

**Information content as a screening tool for rare disease candidates in an EHR
dataset representing 1.2M Singaporeans**

Table S1. Computed prevalence for Genetic conditions.

Orpha code	Disease name	Prevalence
ORPHA:870	Down syndrome	1-5 / 10 000
ORPHA:1900	Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency	1-9 / 1 000 000
ORPHA:841	Sebocystomatosis	1-5 / 10 000
ORPHA:848	Beta-thalassemia	1-9 / 100 000
ORPHA:98944	Coloboma of iris	1-9 / 1 000 000
ORPHA:130	Brugada syndrome	1-5 / 10 000
ORPHA:739	Prader-Willi syndrome	1-9 / 100 000
ORPHA:2978	Chronic intestinal pseudoobstruction	1-9 / 1 000 000
ORPHA:805	Tuberous sclerosis complex	1-9 / 100 000
ORPHA:96346	Anorectal malformation	1-9 / 100 000
ORPHA:91411	Congenital ptosis	1-9 / 100 000
ORPHA:881	Turner syndrome	1-9 / 100 000
ORPHA:1851	Multicystic dysplastic kidney	1-9 / 1 000 000
ORPHA:636	Neurofibromatosis type 1	1-9 / 100 000
ORPHA:648	Noonan syndrome	1-9 / 100 000
ORPHA:247582	Citrin deficiency	1-9 / 1 000 000
ORPHA:3426	Double outlet right ventricle	1-9 / 1 000 000
ORPHA:54595	Craniopharyngioma	1-9 / 100 000
ORPHA:247	Inherited arrhythmogenic cardiomyopathy	1-9 / 1 000 000
ORPHA:892	Von Hippel-Lindau disease	1-9 / 1 000 000
ORPHA:267	Calpain-3-related limb-girdle muscular dystrophy R1	1-9 / 1 000 000
ORPHA:558	Marfan syndrome	1-9 / 100 000
ORPHA:83463	Microtia	1-9 / 100 000
ORPHA:84	Fanconi anemia	1-9 / 1 000 000
ORPHA:44890	Gastrointestinal stromal tumor	1-9 / 100 000
ORPHA:666	Osteogenesis imperfecta	1-9 / 100 000
ORPHA:144	Lynch syndrome	1-9 / 1 000 000
ORPHA:3451	Infantile spasms syndrome	1-9 / 100 000
ORPHA:550	MELAS	1-9 / 1 000 000
ORPHA:733	Familial adenomatous polyposis	1-9 / 100 000
ORPHA:93616	Hemoglobin H disease	1-9 / 100 000
ORPHA:98974	Fuchs endothelial corneal dystrophy	1-9 / 100 000
ORPHA:98878	Hemophilia A	1-9 / 100 000
ORPHA:288	Hereditary elliptocytosis	1-9 / 1 000 000
ORPHA:15	Achondroplasia	1-9 / 100 000

ORPHA:674	Accessory pancreas	<1 / 1 000 000
ORPHA:305	Junctional epidermolysis bullosa	<1 / 1 000 000
ORPHA:79361	Inherited epidermolysis bullosa	1-9 / 1 000 000
ORPHA:268882	Arnold-Chiari malformation type I	1-9 / 1 000 000
ORPHA:379	Chronic granulomatous disease	1-9 / 1 000 000
ORPHA:567	22q11.2 deletion syndrome	1-9 / 100 000
ORPHA:2356	Arachnoid cyst	1-9 / 100 000
ORPHA:442	Congenital hypothyroidism	1-9 / 100 000
ORPHA:388	Hirschsprung disease	1-9 / 100 000
ORPHA:2131	Alternating hemiplegia of childhood	1-9 / 1 000 000
ORPHA:1766	Dysequilibrium syndrome	1-9 / 1 000 000
ORPHA:221083	Hemifacial spasm	1-9 / 100 000
ORPHA:91492	Early-onset non-syndromic cataract	1-9 / 1 000 000
ORPHA:64755	Becker nevus syndrome	1-9 / 1 000 000
ORPHA:79147	Familial reactive perforating collagenosis	1-9 / 1 000 000
ORPHA:233	Duane retraction syndrome	1-9 / 1 000 000
ORPHA:35981	Polymicrogyria	1-9 / 1 000 000
ORPHA:140471	Hereditary sensory and autonomic neuropathy	<1 / 1 000 000
ORPHA:36412	Hypocomplementemic urticarial vasculitis	1-9 / 1 000 000
ORPHA:94068	Spondyloepiphyseal dysplasia congenita	1-9 / 1 000 000
ORPHA:683	Progressive supranuclear palsy	1-9 / 1 000 000
ORPHA:227535	Hereditary breast cancer	1-9 / 1 000 000
ORPHA:730	Autosomal dominant polycystic kidney disease	1-9 / 100 000
ORPHA:904	Williams syndrome	1-9 / 100 000
ORPHA:2573	Moyamoya disease	1-9 / 100 000
ORPHA:652	Multiple endocrine neoplasia type 1	1-9 / 1 000 000
ORPHA:846	Alpha-thalassemia	1-9 / 100 000
ORPHA:3071	Costello syndrome	1-9 / 1 000 000
ORPHA:99169	Epiblepharon	1-9 / 100 000
ORPHA:140997	Orofaciodigital syndrome	1-9 / 1 000 000
ORPHA:138	CHARGE syndrome	1-9 / 1 000 000
ORPHA:85447	ATTRV30M amyloidosis	1-9 / 1 000 000
ORPHA:89936	X-linked hypophosphatemia	<1 / 1 000 000
ORPHA:2398	Multiple symmetric lipomatosis	<1 / 1 000 000
ORPHA:2185	Congenital hydrocephalus	1-9 / 100 000
ORPHA:68335	Rare chromosomal anomaly	1-9 / 1 000 000
ORPHA:68380	Mitochondrial disease	1-9 / 1 000 000
ORPHA:2133	Hemoglobin E disease	1-9 / 100 000
ORPHA:90695	Non-acquired panhypopituitarism	1-9 / 100 000
ORPHA:1114	Aplasia cutis congenita	1-9 / 100 000
ORPHA:35689	Primary lateral sclerosis	<1 / 1 000 000
ORPHA:26793	Very long chain acyl-CoA dehydrogenase deficiency	1-9 / 1 000 000

ORPHA:98261	Progressive myoclonic epilepsy	1-9 / 1 000 000
ORPHA:250	Frontonasal dysplasia	1-9 / 1 000 000
ORPHA:97360	Robinow syndrome	1-9 / 1 000 000
ORPHA:2162	Holoprosencephaly	1-9 / 1 000 000
ORPHA:216820	Osteogenesis imperfecta type 4	1-9 / 1 000 000
ORPHA:42	Medium chain acyl-CoA dehydrogenase deficiency	1-9 / 1 000 000
ORPHA:1203	Duodenal atresia	1-9 / 1 000 000
ORPHA:661	Congenital central hypoventilation syndrome	1-9 / 1 000 000
ORPHA:2322	Kabuki syndrome	1-9 / 1 000 000
ORPHA:718	Isolated Pierre Robin syndrome	1-9 / 100 000
ORPHA:418	Congenital adrenal hyperplasia	1-9 / 100 000
ORPHA:905	Wilson disease	1-9 / 1 000 000
ORPHA:2612	Linear nevus sebaceus syndrome	1-9 / 1 000 000
ORPHA:101063	Situs inversus totalis	1-9 / 1 000 000
ORPHA:214	Cystinuria	1-9 / 1 000 000
ORPHA:1738	Trisomy 4p	<1 / 1 000 000
ORPHA:448	Hemophilia	1-9 / 100 000
ORPHA:1201	Small bowel atresia	<1 / 1 000 000
ORPHA:2128	Isolated hemihyperplasia	1-9 / 1 000 000
ORPHA:87	Apert syndrome	1-9 / 1 000 000
ORPHA:250923	Isolated aniridia	1-9 / 1 000 000
ORPHA:2849	Perlman syndrome	1-9 / 100 000
ORPHA:180074	True unicornuate uterus	1-9 / 1 000 000
ORPHA:98249	Ehlers-Danlos syndrome	1-9 / 1 000 000
ORPHA:307	Juvenile myoclonic epilepsy	1-9 / 100 000
ORPHA:891	Familial exudative vitreoretinopathy	1-9 / 1 000 000
ORPHA:86911	Epilepsy with myoclonic absences	1-9 / 1 000 000
ORPHA:2911	Poland syndrome	1-9 / 1 000 000
ORPHA:46724	Cerebral arteriovenous malformation	1-9 / 100 000
ORPHA:231222	Beta-thalassemia intermedia	1-9 / 100 000
ORPHA:116	Beckwith-Wiedemann syndrome	1-9 / 1 000 000
ORPHA:295051	Lower limb hypertrophy	1-9 / 100 000
ORPHA:54370	Primary membranoproliferative glomerulonephritis	1-9 / 1 000 000
ORPHA:216812	Osteogenesis imperfecta type 3	1-9 / 1 000 000
ORPHA:6	3-methylcrotonyl-CoA carboxylase deficiency	1-9 / 1 000 000
ORPHA:822	Hereditary spherocytosis	1-9 / 100 000
ORPHA:849	Glanzmann thrombasthenia	1-9 / 1 000 000
ORPHA:64739	Ovarian hyperstimulation syndrome	1-9 / 1 000 000
ORPHA:774	Hereditary hemorrhagic telangiectasia	1-9 / 1 000 000
ORPHA:684	Paramyotonia congenita of Von Eulenburg	1-9 / 1 000 000
ORPHA:1199	Esophageal atresia	1-9 / 100 000
ORPHA:199	Cornelia de Lange syndrome	1-9 / 1 000 000

ORPHA:79276	Acute intermittent porphyria	1-9 / 1 000 000
ORPHA:217	Isolated Dandy-Walker malformation	1-9 / 1 000 000
ORPHA:54260	Left ventricular noncompaction	1-9 / 1 000 000
ORPHA:1942	Myoclonic-astatic epilepsy	1-9 / 1 000 000
ORPHA:224	Neonatal diabetes mellitus	<1 / 1 000 000
ORPHA:99885	Isolated permanent neonatal diabetes mellitus	<1 / 1 000 000
ORPHA:93964	Blepharospasm-oromandibular dystonia syndrome	1-9 / 1 000 000
ORPHA:637	Full NF2-related schwannomatosis	1-9 / 1 000 000
ORPHA:2841	Familial benign chronic pemphigus	<1 / 1 000 000
ORPHA:209	Cutis laxa	<1 / 1 000 000
ORPHA:1572	Common variable immunodeficiency	1-9 / 1 000 000
ORPHA:97120	Distal arthrogryposis	1-9 / 1 000 000
ORPHA:216796	Osteogenesis imperfecta type 1	1-9 / 1 000 000
ORPHA:2380	Legg-Calv©-Perthes disease	1-9 / 1 000 000
ORPHA:303	Dystrophic epidermolysis bullosa	<1 / 1 000 000
ORPHA:93110	Posterior urethral valve	1-9 / 100 000
ORPHA:31112	Dermatofibrosarcoma protuberans	1-9 / 1 000 000
ORPHA:823	Spina bifida and other spinal dysraphisms	1-9 / 100 000
ORPHA:192	Coffin-Lowry syndrome	1-9 / 1 000 000
ORPHA:249	Fibrous dysplasia of bone	1-9 / 1 000 000
ORPHA:3205	Sturge-Weber syndrome	1-9 / 1 000 000
ORPHA:98879	Hemophilia B	1-9 / 100 000
ORPHA:3243	Sweet syndrome	1-9 / 1 000 000
ORPHA:93338	Polysyndactyly	1-9 / 1 000 000
ORPHA:511	Maple syrup urine disease	1-9 / 1 000 000
ORPHA:324	Fabry disease	1-9 / 1 000 000
ORPHA:2177	Hydranencephaly	1-9 / 1 000 000
ORPHA:169802	Severe hemophilia A	1-9 / 1 000 000
ORPHA:2314	Autosomal dominant hyper-IgE syndrome due to STAT3 deficiency	<1 / 1 000 000
ORPHA:429	Hypochondroplasia	1-9 / 1 000 000
ORPHA:201	Cowden syndrome	1-9 / 1 000 000
ORPHA:251	Multiple epiphyseal dysplasia	<1 / 1 000 000
ORPHA:1945	Rolandic epilepsy	1-9 / 100 000
ORPHA:98896	Duchenne muscular dystrophy	1-9 / 100 000
ORPHA:275766	Idiopathic pulmonary arterial hypertension	1-9 / 1 000 000
ORPHA:358	Gitelman syndrome	1-9 / 1 000 000
ORPHA:1880	Ebstein malformation of the tricuspid valve	1-9 / 100 000
ORPHA:93969	Open spinal dysraphism with a myelomeningocele	1-9 / 1 000 000
ORPHA:141074	External auditory canal aplasia/hypoplasia	1-9 / 1 000 000
ORPHA:563609	Isolated anencephaly	1-9 / 1 000 000
ORPHA:1941	Juvenile absence epilepsy	1-9 / 1 000 000

ORPHA:98977	Juvenile glaucoma	1-9 / 1 000 000
ORPHA:569	Familial or sporadic hemiplegic migraine	1-9 / 1 000 000
ORPHA:273	Steinert myotonic dystrophy	1-9 / 1 000 000
ORPHA:2869	Peutz-Jeghers syndrome	1-9 / 1 000 000
ORPHA:163596	Hb Bart's hydrops fetalis	1-9 / 1 000 000
ORPHA:98895	Becker muscular dystrophy	1-9 / 1 000 000
ORPHA:91495	Persistent hyperplastic primary vitreous	1-9 / 1 000 000
ORPHA:355	Gaucher disease	1-9 / 1 000 000
ORPHA:295049	Upper limb hypertrophy	1-9 / 1 000 000
ORPHA:1570	Symbrachydactyly of hands and feet	1-9 / 1 000 000
ORPHA:70	Proximal spinal muscular atrophy	1-9 / 1 000 000
ORPHA:83418	Proximal spinal muscular atrophy type 2	1-9 / 1 000 000
ORPHA:280	Wolf-Hirschhorn syndrome	1-9 / 1 000 000
ORPHA:886	Usher syndrome	<1 / 1 000 000
ORPHA:411709	Renal agenesis	1-9 / 1 000 000
ORPHA:72	Angelman syndrome	1-9 / 1 000 000
ORPHA:281	Monosomy 5p	1-9 / 1 000 000
ORPHA:329	Congenital factor XI deficiency	1-9 / 1 000 000
ORPHA:53035	Caroli disease	<1 / 1 000 000
ORPHA:104010	Intestinal polyposis syndrome	<1 / 1 000 000
ORPHA:3109	Mayer-Rokitansky-Kv ^o ster-Hauser syndrome	1-9 / 1 000 000
ORPHA:65753	Charcot-Marie-Tooth disease type 1	1-9 / 1 000 000
ORPHA:753	46,XY difference of sex development due to 5-alpha-reductase 2 deficiency	<1 / 1 000 000
ORPHA:314	Erythroderma desquamativum	<1 / 1 000 000
ORPHA:48471	Lissencephaly	1-9 / 1 000 000
ORPHA:98809	Paroxysmal kinesigenic dyskinesia	1-9 / 1 000 000
ORPHA:93277	Monostotic fibrous dysplasia	1-9 / 1 000 000
ORPHA:2930	Cronkhite-Canada syndrome	1-9 / 1 000 000
ORPHA:524	Li-Fraumeni syndrome	1-9 / 1 000 000
ORPHA:2614	Nail-patella syndrome	1-9 / 1 000 000
ORPHA:217656	Inherited isolated arrhythmogenic cardiomyopathy	<1 / 1 000 000
ORPHA:792	X-linked retinoschisis	1-9 / 1 000 000
ORPHA:2382	Lennox-Gastaut syndrome	1-9 / 1 000 000
ORPHA:47	X-linked agammaglobulinemia	1-9 / 1 000 000
ORPHA:1517	CantVf syndrome	<1 / 1 000 000
ORPHA:35737	Morning glory disc anomaly	1-9 / 1 000 000
ORPHA:3320	Thrombocytopenia-absent radius syndrome	<1 / 1 000 000
ORPHA:464	Incontinentia pigmenti	1-9 / 100 000
ORPHA:93928	Isolated epispadias	<1 / 1 000 000
ORPHA:908	Fragile X syndrome	1-9 / 1 000 000
ORPHA:443079	Central serous chorioretinopathy	1-9 / 1 000 000

