	9.74cd	13.99cd	5.33ad	18.07cd	18.36cd
	Other congenital anomalies with intellectual										
RNG100	disability	2.84m	4.19ad	0	1.13ad	0	0	1.17cd	0	0	0

Supplementary File 4 - Table S4.2

Live births in the period 2012-2013		59832	14947	24934	101456	19550	4187	105169	65128	8389	33514	85376	23454
RNMR Code	ORPHA Code	Toscana	Umbria	Marche	Lazio	Abruzzo	Molise	Campania	Puglia	Basilicata	Calabria	Sicilia	Sardegna
RB0010	654	3.34ad	0	0	1.97ad	0	0	0.95ad	1.54ad	0	2.98ad	0	0
RB0020	790	0	6.69ad	0	2.96ad	30.69ad	0	2.85ad	6.14ad	0	5.97ad	0	0
RB0050	733	0	0	0	0	0	0	0.95ad	1.54cd	0	0	0	0
RC0010	199296	0	0	0	0	0	0	0	0	0	0	1.17cd	0
RC0050	508	0	0	4.01ad	0	0	0	0	0	0	0	0	0
RC0170		0	0	0	0	0	0	0	0	0	0	0	0
RC0180	205	3.34ad	0	0	0	0	0	0	0	0	0	0	0
RD0010		0	0	0	0	0	0	0	0	11.92ad	0	0	0

RD0040	2686	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RD0050	379	0	0	0	0	0	0	0	1.54cd	0	2.98ad	0	0
RF0010	726	0	0	0	0	0	0	0	0	0	0	0	0
RF0030	506	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RF0040	778	0	0	0	0	0	0	0	0	0	0	0	0
RF0060	98261	0	0	0	0	0	0	0	0	0	0	2.34cd	0
RF0100	803	0	0	0	0.99ad	0	0	0	1.54cd	0	0	0	0
RF0130	2382	0	0	0	0	0	0	0	0	0	0	0	0
RF0140	3451	3.34ad	26.76ad	4.01ad	12.81ad	15.35ad	0	0.95cd	1.54ad	0	14.92ad		4.26cd
RF0200	891	0	0	0	0	0	0	0	0	0	0	0	0
RF0280	156071	0	0	0	0	0	0	0	1.54cd	0	0	0	0
RG0040	2331	3.34ad	6.69cd	8.02ad	13.8ad	5.12ad	0	0	0	0	2.98ad	3.51ad	0
RG0100	774	0	0	0	0	0	0	0	1.54cd	0	0	0	0
RI0030	2070	0	0	0	0	0	0	0	1.54cd	0	2.98cd	0	0
RI0040		0	0	0	0	0	0	0	0	0	0	0	0
RI0070	2290	0	0	0	0	0	0	0	0	0	0	0	0
RI0080	36204	0	0	0	0	0	0	0	0	0	0	0	0
RL0040		0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN0010		0	6.69ad	0	1.97ad	5.12ad	23.88ad	0.95ad	1.54ad	23.84ad	0	1.17cd	8.53ad
RN0020	199642	0	0	0	0.99ad	0	0	0	0	0	2.98ad	0	0
RN0040	475	0	0	0	0	0	0	0	0	0	0	1.17ad	0
RN0050		0	0	0	0.99ad	0	0	0	1.54cd	35.76ad	2.98ad	1.17ad	0
RN0060	2162	1.67ad	0	0	0	0	0	0		0	0	1.17cd	4.26ad
RN0090		0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN0100	708	0	0	0	0	0	0	0	0	0	0	0	0
RN0110	77	0	0	0	0.99ad	0	0	0.95cd	0	0	0	1.17ad	0
RN0120		0	0	4.01ad	0	0	0	0	0	0	0	0	0
RN0140		0	0	0	0	0	0	0	0	0	0	0	0
RN0160		10.03ad	0	4.01ad	8.87ad	25.58ad	23.88ad	0.95cd	1.54cd	0	0	1.17cd	0
RN0170	1201	0	0	8.02ad	0.99ad	0	0	0	3.07cd	0	0	0	0
RN0180	1203	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN0190	96346	6.69ad	0	8.02ad	0.99ad	10.23ad	0	0	6.14cd	0	2.98cd	0	0