ORPHA:710	Pfeiffer syndrome	1-9 / 1 000 000
ORPHA:231249	Hemoglobin E-beta-thalassemia syndrome	1-9 / 100 000
ORPHA:93929	Cloacal exstrophy	1-9 / 1 000 000
ORPHA:216804	Osteogenesis imperfecta type 2	<1 / 1 000 000
ORPHA:664	Ornithine transcarbamylase deficiency	1-9 / 1 000 000
ORPHA:1531	Craniosynostosis	1-9 / 100 000
ORPHA:552	MODY	1-9 / 1 000 000
ORPHA:274	Bernard-Soulier syndrome	1-9 / 1 000 000
ORPHA:1059	Blue rubber bleb nevus	1-9 / 1 000 000
ORPHA:77240	Primary lymphedema	1-9 / 1 000 000
ORPHA:783	Rubinstein-Taybi syndrome	1-9 / 1 000 000
ORPHA:506	Leigh syndrome	1-9 / 1 000 000
ORPHA:79201	Glycogen storage disease	1-9 / 1 000 000
ORPHA:100	Ataxia-telangiectasia	<1 / 1 000 000
ORPHA:813	Silver-Russell syndrome	1-9 / 1 000 000
ORPHA:568	Microphthalmia, Lenz type	<1 / 1 000 000
ORPHA:244	Primary ciliary dyskinesia	<1 / 1 000 000
ORPHA:1309	Medullary sponge kidney	1-9 / 1 000 000
ORPHA:1340	Cardiofaciocutaneous syndrome	1-9 / 1 000 000
ORPHA:2451	Mucocutaneous venous malformations	<1 / 1 000 000
ORPHA:1772	45,X/46,XY mixed gonadal dysgenesis	1-9 / 1 000 000
ORPHA:2308	Jacobsen syndrome	<1 / 1 000 000
ORPHA:55	Oculocutaneous albinism	1-9 / 1 000 000
ORPHA:79434	Oculocutaneous albinism type 1B	<1 / 1 000 000
ORPHA:778	Rett syndrome	1-9 / 1 000 000
ORPHA:58	Alexander disease	1-9 / 1 000 000
ORPHA:195	Cat-eye syndrome	<1 / 1 000 000
ORPHA:93403	Syndactyly type 2	1-9 / 1 000 000
ORPHA:685	Hereditary spastic paraplegia	1-9 / 1 000 000
ORPHA:79373	Ectodermal dysplasia syndrome	1-9 / 1 000 000
ORPHA:2929	Juvenile polyposis syndrome	1-9 / 1 000 000
ORPHA:99429	Complete androgen insensitivity syndrome	<1 / 1 000 000
ORPHA:158	Systemic primary carnitine deficiency	1-9 / 1 000 000
ORPHA:791	Retinitis pigmentosa	1-9 / 1 000 000
ORPHA:95719	Thyroid hemiagenesis	<1 / 1 000 000
ORPHA:141127	Congenital tracheal stenosis	1-9 / 1 000 000
ORPHA:60030	Loeys-Dietz syndrome	1-9 / 1 000 000
ORPHA:2440	Isolated split hand-split foot malformation	1-9 / 1 000 000
ORPHA:906	Wiskott-Aldrich syndrome	1-9 / 1 000 000
ORPHA:480	Kearns-Sayre syndrome	<1 / 1 000 000
ORPHA:2686	Cyclic neutropenia	1-9 / 1 000 000
ORPHA:675	Annular pancreas	<1 / 1 000 000

ORPHA:43	X-linked adrenoleukodystrophy	1-9 / 1 000 000
ORPHA:178382	Congenital vertical talus	1-9 / 1 000 000
ORPHA:399	Huntington disease	1-9 / 1 000 000
ORPHA:35	Propionic acidemia	1-9 / 1 000 000
ORPHA:98973	Posterior polymorphous corneal dystrophy	<1 / 1 000 000
ORPHA:232	Sickle cell anemia	1-9 / 1 000 000
ORPHA:33069	Dravet syndrome	1-9 / 1 000 000
ORPHA:52	Alagille syndrome	1-9 / 1 000 000
ORPHA:79153	Idiopathic trachyonychia	<1 / 1 000 000
ORPHA:64734	Iridocorneal endothelial syndrome	1-9 / 1 000 000
ORPHA:2897	Pityriasis rubra pilaris	1-9 / 1 000 000
ORPHA:18	Distal renal tubular acidosis	1-9 / 1 000 000
ORPHA:3269	Congenital radioulnar synostosis	1-9 / 1 000 000
ORPHA:3375	Trisomy X	1-9 / 1 000 000
ORPHA:207	Crouzon syndrome	1-9 / 1 000 000
ORPHA:743	Severe hereditary thrombophilia due to congenital protein S deficiency	1-9 / 1 000 000
ORPHA:2591	Infantile myofibromatosis	<1 / 1 000 000
ORPHA:3193	Supravalvular aortic stenosis	1-9 / 1 000 000
ORPHA:65	Leber congenital amaurosis	<1 / 1 000 000
ORPHA:531	Miller-Dieker syndrome	<1 / 1 000 000
ORPHA:1987	Femoral agenesis/hypoplasia	1-9 / 1 000 000
ORPHA:415	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	<1 / 1 000 000
ORPHA:3157	Septo-optic dysplasia spectrum	1-9 / 1 000 000
ORPHA:3027	Caudal regression syndrome	1-9 / 1 000 000
ORPHA:98757	Spinocerebellar ataxia type 3	1-9 / 1 000 000
ORPHA:657	Congenital isolated hyperinsulinism	1-9 / 1 000 000
ORPHA:738	Porphyria	<1 / 1 000 000
ORPHA:79318	PMM2-CDG	1-9 / 1 000 000
ORPHA:295229	Congenital genu recurvatum	1-9 / 1 000 000
ORPHA:68356	Leukodystrophy	1-9 / 1 000 000
ORPHA:276161	Multiple endocrine neoplasia	1-9 / 1 000 000
ORPHA:163634	Maffucci syndrome	<1 / 1 000 000
ORPHA:296	Ollier disease	<1 / 1 000 000
ORPHA:251375	Sickle cell-hemoglobin E disease syndrome	1-9 / 1 000 000
ORPHA:725	Continuous spikes and waves during sleep	<1 / 1 000 000
ORPHA:110	Bardet-Biedl syndrome	1-9 / 1 000 000
ORPHA:79273	Hereditary coproporphyria	<1 / 1 000 000
ORPHA:308993	Glycerol kinase deficiency	<1 / 1 000 000
ORPHA:71277	Classic glucose transporter type 1 deficiency syndrome	<1 / 1 000 000
ORPHA:141276	Tessier number 7 facial cleft	<1 / 1 000 000

ORPHA:98946	Coloboma of eyelid	1-9 / 1 000 000
ORPHA:294988	Congenital hypoplasia of thumb	1-9 / 1 000 000
ORPHA:2828	Young-onset Parkinson disease	<1 / 1 000 000
ORPHA:64280	Childhood absence epilepsy	1-9 / 1 000 000
ORPHA:731	Autosomal recessive polycystic kidney disease	<1 / 1 000 000
ORPHA:869	Triple A syndrome	<1 / 1 000 000
ORPHA:819	Smith-Magenis syndrome	<1 / 1 000 000
ORPHA:365	Glycogen storage disease due to acid maltase deficiency	1-9 / 1 000 000
ORPHA:166282	Familial sick sinus syndrome	<1 / 1 000 000
ORPHA:99898	Mendelian susceptibility to mycobacterial diseases due to complete IFN γ R1 deficiency	<1 / 1 000 000
ORPHA:583	Mucopolysaccharidosis type 6	1-9 / 1 000 000
ORPHA:79213	Mucopolysaccharidosis	1-9 / 1 000 000
ORPHA:1931	Frontal encephalocele	<1 / 1 000 000
ORPHA:98995	Early-onset zonular cataract	1-9 / 1 000 000
ORPHA:3380	Trisomy 18	1-9 / 1 000 000
ORPHA:124	Diamond-Blackfan anemia	1-9 / 1 000 000
ORPHA:828	Stickler syndrome	<1 / 1 000 000
ORPHA:14	Abetalipoproteinemia	<1 / 1 000 000
ORPHA:100050	Hereditary angioedema type 1	<1 / 1 000 000
ORPHA:91378	Hereditary angioedema	1-9 / 1 000 000
ORPHA:261	Emery-Dreifuss muscular dystrophy	<1 / 1 000 000
ORPHA:231214	Beta-thalassemia major	1-9 / 1 000 000
ORPHA:79143	Isolated congenital anonychia	<1 / 1 000 000
ORPHA:1885	Isolated ectopia lentis	1-9 / 1 000 000
ORPHA:79254	Classic phenylketonuria	1-9 / 1 000 000
ORPHA:1198	Colonic atresia	<1 / 1 000 000
ORPHA:1928	Congenital lobar emphysema	<1 / 1 000 000
ORPHA:447	Paroxysmal nocturnal hemoglobinuria	1-9 / 1 000 000
ORPHA:8	47,XYY syndrome	1-9 / 1 000 000
ORPHA:169349	Immuno-osseous dysplasia	<1 / 1 000 000
ORPHA:36387	Generalized epilepsy with febrile seizures-plus	<1 / 1 000 000
ORPHA:457	Harlequin ichthyosis	<1 / 1 000 000
ORPHA:93930	Bladder exstrophy	1-9 / 1 000 000
ORPHA:83330	Proximal spinal muscular atrophy type 1	1-9 / 1 000 000
ORPHA:190	Coats disease	1-9 / 1 000 000
ORPHA:51	Aicardi-Goutieres syndrome	<1 / 1 000 000
ORPHA:681	Hypokalemic periodic paralysis	1-9 / 1 000 000
ORPHA:1335	Pentalogy of Cantrell	1-9 / 1 000 000
ORPHA:23	Argininosuccinic aciduria	<1 / 1 000 000
ORPHA:821	Sotos syndrome	1-9 / 1 000 000
ORPHA:83419	Proximal spinal muscular atrophy type 3	<1 / 1 000 000

ORPHA:708	Peters anomaly	1-9 / 1 000 000
ORPHA:3440	Waardenburg syndrome	<1 / 1 000 000
ORPHA:93256	Fragile X-associated tremor/ataxia syndrome	<1 / 1 000 000
ORPHA:223	Nephrogenic diabetes insipidus	1-9 / 1 000 000
ORPHA:218	Darier disease	1-9 / 1 000 000
ORPHA:295032	Isolated congenital radial head dislocation	<1 / 1 000 000
ORPHA:184	Cherubism	<1 / 1 000 000
ORPHA:337	Fibrodysplasia ossificans progressiva	1-9 / 1 000 000
ORPHA:33	Isovaleric acidemia	<1 / 1 000 000
ORPHA:1452	Cleidocranial dysplasia	<1 / 1 000 000
ORPHA:2345	Isolated Klippel-Feil syndrome	1-9 / 1 000 000
ORPHA:380	Greig cephalopolysyndactyly syndrome	<1 / 1 000 000
ORPHA:93101	Renal hypoplasia	<1 / 1 000 000
ORPHA:702	Pelizaeus-Merzbacher disease	<1 / 1 000 000
ORPHA:435372	Anterior urethral valve	<1 / 1 000 000
ORPHA:215	Congenital stationary night blindness	<1 / 1 000 000
ORPHA:60041	Congenital heart block	1-9 / 1 000 000
ORPHA:64754	Nevus comedonicus syndrome	<1 / 1 000 000
ORPHA:3447	Weaver syndrome	<1 / 1 000 000
ORPHA:93976	Anotia	1-9 / 1 000 000
ORPHA:135	CACH syndrome	1-9 / 1 000 000
ORPHA:607	Nemaline myopathy	1-9 / 1 000 000
ORPHA:79241	Biotinidase deficiency	<1 / 1 000 000
ORPHA:503	Larsen syndrome	<1 / 1 000 000
ORPHA:758	Pseudoxanthoma elasticum	<1 / 1 000 000
ORPHA:166078	Von Willebrand disease type 1	1-9 / 1 000 000
ORPHA:884	Tetrasomy 12p	1-9 / 1 000 000
ORPHA:77261	Gaucher disease type 3	1-9 / 1 000 000
ORPHA:169808	Mild hemophilia A	<1 / 1 000 000
ORPHA:653	Multiple endocrine neoplasia type 2	<1 / 1 000 000
ORPHA:206443	Late-infantile/juvenile Krabbe disease	<1 / 1 000 000
ORPHA:268823	Occipital encephalocele	<1 / 1 000 000
ORPHA:2257	Primary pulmonary hypoplasia	<1 / 1 000 000
ORPHA:79152	Disseminated superficial actinic porokeratosis	<1 / 1 000 000
ORPHA:221061	Familial cerebral cavernous malformation	<1 / 1 000 000
ORPHA:799	Schizencephaly	1-9 / 1 000 000
ORPHA:52530	Pseudo-von Willebrand disease	<1 / 1 000 000
ORPHA:126	Blepharophimosis-ptosis-epicanthus inversus syndrome	<1 / 1 000 000
ORPHA:586	Cystic fibrosis	1-9 / 1 000 000
ORPHA:2940	Porencephaly	1-9 / 1 000 000
ORPHA:364	Glycogen storage disease due to glucose-6-phosphatase deficiency	1-9 / 1 000 000

ORPHA:275777	Heritable pulmonary arterial hypertension	<1 / 1 000 000
ORPHA:95429	Angioma serpiginosum	1-9 / 1 000 000
ORPHA:405	Familial hypocalciuric hypercalcemia	<1 / 1 000 000
ORPHA:91412	Marcus-Gunn syndrome	1-9 / 1 000 000
ORPHA:565	Menkes disease	1-9 / 1 000 000
ORPHA:3411	Double uterus-hemivagina-renal agenesis syndrome	<1 / 1 000 000
ORPHA:98956	Epithelial basement membrane dystrophy	<1 / 1 000 000
ORPHA:3463	Wolfram syndrome	<1 / 1 000 000
ORPHA:295000	Constriction rings syndrome	1-9 / 1 000 000
ORPHA:838	Susac syndrome	<1 / 1 000 000
ORPHA:100070	Progressive non-fluent aphasia	<1 / 1 000 000
ORPHA:109	Bannayan-Riley-Ruvalcaba syndrome	<1 / 1 000 000
ORPHA:2289	Neuronal intranuclear inclusion disease	<1 / 1 000 000
ORPHA:268813	Myelocystocele	<1 / 1 000 000
ORPHA:466	Fatal familial insomnia	<1 / 1 000 000
ORPHA:352540	Oncogenic osteomalacia	<1 / 1 000 000
ORPHA:309297	Mucopolysaccharidosis type 4A	<1 / 1 000 000
ORPHA:231237	Delta-beta-thalassemia	<1 / 1 000 000
ORPHA:26791	Multiple acyl-CoA dehydrogenase deficiency	<1 / 1 000 000
ORPHA:268817	Cephalocele	1-9 / 1 000 000
ORPHA:29072	Hereditary pheochromocytoma-paraganglioma	<1 / 1 000 000
ORPHA:2052	Fraser syndrome	1-9 / 1 000 000
ORPHA:85173	IMAGe syndrome	<1 / 1 000 000
ORPHA:54247	Posterior cortical atrophy	<1 / 1 000 000
ORPHA:285	Hypermobile Ehlers-Danlos syndrome	1-9 / 1 000 000
ORPHA:37553	Andersen-Tawil syndrome	<1 / 1 000 000
ORPHA:35099	Non-syndromic bicoronal craniosynostosis	<1 / 1 000 000
ORPHA:163927	Pustulosis palmaris et plantaris	<1 / 1 000 000
ORPHA:478	Kallmann syndrome	<1 / 1 000 000
ORPHA:2315	Johanson-Blizzard syndrome	<1 / 1 000 000
ORPHA:3111	Rotor syndrome	<1 / 1 000 000
ORPHA:582	Mucopolysaccharidosis type 4	1-9 / 1 000 000
ORPHA:634	Netherton syndrome	<1 / 1 000 000
ORPHA:382	Guanidinoacetate methyltransferase deficiency	<1 / 1 000 000
ORPHA:1071	Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome	<1 / 1 000 000
ORPHA:1934	Early infantile epileptic encephalopathy	1-9 / 1 000 000
ORPHA:1935	Early myoclonic encephalopathy	1-9 / 1 000 000
ORPHA:275864	Behavioral variant of frontotemporal dementia	<1 / 1 000 000
ORPHA:1606	1p36 deletion syndrome	<1 / 1 000 000
ORPHA:83454	Glomuvenous malformation	<1 / 1 000 000
ORPHA:377	Gorlin syndrome	<1 / 1 000 000
ORPHA:282	Frontotemporal dementia	1-9 / 1 000 000

ORPHA:366	Glycogen storage disease due to glycogen debranching enzyme deficiency	1-9 / 1 000 000
ORPHA:73	Gorham-Stout disease	<1 / 1 000 000
ORPHA:289	Ellis Van Creveld syndrome	<1 / 1 000 000
ORPHA:91490	Isolated congenital sclerocornea	1-9 / 1 000 000
ORPHA:2554	Ear-patella-short stature syndrome	<1 / 1 000 000
ORPHA:983	Testicular regression syndrome	<1 / 1 000 000
ORPHA:974	Adams-Oliver syndrome	<1 / 1 000 000
ORPHA:857	Townes-Brocks syndrome	<1 / 1 000 000
ORPHA:84064	Syndromic diarrhea	<1 / 1 000 000
ORPHA:2875	Phakomatosis pigmentovascularis	<1 / 1 000 000
ORPHA:481	Kennedy disease	<1 / 1 000 000
ORPHA:2134	Atypical hemolytic uremic syndrome	<1 / 1 000 000
ORPHA:2346	Angioosteohypertrophic syndrome	<1 / 1 000 000
ORPHA:672	Pallister-Hall syndrome	<1 / 1 000 000
ORPHA:861	Treacher-Collins syndrome	1-9 / 1 000 000
ORPHA:1393	Cerebrocostomandibular syndrome	<1 / 1 000 000
ORPHA:446	Neonatal hemochromatosis	<1 / 1 000 000
ORPHA:115	Congenital contractural arachnodactyly	<1 / 1 000 000
ORPHA:79145	Dowling-Degos disease	<1 / 1 000 000
ORPHA:97593	Pseudohypoparathyroidism	<1 / 1 000 000
ORPHA:3342	Arterial tortuosity syndrome	<1 / 1 000 000
ORPHA:79234	Crigler-Najjar syndrome type 1	<1 / 1 000 000
ORPHA:45358	Congenital fibrosis of extraocular muscles	<1 / 1 000 000
ORPHA:454750	Isolated tracheoesophageal fistula	<1 / 1 000 000
ORPHA:37	Acrodermatitis enteropathica	<1 / 1 000 000
ORPHA:79665	Gardner syndrome	<1 / 1 000 000
ORPHA:107	BOR syndrome	<1 / 1 000 000
ORPHA:475	Joubert syndrome	<1 / 1 000 000
ORPHA:79204	Lipid storage disease	<1 / 1 000 000
ORPHA:2152	Mowat-Wilson syndrome	<1 / 1 000 000
ORPHA:98818	Landau-Kleffner syndrome	<1 / 1 000 000

Table S2. Computed prevalence for Neoplastic conditions.

Orpha code	Disease name	Prevalence
ORPHA:3394	Soft tissue sarcoma	1-9 / 100 000
ORPHA:213772	Adenocarcinoma of the cervix uteri	1-9 / 100 000
ORPHA:729	Polycythemia vera	1-5 / 10 000
ORPHA:98274	Myeloproliferative neoplasm	1-9 / 100 000
ORPHA:3318	Essential thrombocythemia	1-5 / 10 000
ORPHA:223735	Lymphoma	1-5 / 10 000
ORPHA:52416	Mantle cell lymphoma	1-9 / 100 000

ORPHA:877	Neuroendocrine neoplasm	1-5 / 10 000
ORPHA:213767	Squamous cell carcinoma of the cervix uteri	1-5 / 10 000
ORPHA:519	Acute myeloid leukemia	1-5 / 10 000
ORPHA:29073	Multiple myeloma	1-5 / 10 000
ORPHA:88673	Hepatocellular carcinoma	6-9 / 10 000
ORPHA:424016	Adenocarcinoma of the anal canal	<1 / 1 000 000
ORPHA:252175	Vestibular schwannoma	1-9 / 100 000
ORPHA:521	Chronic myeloid leukemia	1-5 / 10 000
ORPHA:26790	Pseudomyxoma peritonei	1-9 / 1 000 000
ORPHA:52688	Myelodysplastic syndrome	1-5 / 10 000
ORPHA:100088	Rare thyroid carcinoma	1-9 / 1 000 000
ORPHA:824	Primary myelofibrosis	1-9 / 100 000
ORPHA:168829	Primary peritoneal carcinoma	1-9 / 100 000
ORPHA:64720	Leiomyosarcoma	1-9 / 100 000
ORPHA:141112	Nasal glial heterotopia	<1 / 1 000 000
ORPHA:99408	Pituitary adenoma	1-9 / 100 000
ORPHA:178512	Folliculotropic mycosis fungoides	1-9 / 100 000
ORPHA:137583	Vulvar intraepithelial neoplasia	1-9 / 1 000 000
ORPHA:2965	Prolactinoma	1-9 / 100 000
ORPHA:529852	Combined hepatocellular carcinoma and cholangiocarcinoma	1-9 / 1 000 000
ORPHA:66646	Cutaneous mastocytosis	<1 / 1 000 000
ORPHA:98841	Anaplastic large cell lymphoma	1-9 / 100 000
ORPHA:963	Acromegaly	1-9 / 100 000
ORPHA:79467	Verrucous nevus	1-9 / 1 000 000
ORPHA:99861	Precursor T-cell acute lymphoblastic leukemia	1-9 / 100 000
ORPHA:873	Desmoid tumor	1-9 / 100 000
ORPHA:254685	Gestational trophoblastic disease	1-9 / 1 000 000
ORPHA:99927	Hydatidiform mole	1-9 / 1 000 000
ORPHA:91414	Pilomatrixoma	1-9 / 100 000
ORPHA:520	Acute promyelocytic leukemia	1-9 / 100 000
ORPHA:98845	Classic Hodgkin lymphoma, lymphocyte-rich type	1-9 / 1 000 000
ORPHA:98832	Acute myeloid leukemia with minimal differentiation	<1 / 1 000 000
ORPHA:547	Non-Hodgkin lymphoma	1-9 / 1 000 000
ORPHA:180242	Malignant tumor of fallopian tubes	1-9 / 100 000
ORPHA:389	Langerhans cell histiocytosis	1-9 / 100 000
ORPHA:518	Acute megakaryoblastic leukemia	1-9 / 1 000 000
ORPHA:178342	Inflammatory myofibroblastic tumor	1-9 / 1 000 000
ORPHA:3260	Idiopathic hypereosinophilic syndrome	1-9 / 1 000 000
ORPHA:86851	Acute leukemia of ambiguous lineage	1-9 / 1 000 000
ORPHA:33226	Waldenström macroglobulinemia	1-9 / 100 000
ORPHA:100021	Primary plasmacytoma of the bone	1-9 / 100 000
ORPHA:85443	AL amyloidosis	1-9 / 100 000

ORPHA:98838	Primary mediastinal large B-cell lymphoma	1-9 / 1 000 000
ORPHA:254688	Complete hydatidiform mole	1-9 / 100 000
ORPHA:263425	Nevus of Ota	1-9 / 100 000
ORPHA:530995	Mixed phenotype acute leukemia	1-9 / 1 000 000
ORPHA:70568	Post-transplant lymphoproliferative disease	1-9 / 100 000
ORPHA:97279	Insulinoma	1-9 / 1 000 000
ORPHA:86854	Splenic marginal zone lymphoma	1-9 / 100 000
ORPHA:75564	Acquired idiopathic sideroblastic anemia	1-9 / 1 000 000
ORPHA:874	Primary adult heart tumor	1-9 / 1 000 000
ORPHA:263413	Angiosarcoma	1-9 / 100 000
ORPHA:69078	Liposarcoma	1-9 / 1 000 000
ORPHA:86886	Angioimmunoblastic T-cell lymphoma	1-9 / 100 000
ORPHA:252183	Neurofibroma	1-9 / 100 000
ORPHA:86875	Adult T-cell leukemia/lymphoma	1-9 / 100 000
ORPHA:33276	Kaposi sarcoma	1-9 / 1 000 000
ORPHA:780	Rhabdomyosarcoma	1-9 / 100 000
ORPHA:876	Yolk sac tumor	1-9 / 1 000 000
ORPHA:543	Burkitt lymphoma	1-9 / 100 000
ORPHA:251912	Pineocytoma	1-9 / 1 000 000
ORPHA:99860	Precursor B-cell acute lymphoblastic leukemia	1-9 / 100 000
ORPHA:2905	POEMS syndrome	1-9 / 1 000 000
ORPHA:58017	Classic hairy cell leukemia	1-9 / 1 000 000
ORPHA:364043	ALK-positive large B-cell lymphoma	1-9 / 1 000 000
ORPHA:83468	Solitary bone cyst	<1 / 1 000 000
ORPHA:391	Classic Hodgkin lymphoma	1-9 / 1 000 000
ORPHA:98844	Classic Hodgkin lymphoma, mixed cellularity type	1-9 / 1 000 000
ORPHA:300903	ALK-negative anaplastic large cell lymphoma	1-9 / 1 000 000
ORPHA:96253	Cushing disease	1-9 / 100 000
ORPHA:289666	Plasmablastic lymphoma	1-9 / 1 000 000
ORPHA:301	Ependymal tumor	1-9 / 1 000 000
ORPHA:363976	Giant cell tumor of bone	1-9 / 1 000 000
ORPHA:99756	Alveolar rhabdomyosarcoma	1-9 / 1 000 000
ORPHA:99925	Invasive mole	1-9 / 1 000 000
ORPHA:447777	Keratocystic odontogenic tumor	<1 / 1 000 000
ORPHA:223727	Bone sarcoma	1-9 / 100 000
ORPHA:2467	Systemic mastocytosis	1-9 / 1 000 000
ORPHA:494418	Vulvar carcinoma	1-9 / 1 000 000
ORPHA:98275	Myelodysplastic/myeloproliferative disease	1-9 / 1 000 000
ORPHA:86870	Blastic plasmacytoid dendritic cell neoplasm	1-9 / 1 000 000
ORPHA:454714	Plasma cell leukemia	1-9 / 1 000 000
ORPHA:199251	Ledderhose disease	1-9 / 1 000 000
ORPHA:86869	Lymphomatoid granulomatosis	1-9 / 1 000 000

ORPHA:97338	Melanoma of soft tissue	<1 / 1 000 000
ORPHA:86879	Extranodal nasal NK/T cell lymphoma	1-9 / 1 000 000
ORPHA:99867	Thymoma	1-9 / 1 000 000
ORPHA:86829	Chronic neutrophilic leukemia	1-9 / 1 000 000
ORPHA:160	Castleman disease	1-9 / 1 000 000
ORPHA:279904	Primary intraocular lymphoma	1-9 / 1 000 000
ORPHA:2126	Solitary fibrous tumor	1-9 / 1 000 000
ORPHA:99977	Squamous cell carcinoma of the esophagus	<1 / 1 000 000
ORPHA:98823	Chronic myelomonocytic leukemia	1-9 / 1 000 000
ORPHA:254693	Partial hydatidiform mole	1-9 / 1 000 000
ORPHA:480553	Aneurysmal bone cyst	1-9 / 1 000 000
ORPHA:52417	MALT lymphoma	1-9 / 1 000 000
ORPHA:180261	Phyllodes tumor of the breast	1-9 / 1 000 000
ORPHA:252054	Hemangioblastoma	1-9 / 1 000 000
ORPHA:247762	Lipoblastoma	1-9 / 1 000 000
ORPHA:86855	Plasmacytoma	<1 / 1 000 000
ORPHA:477742	Nodular fasciitis	<1 / 1 000 000
ORPHA:79140	Cutaneous neuroendocrine carcinoma	<1 / 1 000 000
ORPHA:391651	Glomus tumor	1-9 / 1 000 000
ORPHA:98825	Unclassified myelodysplastic/myeloproliferative disease	1-9 / 1 000 000
ORPHA:86834	Juvenile myelomonocytic leukemia	1-9 / 1 000 000
ORPHA:1332	Medullary thyroid carcinoma	1-9 / 1 000 000
ORPHA:2086	Optic pathway glioma	1-9 / 1 000 000
ORPHA:100075	Neuroendocrine tumor of stomach	<1 / 1 000 000
ORPHA:2122	Kaposiform hemangioendothelioma	1-9 / 1 000 000
ORPHA:99889	Cushing syndrome due to ectopic ACTH secretion	<1 / 1 000 000
ORPHA:178533	Primary cutaneous gamma/delta-positive T-cell lymphoma	<1 / 1 000 000
ORPHA:99967	Myxoid/round cell liposarcoma	1-9 / 1 000 000
ORPHA:90389	Telangiectasia macularis eruptiva perstans	<1 / 1 000 000
ORPHA:86867	Nodal marginal zone B-cell lymphoma	<1 / 1 000 000
ORPHA:544	Diffuse large B-cell lymphoma	<1 / 1 000 000
ORPHA:100020	Refractory anemia with excess blasts type 2	<1 / 1 000 000
ORPHA:99869	Thymic neuroendocrine carcinoma	<1 / 1 000 000
ORPHA:86884	Subcutaneous panniculitis-like T-cell lymphoma	1-9 / 1 000 000
ORPHA:98824	Atypical chronic myeloid leukemia	<1 / 1 000 000
ORPHA:86850	Myeloid sarcoma	1-9 / 1 000 000
ORPHA:420611	Transient myeloproliferative syndrome	<1 / 1 000 000
ORPHA:98282	Plasma cell tumor	<1 / 1 000 000
ORPHA:168966	Composite lymphoma	<1 / 1 000 000
ORPHA:569164	Angiomatoid fibrous histiocytoma	<1 / 1 000 000
ORPHA:289596	Juvenile nasopharyngeal angiofibroma	<1 / 1 000 000
ORPHA:314473	Ovarian fibroma	<1 / 1 000 000

ORPHA:398058	Squamous cell carcinoma of the penis	<1 / 1 000 000
ORPHA:86852	B-cell prolymphocytic leukemia	<1 / 1 000 000
ORPHA:93292	Adenoma of pancreas	<1 / 1 000 000

Table S3. Computed prevalence for Other RDs.

Orpha code	Disease name	Prevalence
ORPHA:732	Polymyositis	1-9 / 100 000
ORPHA:90291	Systemic sclerosis	1-9 / 100 000
ORPHA:536	Systemic lupus erythematosus	1-5 / 10 000
ORPHA:91355	Sheehan syndrome	1-9 / 1 000 000
ORPHA:70589	Bronchopulmonary dysplasia	1-9 / 100 000
ORPHA:700	Alopecia totalis	1-9 / 1 000 000
ORPHA:761	Immunoglobulin A vasculitis	1-5 / 10 000
ORPHA:703	Bullous pemphigoid	1-5 / 10 000
ORPHA:199302	Isolated cleft lip	1-9 / 100 000
ORPHA:70475	Radiation proctitis	1-9 / 100 000
ORPHA:221091	Trigeminal neuralgia	1-5 / 10 000
ORPHA:36426	Stevens-Johnson syndrome	1-9 / 100 000
ORPHA:570762	Infective endocarditis	1-5 / 10 000
ORPHA:50918	Kikuchi-Fujimoto disease	1-9 / 100 000
ORPHA:280886	Anterior uveitis	1-9 / 100 000
ORPHA:1656	Dermatitis herpetiformis	<1 / 1 000 000
ORPHA:99054	Valvular pulmonary stenosis	1-5 / 10 000
ORPHA:94059	Uremic pruritus	1-9 / 1 000 000
ORPHA:90050	Retinopathy of prematurity	1-5 / 10 000
ORPHA:2032	Idiopathic pulmonary fibrosis	1-9 / 100 000
ORPHA:275555	Preeclampsia	1-5 / 10 000
ORPHA:589	Myasthenia gravis	1-5 / 10 000
ORPHA:1302	Cryptogenic organizing pneumonia	1-9 / 100 000
ORPHA:186	Primary biliary cholangitis	1-9 / 100 000
ORPHA:1163	Aspergillosis	1-9 / 1 000 000
ORPHA:139417	Acute transverse myelitis	1-9 / 1 000 000
ORPHA:102	Multiple system atrophy	1-9 / 100 000
ORPHA:98919	Miller Fisher syndrome	1-9 / 100 000
ORPHA:3280	Syringomyelia	1-9 / 1 000 000
ORPHA:178029	Central diabetes insipidus	1-9 / 100 000
ORPHA:2331	Kawasaki disease	1-5 / 10 000
ORPHA:480501	Choledochal cyst	1-9 / 100 000
ORPHA:37202	Interstitial cystitis	1-9 / 100 000
ORPHA:217080	Pulmonary fungal infections in patients deemed at risk	1-9 / 1 000 000
ORPHA:411703	Pulmonary non-tuberculous mycobacterial infection	1-5 / 10 000
ORPHA:3161	Congenital pulmonary sequestration	1-9 / 1 000 000

ORPHA:228113	Anal fistula	1-5 / 10 000
ORPHA:30391	Isolated biliary atresia	1-9 / 100 000
ORPHA:137914	Choanal atresia	1-9 / 1 000 000
ORPHA:182095	Interstitial lung disease	1-5 / 10 000
ORPHA:91364	Non-specific interstitial pneumonia	1-9 / 100 000
ORPHA:39812	Graft versus host disease	1-9 / 100 000
ORPHA:117	Behçet disease	1-9 / 100 000
ORPHA:2137	Autoimmune hepatitis	1-9 / 100 000
ORPHA:69	Amyloidosis	1-9 / 100 000
ORPHA:2932	Chronic inflammatory demyelinating polyneuropathy	1-9 / 100 000
ORPHA:1303	Bronchiolitis obliterans	1-9 / 100 000
ORPHA:90281	Discoid lupus erythematosus	1-9 / 1 000 000
ORPHA:3303	Tetralogy of Fallot	1-9 / 100 000
ORPHA:180086	Didelphys uterus	1-9 / 100 000
ORPHA:171	Primary sclerosing cholangitis	1-9 / 1 000 000
ORPHA:85414	Systemic-onset juvenile idiopathic arthritis	1-9 / 100 000
ORPHA:92	Juvenile idiopathic arthritis	1-9 / 100 000
ORPHA:36235	Staphylococcal scarlet fever	1-9 / 100 000
ORPHA:137577	Neonatal hypoxic and ischemic brain injury	1-9 / 1 000 000
ORPHA:809	Mixed connective tissue disease	1-9 / 100 000
ORPHA:180122	Septate uterus	1-9 / 1 000 000
ORPHA:180129	Partial septate uterus	1-9 / 1 000 000
ORPHA:66529	Tako-Tsubo cardiomyopathy	1-9 / 100 000
ORPHA:399329	Epiphysiolysis of the hip	1-9 / 100 000
ORPHA:482	Kimura disease	1-9 / 1 000 000
ORPHA:95708	Rare precocious puberty	1-9 / 100 000
ORPHA:254395	Actinic lichen planus	<1 / 1 000 000
ORPHA:158014	Rosai-Dorfman disease	1-9 / 1 000 000
ORPHA:85438	Enthesitis-related juvenile idiopathic arthritis	1-9 / 100 000
ORPHA:199306	Cleft lip/palate	1-9 / 100 000
ORPHA:411527	Central retinal vein occlusion	1-9 / 100 000
ORPHA:238624	Idiopathic intracranial hypertension	1-9 / 100 000
ORPHA:1457	Aorta coarctation	1-9 / 100 000
ORPHA:83465	Narcolepsy type 2	1-9 / 1 000 000
ORPHA:90056	Moderate and severe traumatic brain injury	1-9 / 100 000
ORPHA:3287	Takayasu arteritis	1-9 / 100 000
ORPHA:94058	Neovascular glaucoma	1-9 / 100 000
ORPHA:90033	Autoimmune hemolytic anemia, warm type	1-9 / 100 000
ORPHA:98375	Autoimmune hemolytic anemia	1-9 / 100 000
ORPHA:402035	Eosinophilic colitis	1-9 / 1 000 000
ORPHA:660	Omphalocele	1-9 / 100 000
ORPHA:180182	Supernumerary breasts	1-9 / 100 000