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RN0910	374	6.69ad	0	0	1.97ad	0	0	0	3.07cd	0	2.98ad	3.51ad	4.26cd
RN0940	2322	1.67ad	0	0	0	0	0	0	0	0	0	0	0
RN0950	98861	0	6.69ad	0	0	0	0	0	0	0	0	1.17ad	0
RN0990	570	0	0	0	0.99ad	0	0	0	1.54cd	0	0	0	0
RN1000	245	0	0	0	0	0	0	0	0	0	0	0	0
RN1010	648	1.67ad	0	8.02ad	7.89ad	0	0	0.95cd	0	23.84ad	2.98ad	3.51ad	0
RN1040	710	0	0	0	0	0	0	0	0	0	0	0	0
RN1080	813	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN1130	1297	0	0	0	0	0	0	0	0	0	0	0	0
RN1140	107	0	0	0	0	0	0	0	0	0	0	0	0
RN1170	744	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN1180	324764	0	0	0	0	0	0	0.95cd	0	0	0	0	0
RN1210	819	0	0	0	0	0	0	0	0	0	0	1.17ad	0
RN1220	828	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN1240	857	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RN1250	887	0	0	0		0	0	0	1.54cd	0	0	0	0
RN1270	904	3.34ad	0	0	1.97ad	5.12ad	0	0.95ad	0	0	2.98ad	1.17ad	0
RN1300	72	0	0	4.01ad	0	0	0	0	0	0	0	0	0
RN1310	739	0	6.69ad	0	1.97ad	5.12ad	0		3.07cd	0	0	3.51ad	12.79ad
RN1320	558	0	0	0	0	0	0	0	0	0	0	0	0
RN1330	908	1.67ad	0	0	0	0	0	0.95cd	0	0	0	0	0
RN1350		0	6.69ad	0	0	0	0	0	0	0	0	3.51ad	0
RN1410	199	0	0	0	0	5.12ad	0	0	0	0	0	0	0
RN1460	2052	0	0	0	0	0	0	0	0	0	0	0	0
RN1480	435	0	0	0	0	0	0	0	0	0	0	0	0
RN1510	90308	0	0	0	0	0	0	0	0	0	0	0	0
RN1530	500	0	0	0	0	0	0	0	0	0	0	1.17ad	0
RN1590	884	0	0	0	0	0	0	0	0	0	0	0	0
RN1620	783	0	0	0	0	0	0	0	1.54cd	0	0	0	0
RN1760	912	0	0	0	0.99ad	0	0	0.95cd	0	0	0	1.17ad	0
RP0010	290	0	0	0	0	0	0	14.26cd	0	0	0	0	0
RP0040	1915	0	0	0	2.96ad	0	0	0	0	0	0	0	0
RP0050	70590	0	0	0	0.99ad	0	0	0		0	0	0	0

RBG010	8.36ad	0	0	2.96ad	10.23ad	0	0.95ad	1.54cd	0	2.34ad	0	
RCG010	0	0	4.01ad	0	0	0	0	0	0	0	0	
RCG020	0	0	0		10.23ad	0	1.9ad	1.54cd	11.92ad	2.98ad	3.51ad	4.26ad
RCG040	31.76ad			26.61ad	25.58ad	0		73.7cd	47.68cd	26.85ad	26.94ad	0
RCG050	1.67ad	6.69ad	0	3.94ad	0	0	0	0	0	0	0	8.53ad
RCG060	3.34ad	0	0	13.8ad	5.12ad	0	1.9cd		0	0	4.69ad	8.53ad
RCG070		20.07ad	0	4.93ad	0	23.88ad	1.9cd		0	2.98ad	1.17ad	0
RCG080	3.34ad	0	0	0	0	0	0.95ad	0	11.92ad	2.98ad	1.17ad	0
RCG090	0	0	0	0	0	0	0	0	0	0	1.17ad	0
RCG100	0	0	0	0	0	0	0	1.54cd	0	0	0	0
RCG140	0	0	0	0	0	0	0	1.54ad	0	0	0	0
RCG150	0	13.38cd	0	1.97ad	0	0	0	0	0	0	0	0
RCG160	10.03ad	6.69ad	4.01ad	2.96ad	15.35ad	0	3.8cd		0	2.34ad	4.26cd	
RDG010	20.06ad	26.76cd	4.01ad	20.7ad	5.12ad	0	0	15.35cd	0	2.98ad	5.86ad	0
RDG020	3.34ad	0		5.91ad	0	0	0	10.75cd	11.92ad	0	2.34ad	0
RDG030	0	0	0	0	0	0	0	0	0	0	0	0
RDG040	0	0	0	0	0	0	0	0	0	0	0	0
RFG010	0	0	0	0.99ad	0	0	0	0	0	0	1.17ad	4.26cd
RFG030	0	0	4.01ad	1.97ad	0	0	0.95cd	1.54cd	0	0	0	0
RFG040	1.67ad	0	0	0	0	0	0.95ad	0	0	0	1.17ad	0
RFG050	0		4.01ad	9.86ad	5.12ad	0	1.9cd	3.07cd	0	5.97ad		0
RFG060	3.34ad	0	0	0.99ad	0	0	0	1.54cd	0	0	0	0
RFG070	0	0	4.01ad	0.99ad	0	0	0.95ad	1.54ad	0	0	0	0
RFG080	1.67ad	0	0	2.96ad	10.23ad	0			11.92ad	2.98ad	0	0

RFG090	0	0	0	0	0	0	0.95cd	1.54cd	0	0	0	4.26ad
RFG110	3.34ad	0	0	3.94ad	0	0	0.95ad	0	0	0	0	0
RMG010	0	0	0	0	0	0	0	0	0	2.98ad	0	0
RNG010	0	0	0	4.93ad	0	0	0	0	0	0	0	0
RNG020	0	0	4.01ad	2.96ad	5.12ad	0	0	1.54cd	0	0	0	0
RNG030	0	0	0	0.99ad	0	0	0	0	0	0	0	0
RNG040	6.69ad	6.69ad		24.64ad		0			0	8.95ad	4.69ad	4.26ad
RNG050		0	4.01ad		5.12ad	0	2.85cd	3.07cd	0	0	3.51ad	0
RNG060	3.34ad	0	0	9.86ad	10.23ad	0	0	3.07cd	0	8.95ad	2.34ad	0
RNG070	1.67ad	0	8.02ad	2.96ad	5.12ad	0		1.54cd	11.92ad	0	4.69ad	0
RNG080	1.67ad	0	0		5.12ad	23.88ad	3.8cd	4.61cd	0	0	1.17ad	0
RNG090	8.36ad	13.38ad		8.87ad	5.12ad	0		4.61cd	0	8.95ad	12.88ad	0
RNG100	8.36ad	0	12.03ad	5.91ad	0	0	1.9ad		11.92ad	0	2.34ad	4.26ad