ORPHA:2248	Hypoplastic left heart syndrome	1-9 / 1 000 000
ORPHA:70476	Vernal keratoconjunctivitis	1-9 / 100 000
ORPHA:391673	Necrotizing enterocolitis	1-9 / 100 000
ORPHA:2302	Asbestos intoxication	1-9 / 1 000 000
ORPHA:622014	Autoimmune encephalitis	1-9 / 100 000
ORPHA:54057	Thrombotic thrombocytopenic purpura	1-9 / 100 000
ORPHA:31202	Melioidosis	1-9 / 100 000
ORPHA:180134	Bicornuate uterus	1-9 / 100 000
ORPHA:2140	Congenital diaphragmatic hernia	1-9 / 100 000
ORPHA:2014	Cleft palate	1-9 / 100 000
ORPHA:158000	Juvenile xanthogranuloma	1-9 / 100 000
ORPHA:52759	Vasculitis	1-9 / 100 000
ORPHA:29207	Reactive arthritis	1-9 / 100 000
ORPHA:183	Eosinophilic granulomatosis with polyangiitis	1-9 / 100 000
ORPHA:64745	Pruritic urticarial papules and plaques of pregnancy	1-9 / 100 000
ORPHA:99772	Cleft velum	1-9 / 100 000
ORPHA:728	Relapsing polychondritis	1-9 / 1 000 000
ORPHA:900	Granulomatosis with polyangiitis	1-9 / 100 000
ORPHA:49804	Lichen amyloidosis	1-9 / 1 000 000
ORPHA:563	Peripartum cardiomyopathy	1-9 / 1 000 000
ORPHA:69665	Intrahepatic cholestasis of pregnancy	1-9 / 100 000
ORPHA:3020	Ramsay Hunt syndrome	1-9 / 100 000
ORPHA:3389	Tuberculosis	1-9 / 100 000
ORPHA:93569	Polymyalgia rheumatica	1-9 / 100 000
ORPHA:85408	Rheumatoid factor-negative polyarticular juvenile idiopathic arthritis	1-9 / 1 000 000
ORPHA:854	Primitive portal vein thrombosis	1-9 / 100 000
ORPHA:85410	Oligoarticular juvenile idiopathic arthritis	1-9 / 1 000 000
ORPHA:263479	Fuchs heterochromic iridocyclitis	<1 / 1 000 000
ORPHA:535	Rare cutaneous lupus erythematosus	1-9 / 1 000 000
ORPHA:889	Cutaneous small vessel vasculitis	1-9 / 100 000
ORPHA:71211	Neuromyelitis optica spectrum disorder	1-9 / 100 000
ORPHA:178493	Myopic macular degeneration	1-9 / 1 000 000
ORPHA:70591	Chronic thromboembolic pulmonary hypertension	1-9 / 100 000
ORPHA:1214	Progressive hemifacial atrophy	1-9 / 1 000 000
ORPHA:228312	Autoimmune hemolytic anemia, cold type	1-9 / 100 000
ORPHA:93672	Juvenile dermatomyositis	1-9 / 100 000
ORPHA:280062	Calciphylaxis	1-9 / 100 000
ORPHA:2368	Gastroschisis	1-9 / 1 000 000
ORPHA:521219	Mirizzi syndrome	1-9 / 100 000
ORPHA:488	Urachal cyst	1-9 / 100 000
ORPHA:66627	Tenosynovial giant cell tumor	1-9 / 100 000

ORPHA:583861	Isolated mesenteric vein thrombosis	1-9 / 1 000 000
ORPHA:103919	Autoimmune pancreatitis	1-9 / 100 000
ORPHA:499096	Isolated optic neuritis	1-9 / 100 000
ORPHA:2901	Neuralgic amyotrophy	1-9 / 100 000
ORPHA:95613	Pituitary apoplexy	1-9 / 100 000
ORPHA:90002	Undifferentiated connective tissue syndrome	1-9 / 100 000
ORPHA:3437	Vogt-Koyanagi-Harada disease	1-9 / 1 000 000
ORPHA:97214	Eisenmenger syndrome	1-9 / 1 000 000
ORPHA:79128	Lymphoid interstitial pneumonia	1-9 / 1 000 000
ORPHA:210	Cyclosporiasis	<1 / 1 000 000
ORPHA:329217	Cerebral sinovenous thrombosis	1-9 / 100 000
ORPHA:90058	Spinal cord injury	1-9 / 100 000
ORPHA:99981	Apnea of prematurity	1-9 / 1 000 000
ORPHA:70588	Meconium aspiration syndrome	1-9 / 1 000 000
ORPHA:3465	Worster-Drought syndrome	1-9 / 1 000 000
ORPHA:1164	Allergic bronchopulmonary aspergillosis	1-9 / 1 000 000
ORPHA:294422	Chronic intestinal failure	1-9 / 1 000 000
ORPHA:97335	Osgood-Schlatter disease	1-9 / 1 000 000
ORPHA:99745	Typhoid	1-9 / 100 000
ORPHA:1959	Evans syndrome	1-9 / 100 000
ORPHA:31204	Nocardiosis	1-9 / 1 000 000
ORPHA:1489	Whooping cough	1-9 / 1 000 000
ORPHA:79086	Acquired generalized lipodystrophy	<1 / 1 000 000
ORPHA:767	Polyarteritis nodosa	1-9 / 1 000 000
ORPHA:90062	Acute liver failure	1-9 / 1 000 000
ORPHA:280892	Posterior uveitis	1-9 / 1 000 000
ORPHA:2444	Congenital pulmonary airway malformation	1-9 / 100 000
ORPHA:443173	Postpartum psychosis	1-9 / 1 000 000
ORPHA:99931	Idiopathic pulmonary hemosiderosis	1-9 / 1 000 000
ORPHA:99124	Congenital partial pulmonary venous return anomaly	1-9 / 1 000 000
ORPHA:158032	Hemophagocytic syndrome	1-9 / 1 000 000
ORPHA:36204	Intestinal lymphangiectasia	<1 / 1 000 000
ORPHA:85435	Rheumatoid factor-positive polyarticular juvenile idiopathic arthritis	1-9 / 1 000 000
ORPHA:99921	Chronic graft versus host disease	1-9 / 1 000 000
ORPHA:104008	Short bowel syndrome	1-9 / 100 000
ORPHA:79138	Bickerstaff brainstem encephalitis	<1 / 1 000 000
ORPHA:533	Listeriosis	1-9 / 1 000 000
ORPHA:81	Antisynthetase syndrome	1-9 / 100 000
ORPHA:2583	Mycetoma	1-9 / 1 000 000
ORPHA:2103	Guillain-Barré syndrome	1-9 / 100 000
ORPHA:280898	Panuveitis	1-9 / 1 000 000

ORPHA:139402	Drug reaction with eosinophilia and systemic symptoms	1-9 / 100 000
ORPHA:97292	Cardiogenic shock	1-9 / 1 000 000
ORPHA:36237	Bullous impetigo	1-9 / 1 000 000
ORPHA:90059	Sudden sensorineural hearing loss	1-9 / 1 000 000
ORPHA:167635	Scleromyxedema	1-9 / 1 000 000
ORPHA:829	Adult-onset Still disease	1-9 / 1 000 000
ORPHA:90065	Acquired aneurysmal subarachnoid hemorrhage	1-9 / 1 000 000
ORPHA:68354	Rare sleep disorder	1-9 / 1 000 000
ORPHA:1183	Opsoclonus-myoclonus syndrome	1-9 / 1 000 000
ORPHA:100057	Renin-angiotensin-aldosterone system-blocker-induced angioedema	<1 / 1 000 000
ORPHA:90076	Partial deep dermal and full thickness burns	1-9 / 100 000
ORPHA:71203	Autoimmune thrombocytopenia	1-9 / 1 000 000
ORPHA:97337	Sinding-Larsen-Johansson disease	1-9 / 1 000 000
ORPHA:99125	Congenital total pulmonary venous return anomaly	1-9 / 1 000 000
ORPHA:279882	Spasmus nutans	1-9 / 1 000 000
ORPHA:64722	Granulomatous mastitis	1-9 / 1 000 000
ORPHA:99107	Atrial septal aneurysm	1-9 / 1 000 000
ORPHA:803	Amyotrophic lateral sclerosis	1-9 / 1 000 000
ORPHA:69061	Idiopathic steroid-sensitive nephrotic syndrome	1-9 / 1 000 000
ORPHA:300564	Combined pulmonary fibrosis-emphysema syndrome	1-9 / 1 000 000
ORPHA:137686	Asherman syndrome	1-9 / 1 000 000
ORPHA:1207	Pulmonary atresia with ventricular septal defect	1-9 / 100 000
ORPHA:548	Leprosy	1-9 / 100 000
ORPHA:797	Sarcoidosis	1-9 / 100 000
ORPHA:33208	Idiopathic hypersomnia	1-9 / 1 000 000
ORPHA:97330	Thoracic outlet syndrome	1-9 / 1 000 000
ORPHA:431341	Patent urachus	1-9 / 100 000
ORPHA:199323	Endophthalmitis	1-9 / 100 000
ORPHA:499009	Congenital syphilis	<1 / 1 000 000
ORPHA:93126	Pauci-immune glomerulonephritis	1-9 / 1 000 000
ORPHA:1546	Cryptococcosis	1-9 / 100 000
ORPHA:353253	Burning mouth syndrome	1-9 / 1 000 000
ORPHA:155878	Submucosal cleft palate	1-9 / 100 000
ORPHA:853	Fetal and neonatal alloimmune thrombocytopenia	1-9 / 1 000 000
ORPHA:1208	Pulmonary atresia-intact ventricular septum syndrome	1-9 / 1 000 000
ORPHA:251312	Overlapping connective tissue disease	1-9 / 1 000 000
ORPHA:945	Acalvaria	1-9 / 1 000 000
ORPHA:137814	Macular amyloidosis	1-9 / 1 000 000
ORPHA:1041	Hydrops fetalis	1-9 / 1 000 000
ORPHA:2764	Osteochondritis dissecans	1-9 / 1 000 000
ORPHA:43393	Lambert-Eaton myasthenic syndrome	<1 / 1 000 000

ORPHA:2041	Coronary arterial fistula	1-9 / 1 000 000
ORPHA:140949	Low-flow priapism	<1 / 1 000 000
ORPHA:2070	Eosinophilic gastroenteritis	1-9 / 1 000 000
ORPHA:95707	Idiopathic isolated micropenis	1-9 / 1 000 000
ORPHA:83452	Complex regional pain syndrome	1-9 / 100 000
ORPHA:84085	Hinman syndrome	<1 / 1 000 000
ORPHA:228119	Fusariosis	1-9 / 1 000 000
ORPHA:727	Microscopic polyangiitis	1-9 / 1 000 000
ORPHA:141291	Cleft lip and alveolus	1-9 / 1 000 000
ORPHA:399293	Osteonecrosis of the jaw	1-9 / 1 000 000
ORPHA:93573	Thrombotic microangiopathy	1-9 / 1 000 000
ORPHA:764	Pyomyositis	1-9 / 1 000 000
ORPHA:68364	Hemoglobinopathy	1-9 / 1 000 000
ORPHA:441	Pure autonomic failure	1-9 / 1 000 000
ORPHA:158048	Hemophagocytic syndrome associated with an infection	<1 / 1 000 000
ORPHA:447774	Secondary sclerosing cholangitis	<1 / 1 000 000
ORPHA:71273	Renal nutcracker syndrome	1-9 / 1 000 000
ORPHA:1081	Coronary artery congenital malformation	1-9 / 1 000 000
ORPHA:158061	Macrophage activation syndrome	1-9 / 1 000 000
ORPHA:3181	Sprengel deformity	1-9 / 1 000 000
ORPHA:2035	Lymphatic filariasis	1-9 / 1 000 000
ORPHA:70578	Adult acute respiratory distress syndrome	1-9 / 1 000 000
ORPHA:99095	Congenital Gerbode defect	1-9 / 1 000 000
ORPHA:169105	Good syndrome	1-9 / 1 000 000
ORPHA:60032	Recurrent respiratory papillomatosis	1-9 / 1 000 000
ORPHA:293812	Fixed drug eruption	1-9 / 100 000
ORPHA:67043	Amoebic keratitis	1-9 / 1 000 000
ORPHA:279914	Intermediate uveitis	1-9 / 1 000 000
ORPHA:3282	Multifocal atrial tachycardia	1-9 / 1 000 000
ORPHA:411501	Williams-Campbell syndrome	1-9 / 1 000 000
ORPHA:131	Budd-Chiari syndrome	1-9 / 1 000 000
ORPHA:141234	Median facial cleft	<1 / 1 000 000
ORPHA:217067	Pouchitis	1-9 / 1 000 000
ORPHA:48104	Pyoderma gangrenosum	1-9 / 1 000 000
ORPHA:99131	Pleuro-pericardial cyst	1-9 / 1 000 000
ORPHA:200	Isolated corpus callosum agenesis	1-9 / 1 000 000
ORPHA:1467	Cogan syndrome	1-9 / 1 000 000
ORPHA:49041	IgG4-related retroperitoneal fibrosis	1-9 / 1 000 000
ORPHA:221098	Glossopharyngeal neuralgia	1-9 / 1 000 000
ORPHA:2477	Megalencephaly	1-9 / 1 000 000
ORPHA:91354	Pituitary deficiency due to empty sella turcica syndrome	<1 / 1 000 000
ORPHA:641	Multifocal motor neuropathy	1-9 / 100 000

ORPHA:140896	Severe acute respiratory syndrome	1-9 / 1 000 000
ORPHA:188	Systemic capillary leak syndrome	1-9 / 1 000 000
ORPHA:293173	Acute generalized exanthematous pustulosis	1-9 / 100 000
ORPHA:3033	Renal tubular dysgenesis	1-9 / 1 000 000
ORPHA:185	Scimitar syndrome	1-9 / 1 000 000
ORPHA:163934	Atopic keratoconjunctivitis	<1 / 1 000 000
ORPHA:398124	Neonatal lupus erythematosus	1-9 / 1 000 000
ORPHA:289390	Primary Sjögren syndrome	<1 / 1 000 000
ORPHA:519408	Mooren ulcer	1-9 / 1 000 000
ORPHA:220393	Diffuse cutaneous systemic sclerosis	1-9 / 1 000 000
ORPHA:171700	Diffuse panbronchiolitis	1-9 / 1 000 000
ORPHA:3099	Rheumatic fever	1-9 / 1 000 000
ORPHA:216694	Congenitally corrected transposition of the great arteries	1-9 / 1 000 000
ORPHA:178478	Infant botulism	1-9 / 1 000 000
ORPHA:704	Pemphigus vulgaris	1-9 / 100 000
ORPHA:79481	Pemphigus foliaceus	1-9 / 1 000 000
ORPHA:65684	Monomelic amyotrophy	<1 / 1 000 000
ORPHA:95498	Congenital anomaly of superior vena cava	1-9 / 1 000 000
ORPHA:209964	Solitary rectal ulcer syndrome	1-9 / 1 000 000
ORPHA:85138	Addison disease	<1 / 1 000 000
ORPHA:206569	Immune-mediated necrotizing myopathy	1-9 / 1 000 000
ORPHA:171673	Limbic stem cell deficiency	1-9 / 1 000 000
ORPHA:180139	Uterine hypoplasia	<1 / 1 000 000
ORPHA:2038	Pulmonary arteriovenous malformation	<1 / 1 000 000
ORPHA:549	Legionnaires disease	<1 / 1 000 000
ORPHA:858	Congenital toxoplasmosis	<1 / 1 000 000
ORPHA:254492	Frontal fibrosing alopecia	<1 / 1 000 000
ORPHA:46488	Linear IgA dermatosis	<1 / 1 000 000
ORPHA:36397	Adiposis dolorosa	1-9 / 1 000 000
ORPHA:86908	Idiopathic hemiconvulsion-hemiplegia syndrome	<1 / 1 000 000
ORPHA:99892	ACTH-dependent Cushing syndrome	1-9 / 1 000 000
ORPHA:2842	Penoscrotal transposition	1-9 / 1 000 000
ORPHA:93568	Juvenile polymyositis	<1 / 1 000 000
ORPHA:90285	Lupus erythematosus panniculitis	1-9 / 1 000 000
ORPHA:2494	MV@nv@trier disease	<1 / 1 000 000
ORPHA:2902	Idiopathic chronic eosinophilic pneumonia	1-9 / 1 000 000
ORPHA:99083	Pulmonary artery hypoplasia	1-9 / 1 000 000
ORPHA:85436	Psoriasis-related juvenile idiopathic arthritis	<1 / 1 000 000
ORPHA:98723	Hypoplastic right heart syndrome	<1 / 1 000 000
ORPHA:95431	Twin to twin transfusion syndrome	1-9 / 1 000 000
ORPHA:99081	Right aortic arch	<1 / 1 000 000
ORPHA:220402	Limited cutaneous systemic sclerosis	1-9 / 1 000 000

ORPHA:457095	Actinomycosis	1-9 / 1 000 000
ORPHA:431347	Urachal diverticulum	<1 / 1 000 000
ORPHA:220407	Limited systemic sclerosis	1-9 / 1 000 000
ORPHA:35687	Erdheim-Chester disease	1-9 / 1 000 000
ORPHA:139436	Multicentric reticulohistiocytosis	<1 / 1 000 000
ORPHA:330064	Chronic actinic dermatitis	<1 / 1 000 000
ORPHA:447788	Cerebral visual impairment	<1 / 1 000 000
ORPHA:70587	Infant acute respiratory distress syndrome	1-9 / 1 000 000
ORPHA:36258	Buerger disease	1-9 / 1 000 000
ORPHA:264688	Congenital chylothorax	<1 / 1 000 000
ORPHA:673	Malaria	<1 / 1 000 000
ORPHA:499	Kerion celsi	1-9 / 1 000 000
ORPHA:2037	Congenital aortopulmonary window	1-9 / 1 000 000
ORPHA:83597	Acute disseminated encephalomyelitis	1-9 / 1 000 000
ORPHA:99920	Acute graft versus host disease	1-9 / 1 000 000
ORPHA:1556	Cutis marmorata telangiectatica congenita	1-9 / 1 000 000
ORPHA:282196	Autoimmune polyendocrinopathy	<1 / 1 000 000
ORPHA:206991	Viral myositis	1-9 / 1 000 000
ORPHA:1166	Congenital unilateral hypoplasia of depressor anguli oris	1-9 / 1 000 000
ORPHA:99099	Cor triatriatum sinister	<1 / 1 000 000
ORPHA:1209	Tricuspid atresia	<1 / 1 000 000
ORPHA:793	SAPHO syndrome	1-9 / 1 000 000
ORPHA:46486	Mucous membrane pemphigoid	1-9 / 1 000 000
ORPHA:90078	Invasive infections due to vancomycin-resistant enterococci	<1 / 1 000 000
ORPHA:157843	Trigeminal autonomic cephalalgia	1-9 / 1 000 000
ORPHA:36236	Staphylococcal scalded skin syndrome	1-9 / 1 000 000
ORPHA:99771	Bifid uvula	1-9 / 1 000 000
ORPHA:31740	Hypersensitivity pneumonitis	1-9 / 1 000 000
ORPHA:97252	Mega-cisterna magna	1-9 / 1 000 000
ORPHA:397	Giant cell arteritis	<1 / 1 000 000
ORPHA:244242	HELLP syndrome	1-9 / 1 000 000
ORPHA:141288	Midline cervical cleft	<1 / 1 000 000
ORPHA:137596	Neurotrophic keratopathy	<1 / 1 000 000
ORPHA:519930	Fungal keratitis	1-9 / 1 000 000
ORPHA:254463	Lichen planus pigmentosus	<1 / 1 000 000
ORPHA:79087	Acquired partial lipodystrophy	<1 / 1 000 000
ORPHA:1676	Idiopathic pulmonary artery dilatation	1-9 / 1 000 000
ORPHA:396	Chronic hiccup	<1 / 1 000 000
ORPHA:99922	Ocular cicatricial pemphigoid	1-9 / 1 000 000
ORPHA:448270	Ectopia cordis	<1 / 1 000 000
ORPHA:390	Histoplasmosis	<1 / 1 000 000
ORPHA:449266	Pleural empyema	1-9 / 1 000 000

ORPHA:98871	Transient erythroblastopenia of childhood	<1 / 1 000 000
ORPHA:98725	Ascending aorta anomaly	1-9 / 1 000 000
ORPHA:3325	Heparin-induced thrombocytopenia	<1 / 1 000 000
ORPHA:71276	Silent sinus syndrome	<1 / 1 000 000
ORPHA:36234	Bacterial toxic-shock syndrome	1-9 / 1 000 000
ORPHA:96269	Isolated partial vaginal agenesis	<1 / 1 000 000
ORPHA:65250	Perineural cyst	<1 / 1 000 000
ORPHA:95443	Mesocardia	<1 / 1 000 000
ORPHA:1047	Sideroblastic anemia	<1 / 1 000 000
ORPHA:137807	Primary cutaneous amyloidosis	1-9 / 1 000 000
ORPHA:141171	Maxillary arteriovenous malformation	<1 / 1 000 000
ORPHA:1304	Brucellosis	1-9 / 1 000 000
ORPHA:583856	Isolated splenic vein thrombosis	1-9 / 1 000 000
ORPHA:83315	Murine typhus	<1 / 1 000 000
ORPHA:1463	Triatrial heart	1-9 / 1 000 000
ORPHA:79102	Thyrotoxic periodic paralysis	1-9 / 1 000 000
ORPHA:243367	Acute fatty liver of pregnancy	<1 / 1 000 000
ORPHA:141064	Lower lip fistula	<1 / 1 000 000
ORPHA:3198	Stiff person spectrum disorder	1-9 / 1 000 000
ORPHA:464318	Verrucous hemangioma	<1 / 1 000 000
ORPHA:2330	Kasabach-Merritt phenomenon	<1 / 1 000 000
ORPHA:98917	Acute motor and sensory axonal neuropathy	<1 / 1 000 000
ORPHA:509	Leptospirosis	<1 / 1 000 000
ORPHA:90283	Lupus erythematosus tumidus	<1 / 1 000 000
ORPHA:99111	Persistent left superior vena cava connecting to the roof of left-sided atrium	<1 / 1 000 000
ORPHA:163525	Subacute cutaneous lupus erythematosus	<1 / 1 000 000
ORPHA:99084	Peripheral pulmonary stenosis	<1 / 1 000 000
ORPHA:447881	Idiopathic dropped head syndrome	<1 / 1 000 000
ORPHA:217260	Progressive multifocal leukoencephalopathy	<1 / 1 000 000
ORPHA:810	Shigellosis	<1 / 1 000 000
ORPHA:3165	Eosinophilic fasciitis	<1 / 1 000 000
ORPHA:79126	Acute interstitial pneumonia	<1 / 1 000 000
ORPHA:221117	Gerstmann syndrome	<1 / 1 000 000
ORPHA:542592	Necrobiosis lipoidica	<1 / 1 000 000
ORPHA:76	Strongyloidiasis	1-9 / 1 000 000
ORPHA:541454	Anomalous aortic origin of the right coronary artery	<1 / 1 000 000
ORPHA:490	Omphalomesenteric cyst	<1 / 1 000 000
ORPHA:521127	Osteoradionecrosis of the mandible	<1 / 1 000 000
ORPHA:48377	Subcorneal pustular dermatosis	<1 / 1 000 000
ORPHA:3092	Fixed subaortic stenosis	<1 / 1 000 000
ORPHA:91546	Lyme disease	<1 / 1 000 000

ORPHA:99828	Dengue fever	1-9 / 1 000 000
ORPHA:91138	Cryoglobulinemic vasculitis	<1 / 1 000 000
ORPHA:99057	Congenital mitral stenosis	<1 / 1 000 000
ORPHA:1054	Aneurysm of sinus of Valsalva	<1 / 1 000 000
ORPHA:99147	Acquired von Willebrand syndrome	<1 / 1 000 000
ORPHA:2406	Locked-in syndrome	<1 / 1 000 000

Table S4. Comparative overview of the Orphanet prevalence against the computed prevalence for underdiagnosed conditions.

Orpha code	Disease name	Orphanet prevalence	Computed prevalence
ORPHA:95719	Thyroid hemiagenesis	1-5 / 10 000	<1 / 1 000 000
ORPHA:2828	Young-onset Parkinson disease	1-5 / 10 000	<1 / 1 000 000
ORPHA:221061	Familial cerebral cavernous malformation	1-5 / 10 000	<1 / 1 000 000
ORPHA:247	Inherited arrhythmogenic cardiomyopathy	1-5 / 10 000	1-9 / 1 000 000
ORPHA:288	Hereditary elliptocytosis	1-5 / 10 000	1-9 / 1 000 000
ORPHA:214	Cystinuria	1-5 / 10 000	1-9 / 1 000 000
ORPHA:273	Steinert myotonic dystrophy	1-5 / 10 000	1-9 / 1 000 000
ORPHA:3109	Mayer-Rokitansky-Küster-Hauser syndrome	1-5 / 10 000	1-9 / 1 000 000
ORPHA:65753	Charcot-Marie-Tooth disease type 1	1-5 / 10 000	1-9 / 1 000 000
ORPHA:908	Fragile X syndrome	1-5 / 10 000	1-9 / 1 000 000
ORPHA:791	Retinitis pigmentosa	1-5 / 10 000	1-9 / 1 000 000
ORPHA:100088	Rare thyroid carcinoma	1-5 / 10 000	1-9 / 1 000 000
ORPHA:2467	Systemic mastocytosis	1-5 / 10 000	1-9 / 1 000 000
ORPHA:853	Fetal and neonatal alloimmune thrombocytopenia	1-5 / 10 000	1-9 / 1 000 000
ORPHA:36258	Buerger disease	1-5 / 10 000	1-9 / 1 000 000
ORPHA:648	Noonan syndrome	1-5 / 10 000	1-9 / 100 000
ORPHA:558	Marfan syndrome	1-5 / 10 000	1-9 / 100 000
ORPHA:83463	Microtia	1-5 / 10 000	1-9 / 100 000
ORPHA:666	Osteogenesis imperfecta	1-5 / 10 000	1-9 / 100 000
ORPHA:388	Hirschsprung disease	1-5 / 10 000	1-9 / 100 000
ORPHA:1114	Aplasia cutis congenita	1-5 / 10 000	1-9 / 100 000
ORPHA:2032	Idiopathic pulmonary fibrosis	1-5 / 10 000	1-9 / 100 000
ORPHA:186	Primary biliary cholangitis	1-5 / 10 000	1-9 / 100 000
ORPHA:2137	Autoimmune hepatitis	1-5 / 10 000	1-9 / 100 000
ORPHA:92	Juvenile idiopathic arthritis	1-5 / 10 000	1-9 / 100 000
ORPHA:391673	Necrotizing enterocolitis	1-5 / 10 000	1-9 / 100 000
ORPHA:797	Sarcoidosis	1-5 / 10 000	1-9 / 100 000
ORPHA:99429	Complete androgen insensitivity syndrome	1-9 / 1 000 000	<1 / 1 000 000
ORPHA:71277	Classic glucose transporter type 1 deficiency syndrome	1-9 / 1 000 000	<1 / 1 000 000
ORPHA:1452	Cleidocranial dysplasia	1-9 / 1 000 000	<1 / 1 000 000

ORPHA:3463	Wolfram syndrome	1-9 / 1 000 000	<1 / 1 000 000
ORPHA:43393	Lambert-Eaton myasthenic syndrome	1-9 / 1 000 000	<1 / 1 000 000
ORPHA:89936	X-linked hypophosphatemia	1-9 / 100 000	<1 / 1 000 000
ORPHA:886	Usher syndrome	1-9 / 100 000	<1 / 1 000 000
ORPHA:79434	Oculocutaneous albinism type 1B	1-9 / 100 000	<1 / 1 000 000
ORPHA:65	Leber congenital amaurosis	1-9 / 100 000	<1 / 1 000 000
ORPHA:738	Porphyria	1-9 / 100 000	<1 / 1 000 000
ORPHA:819	Smith-Magenis syndrome	1-9 / 100 000	<1 / 1 000 000
ORPHA:1928	Congenital lobar emphysema	1-9 / 100 000	<1 / 1 000 000
ORPHA:79241	Biotinidase deficiency	1-9 / 100 000	<1 / 1 000 000
ORPHA:377	Gorlin syndrome	1-9 / 100 000	<1 / 1 000 000
ORPHA:454750	Isolated tracheoesophageal fistula	1-9 / 100 000	<1 / 1 000 000
ORPHA:107	BOR syndrome	1-9 / 100 000	<1 / 1 000 000
ORPHA:683	Progressive supranuclear palsy	1-9 / 100 000	1-9 / 1 000 000
ORPHA:42	Medium chain acyl-CoA dehydrogenase deficiency	1-9 / 100 000	1-9 / 1 000 000
ORPHA:905	Wilson disease	1-9 / 100 000	1-9 / 1 000 000
ORPHA:87	Apert syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:192	Coffin-Lowry syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:429	Hypochondroplasia	1-9 / 100 000	1-9 / 1 000 000
ORPHA:98895	Becker muscular dystrophy	1-9 / 100 000	1-9 / 1 000 000
ORPHA:83418	Proximal spinal muscular atrophy type 2	1-9 / 100 000	1-9 / 1 000 000
ORPHA:72	Angelman syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:2614	Nail-patella syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:792	X-linked retinoschisis	1-9 / 100 000	1-9 / 1 000 000
ORPHA:55	Oculocutaneous albinism	1-9 / 100 000	1-9 / 1 000 000
ORPHA:778	Rett syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:685	Hereditary spastic paraplegia	1-9 / 100 000	1-9 / 1 000 000
ORPHA:399	Huntington disease	1-9 / 100 000	1-9 / 1 000 000
ORPHA:98757	Spinocerebellar ataxia type 3	1-9 / 100 000	1-9 / 1 000 000
ORPHA:79254	Classic phenylketonuria	1-9 / 100 000	1-9 / 1 000 000
ORPHA:821	Sotos syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:171	Primary sclerosing cholangitis	1-9 / 100 000	1-9 / 1 000 000
ORPHA:99125	Congenital total pulmonary venous return anomaly	1-9 / 100 000	1-9 / 1 000 000
ORPHA:803	Amyotrophic lateral sclerosis	1-9 / 100 000	1-9 / 1 000 000
ORPHA:727	Microscopic polyangiitis	1-9 / 100 000	1-9 / 1 000 000
ORPHA:131	Budd-Chiari syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:36234	Bacterial toxic-shock syndrome	1-9 / 100 000	1-9 / 1 000 000
ORPHA:79373	Ectodermal dysplasia syndrome	6-9 / 10 000	1-9 / 1 000 000

Figure S1. Distribution of the median information content and the information entropy per clinical encounter for Neoplasms

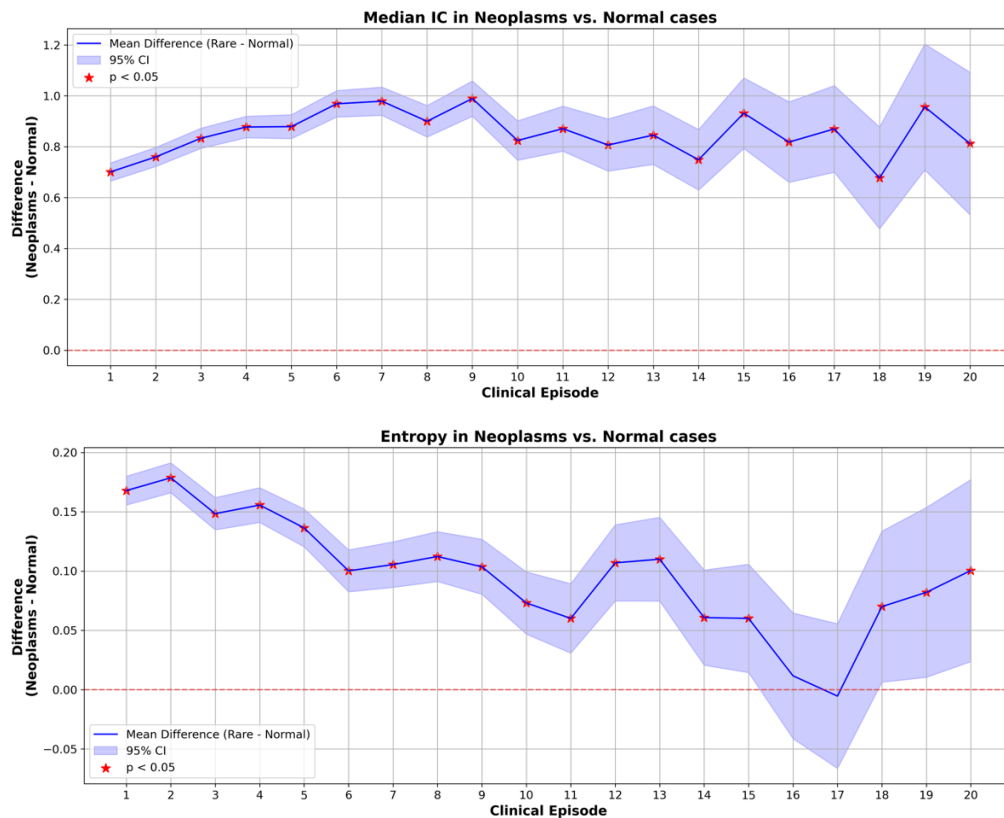


Figure S1. Distribution of the median information content and the information entropy per clinical encounter for Other (non-genetic, non-neoplastic) rare diseases

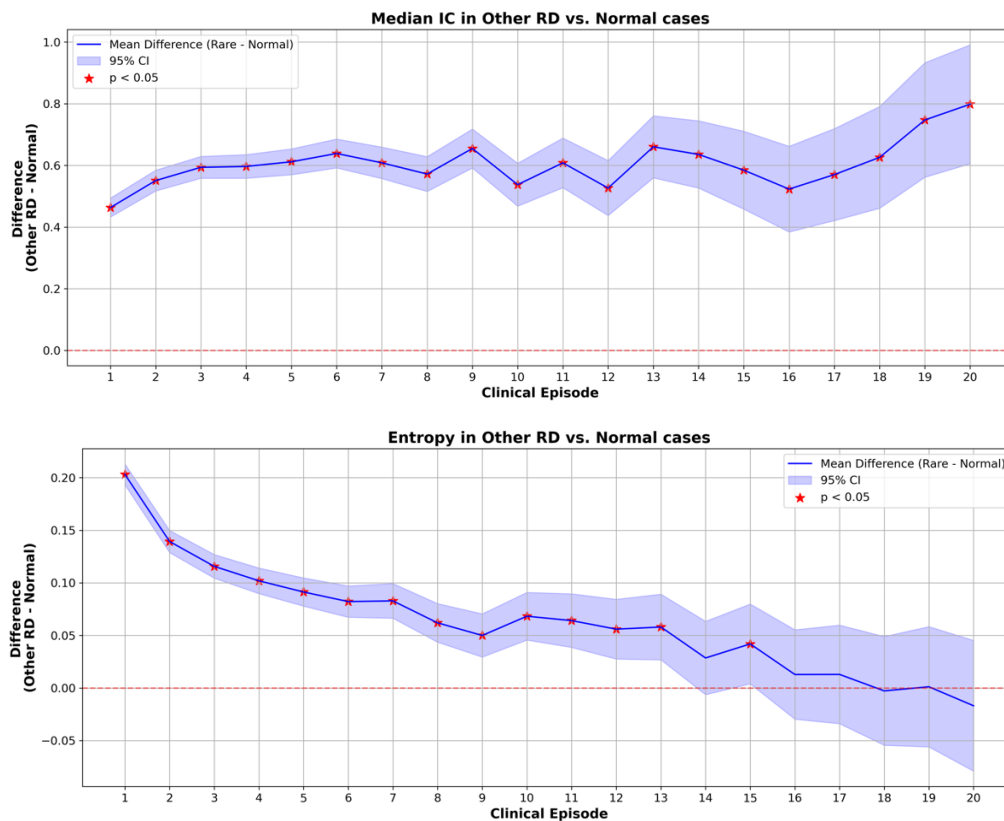


Figure S3. Distribution of the median information content and the information entropy per clinical encounter for underdiagnosed conditions

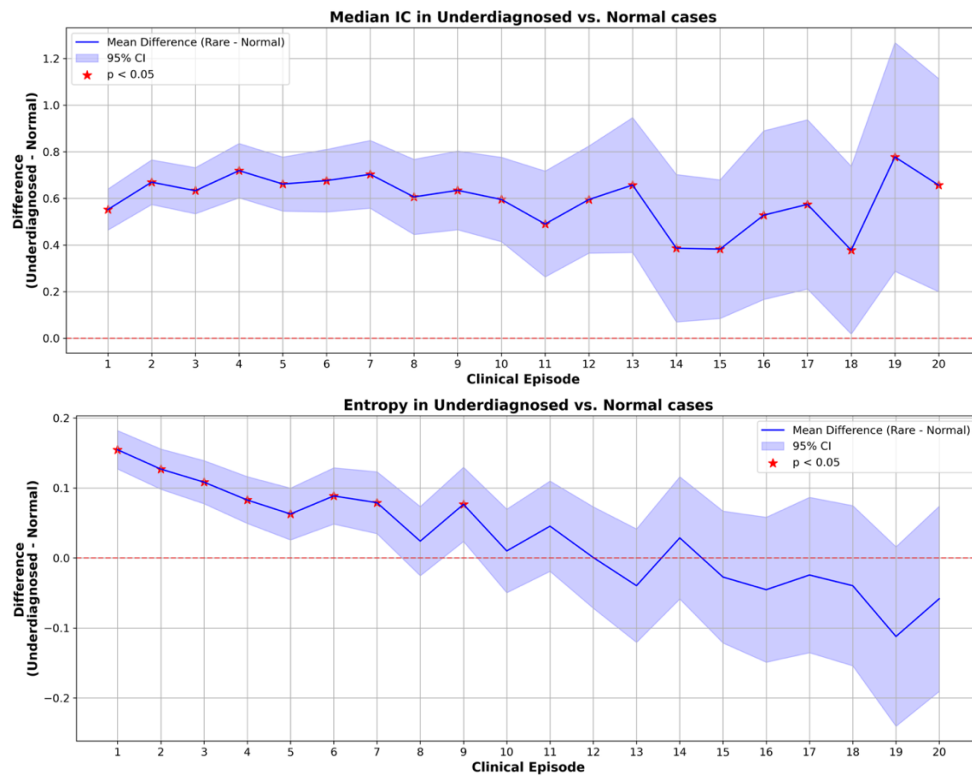


Figure S4. Distribution of the median information content and the information entropy per clinical encounter for Genetic conditions in children vs adults

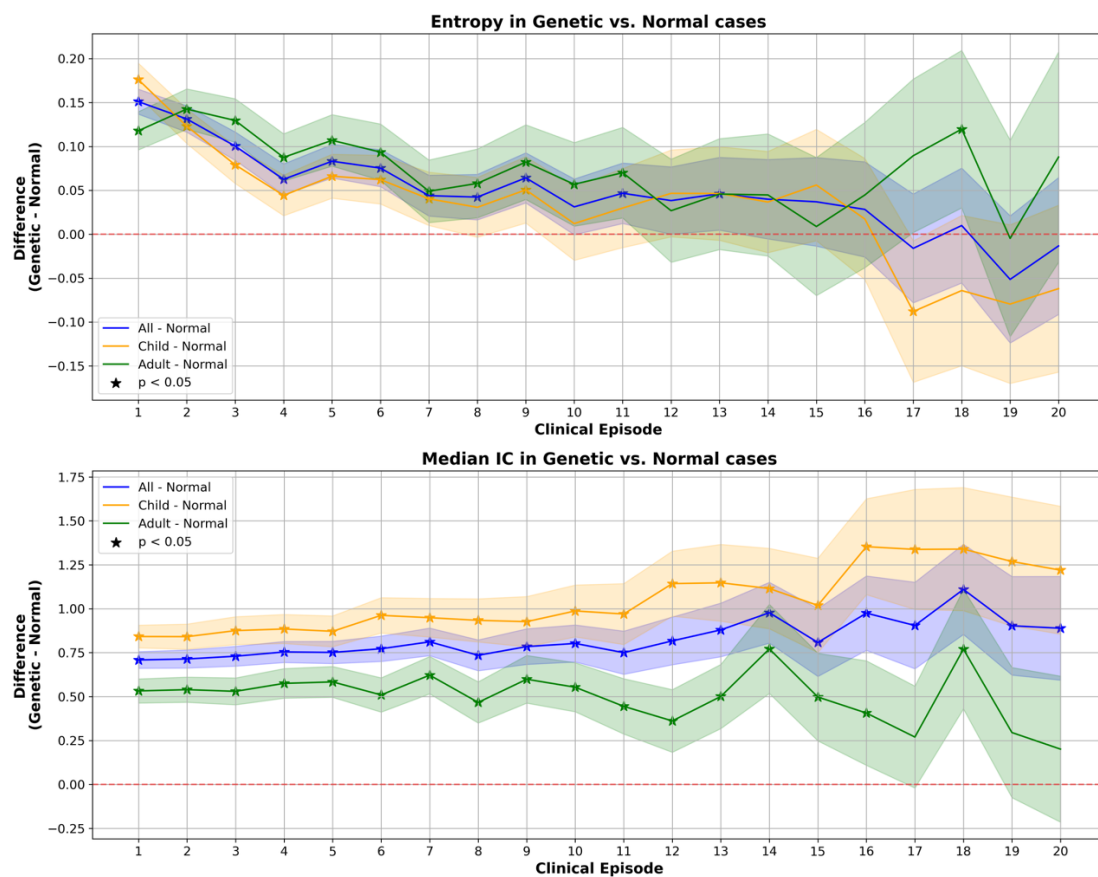


Figure S5. Distribution of the median information content and the information entropy per clinical encounter for Neoplasms in children vs adults

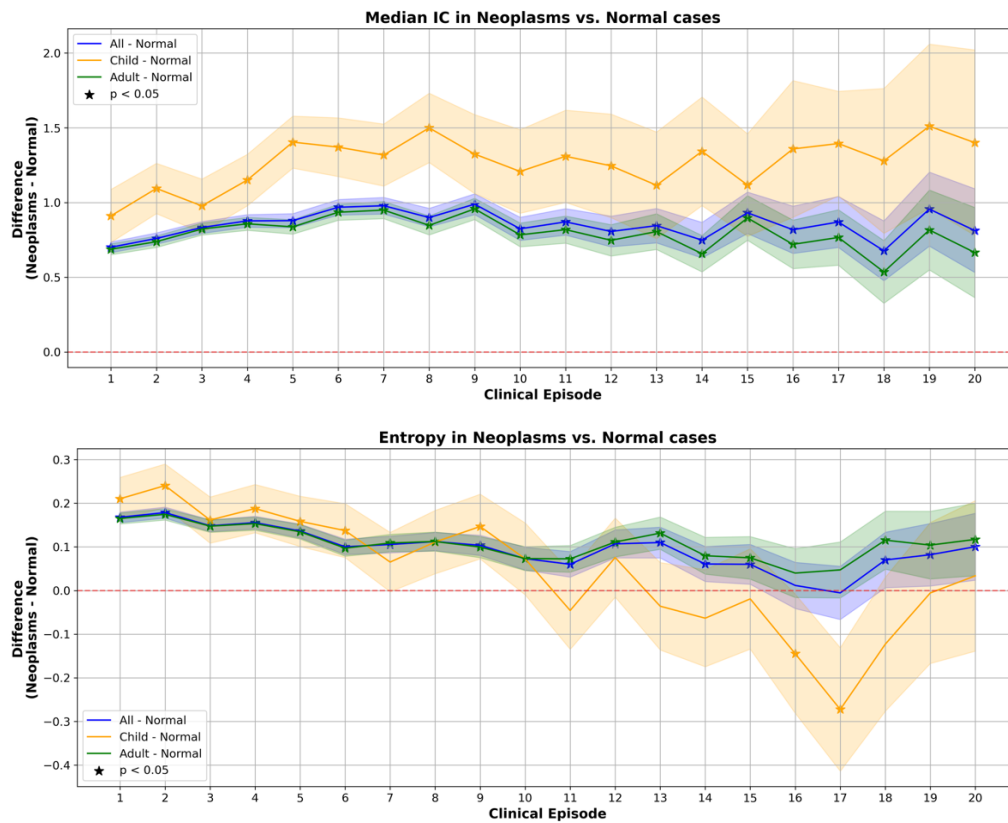


Figure S6. Distribution of the median information content and the information entropy per clinical encounter for Other RDs in children vs adults

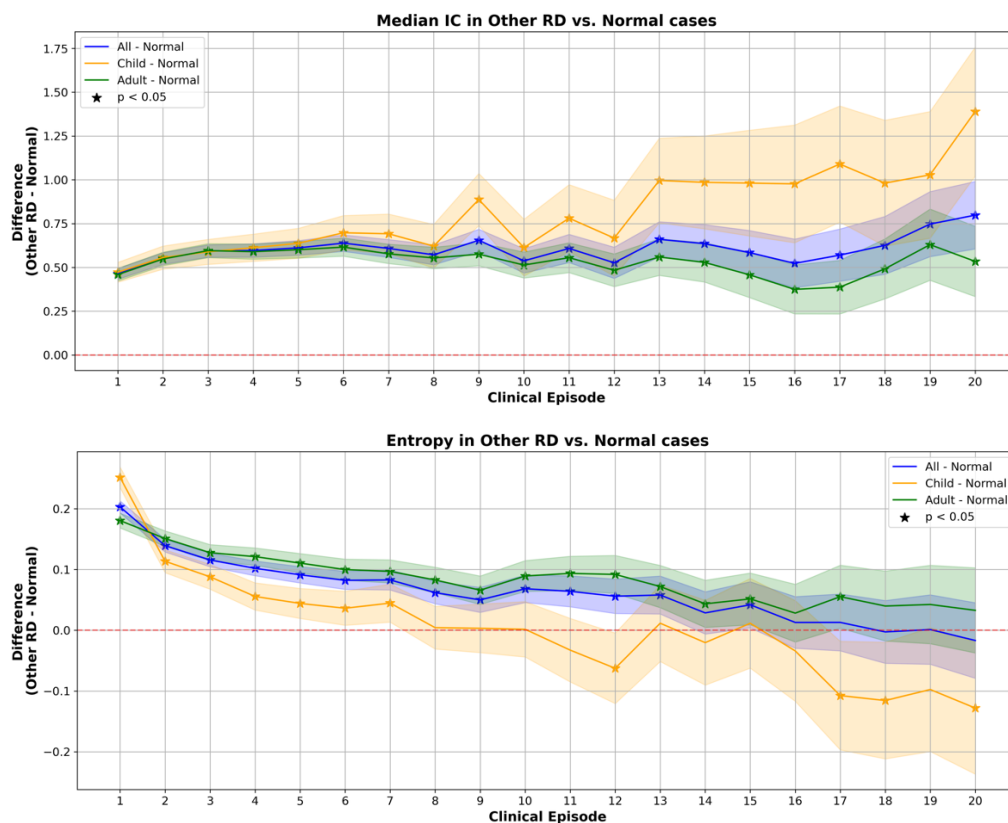


Figure S7. Distribution of the median information content and the information entropy per clinical encounter for Genetic conditions in Males vs Females

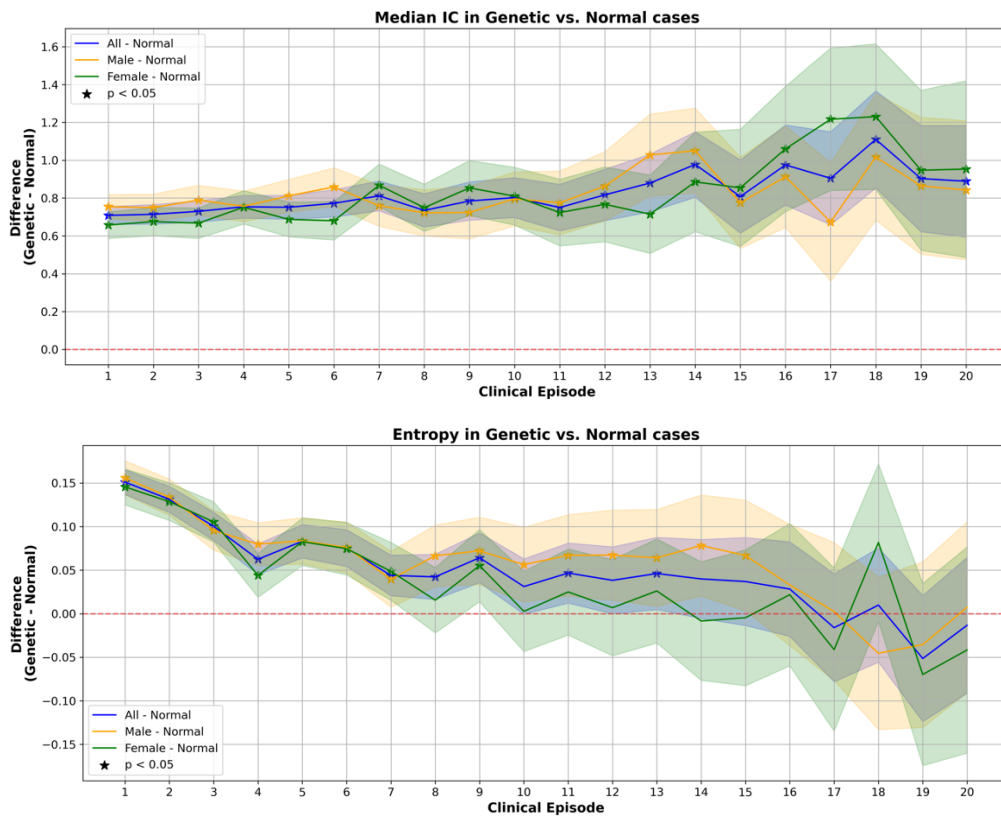


Figure S8. Distribution of the median information content and the information entropy per clinical encounter for Neoplasms in Males vs Females

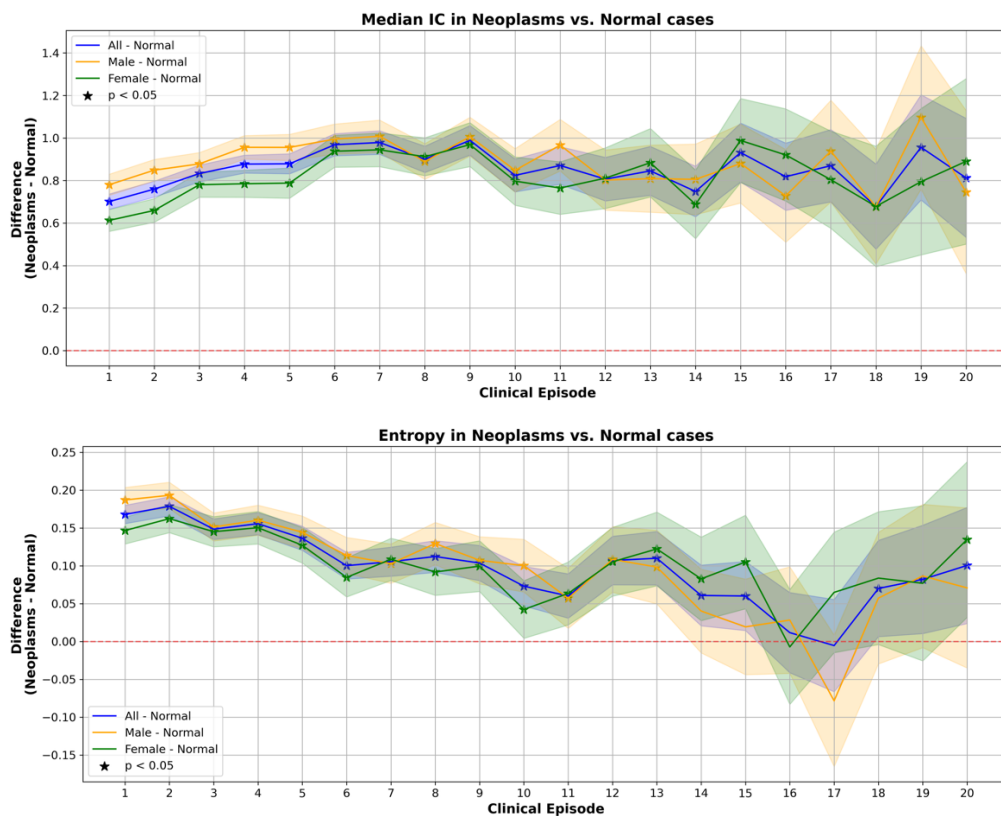


Figure S9. Distribution of the median information content and the information entropy per clinical encounter for Other RDs in Males vs Females

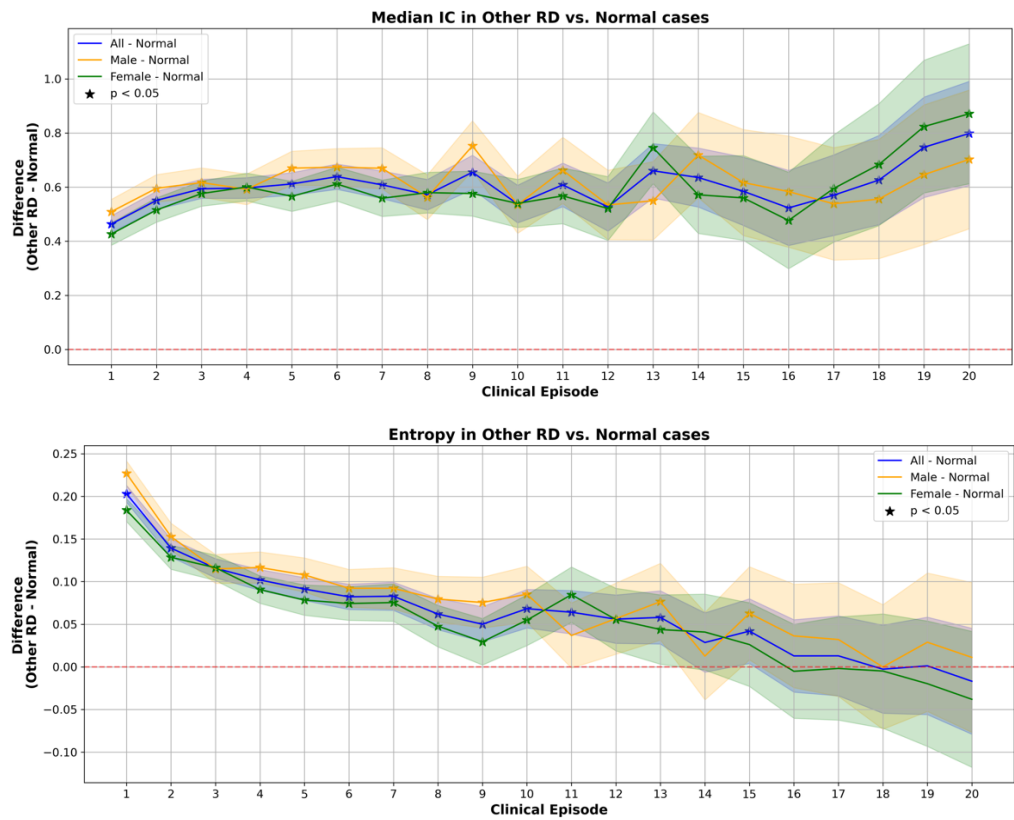


Figure S10. High-level overview of the stratification outcomes subject to the encounter that contained the initial diagnosis and split onto the three major classes of diseases: Genetic, Neoplasms and Other RDs.

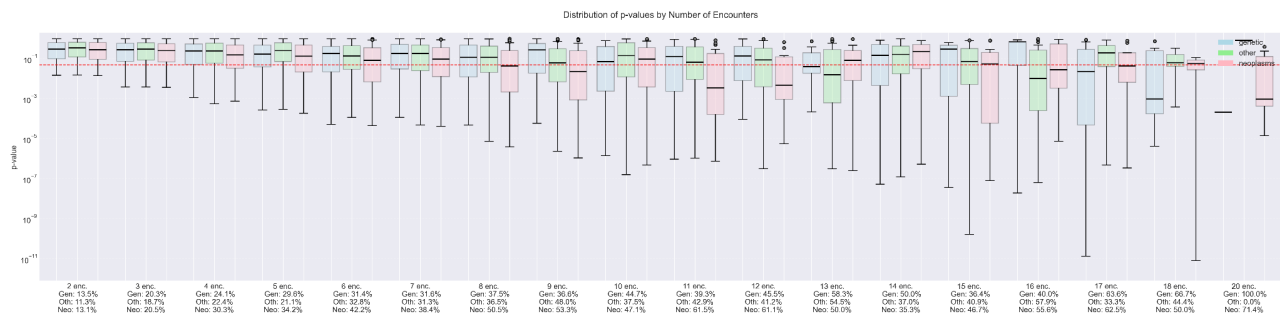


Figure S11. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Neoplastic disease patients. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

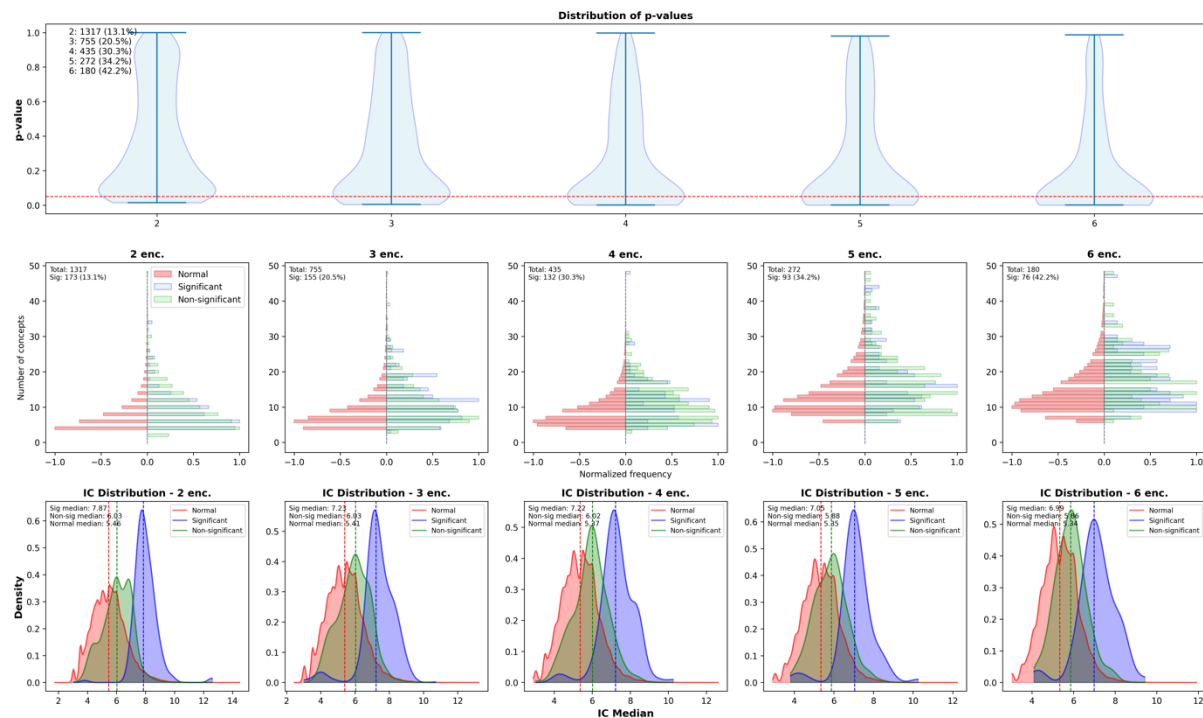


Figure S12. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Other RD patients. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

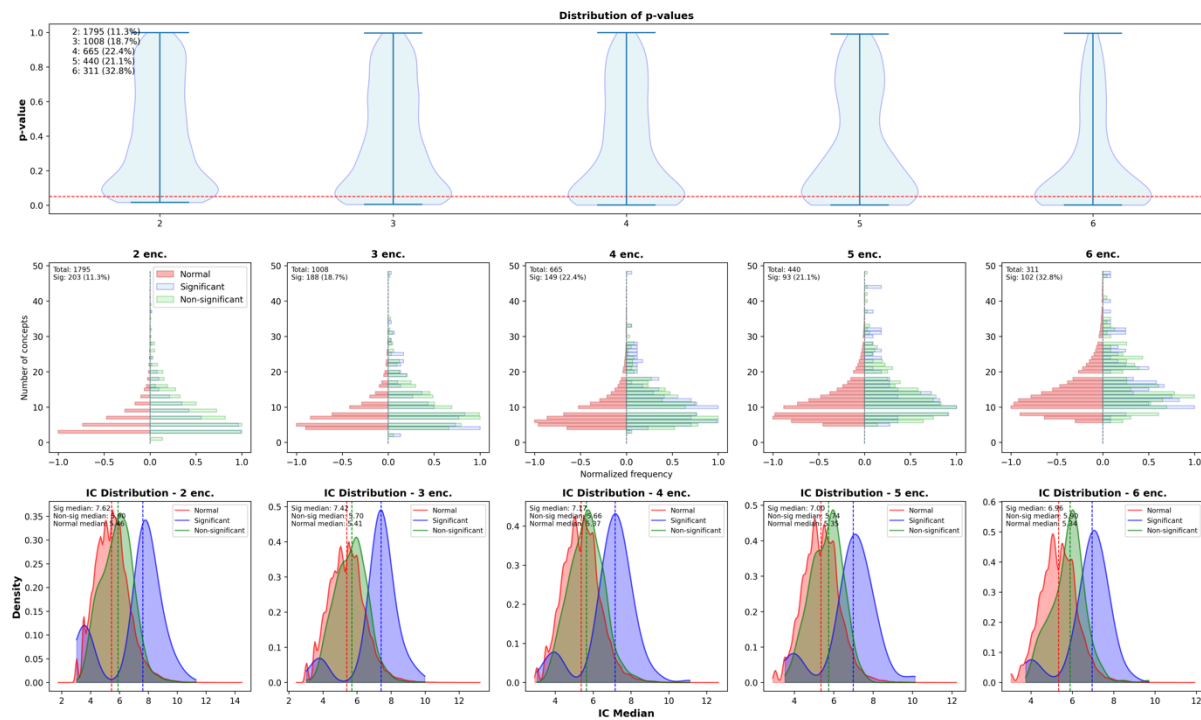


Figure S13. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Genetic disease patients. The top panel displays the distribution of p-values for each stratified group (ranging from 7 to 11 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

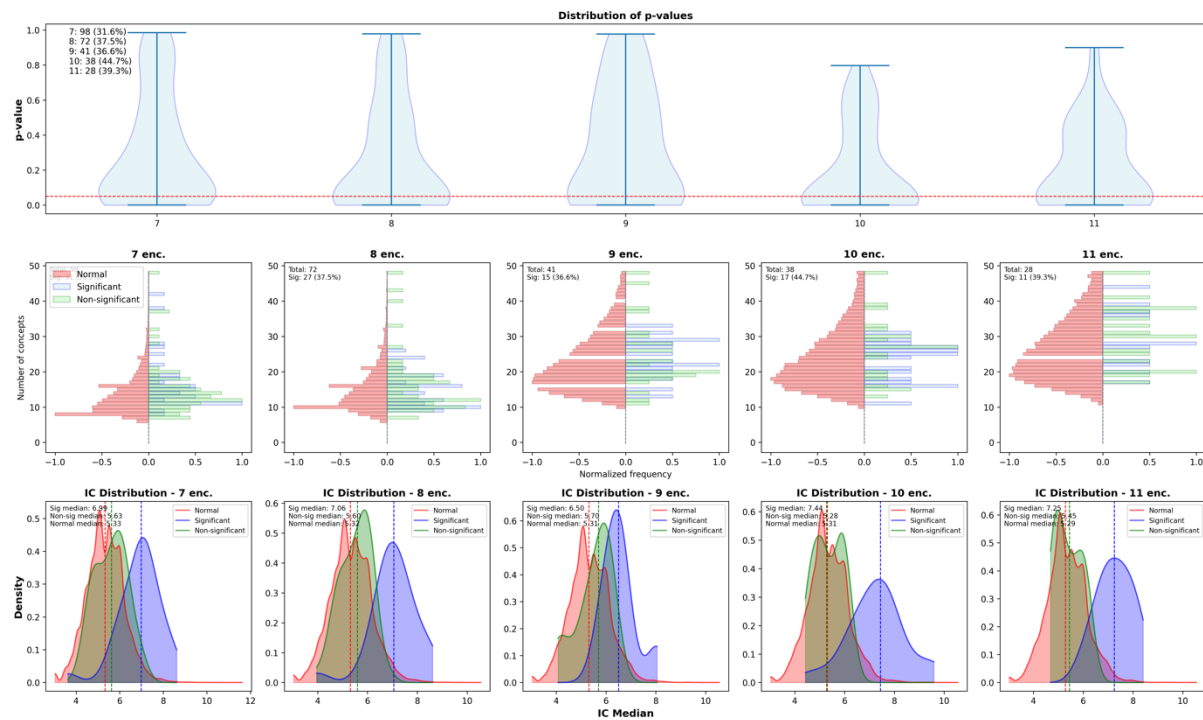


Figure S14. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Neoplastic disease patients. The top panel displays the distribution of p-values for each stratified group (ranging from 7 to 11 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

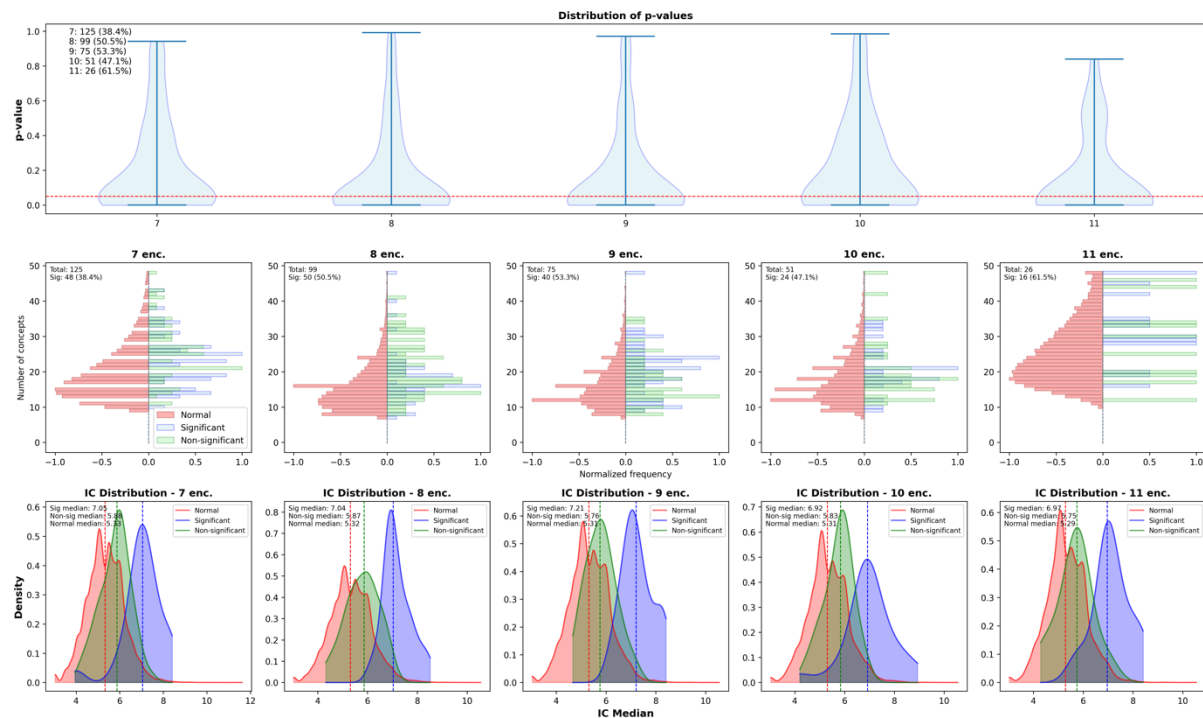


Figure S15. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Other RD patients. The top panel displays the distribution of p-values for each stratified group (ranging from 7 to 11 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

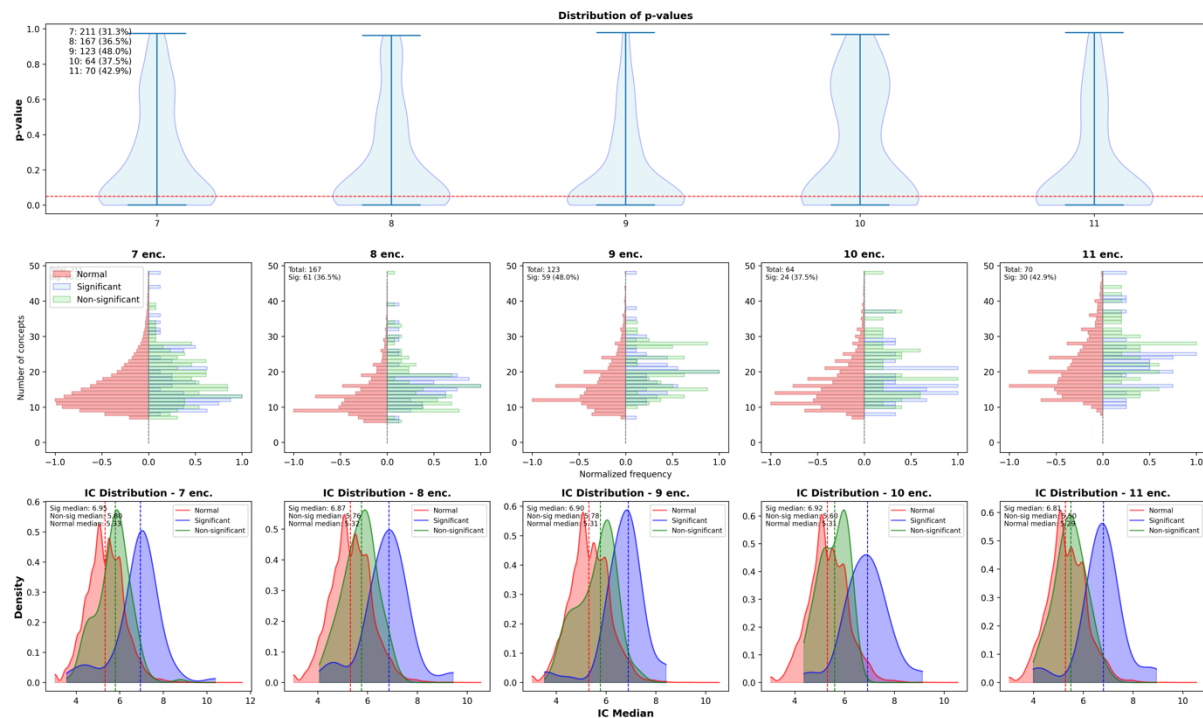


Figure S16. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Underdiagnosed disease patients. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

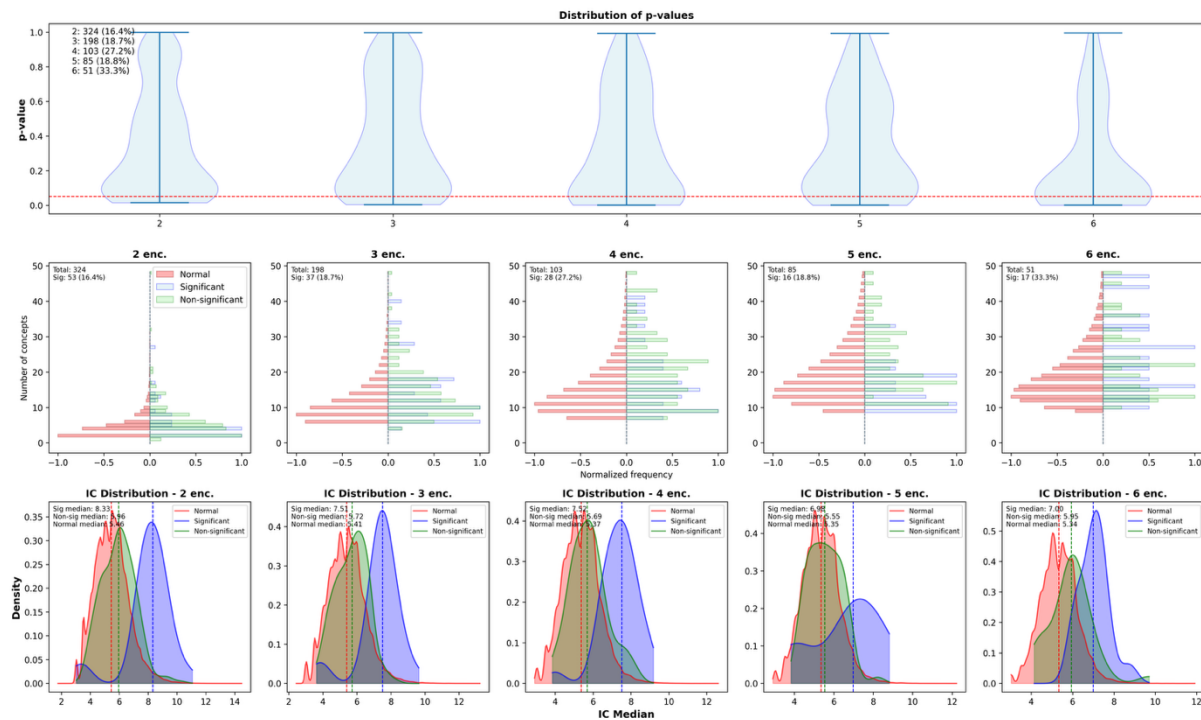


Figure S17. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Children across all disease groups. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

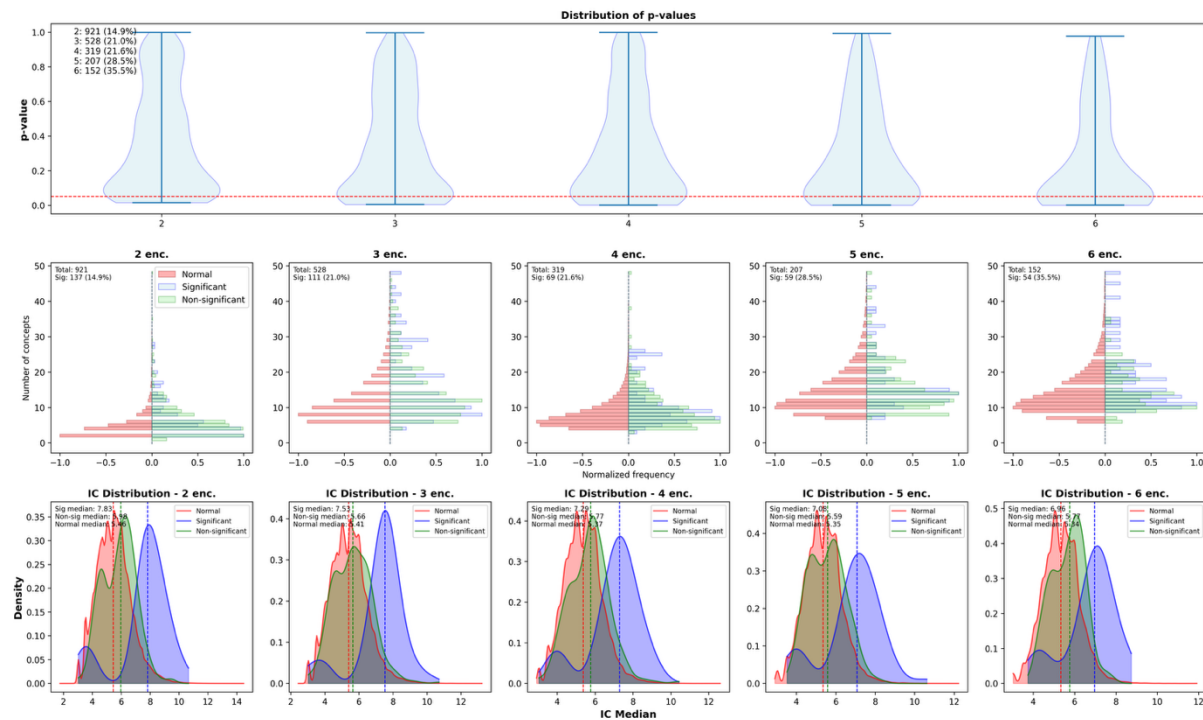


Figure S18. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Adults across all disease groups. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

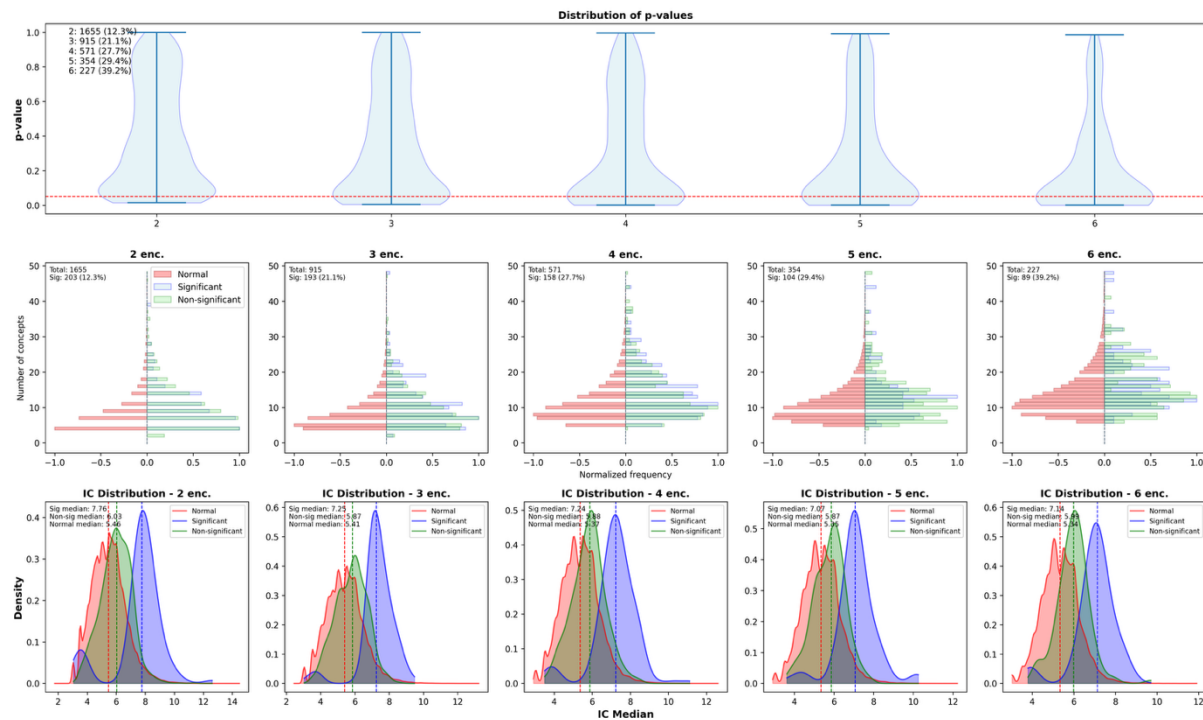


Figure S19. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Females across all disease groups. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

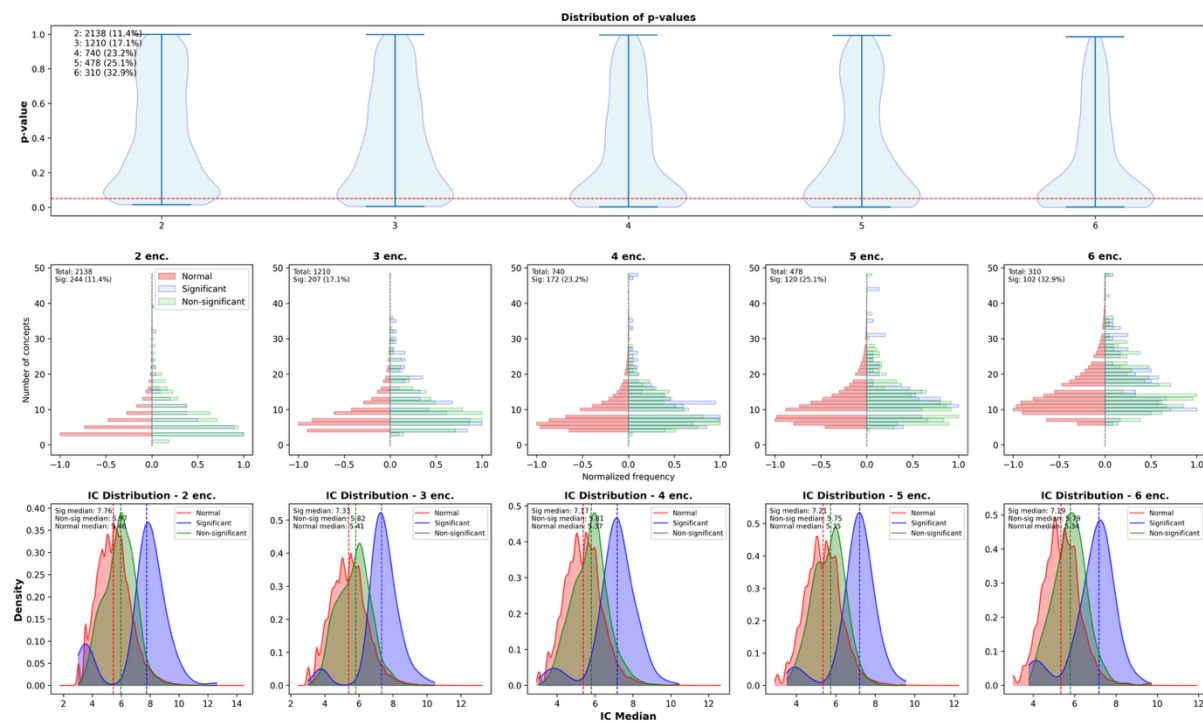


Figure S20. Fine-grained view over the distribution of the p-values obtained from performing a statistical significance test of the distribution of the median IC of a longitudinal profile against the control group at different numbers of encounters for Males across all disease groups. The top panel displays the distribution of p-values for each stratified group (ranging from 2 to 6 encounters), indicating the statistical significance of differences observed in the dataset. The middle panel provides histograms showing the normalized frequency of medical concepts across different stratification groups, with red bars representing the control population, blue the significant cases and green the non-significant cases. The bottom panel presents the distribution of IC values for each stratification group, comparing significant, non-significant, and normal cases.

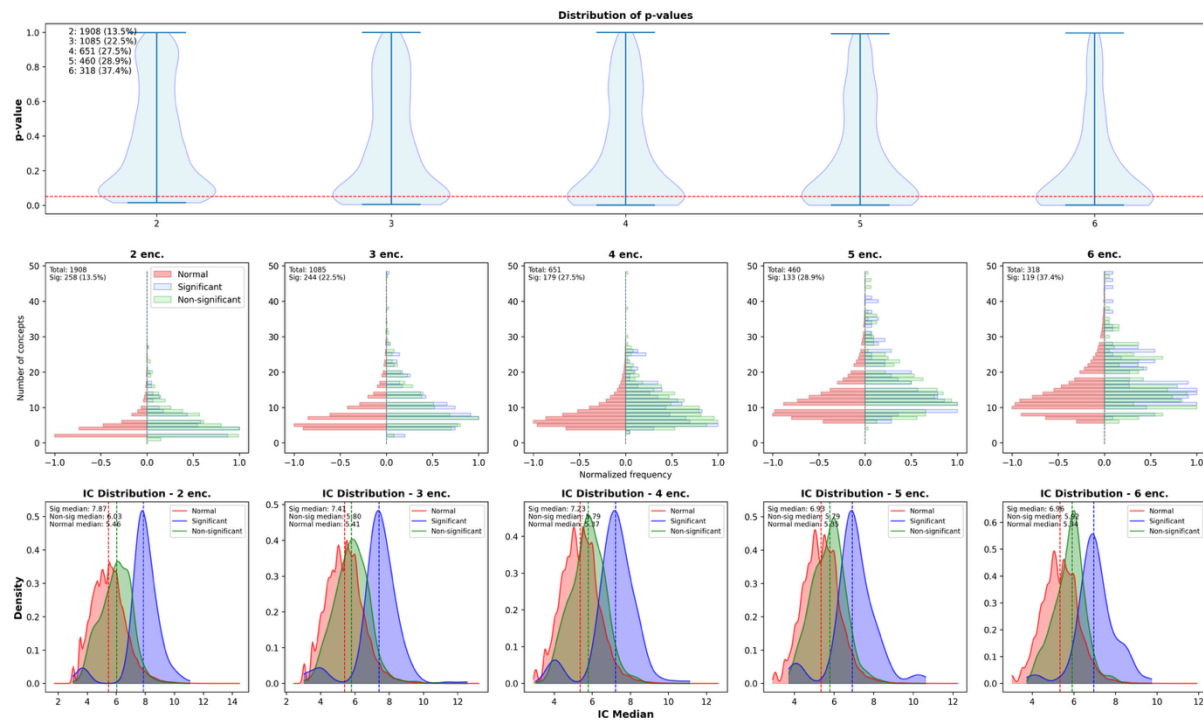


Figure S21. Receiver operating characteristic (ROC) curves showcasing the diagnostic performance of information content (IC) as a potential screening test for rare disease patients. Each panel focuses on a cohort stratified by the number of encounters at the time of diagnosis. Values are provided overall, as well as based on the major class of conditions: Genetic, Neoplasms and Other RDs.

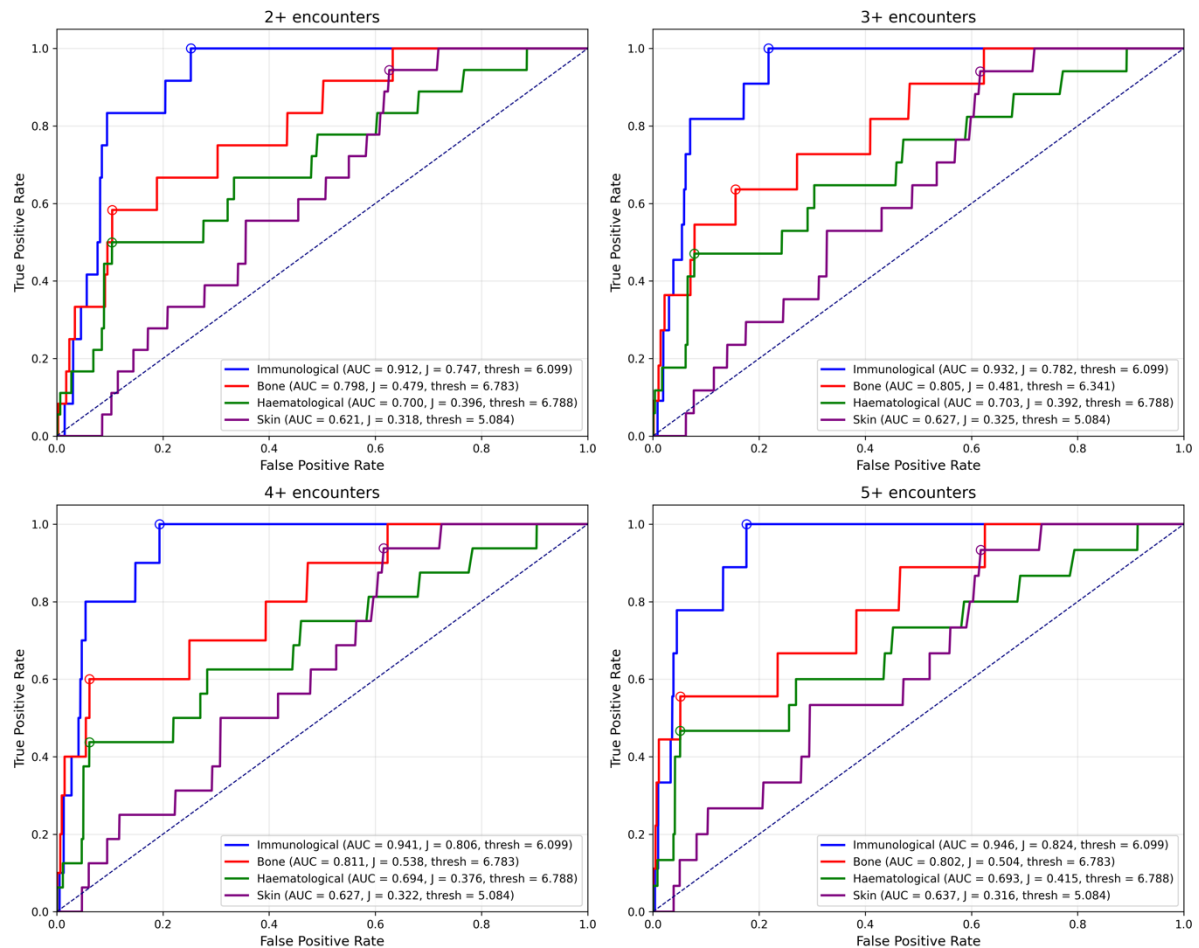


Figure S22. Receiver operating characteristic (ROC) curves showcasing the diagnostic performance of information content (IC) as a potential screening test for rare disease patients. Each panel focuses on a cohort stratified by the number of encounters at the time of diagnosis. Values are provided overall, as well as based on the major class of conditions: Genetic, Neoplasms and Other RDs.

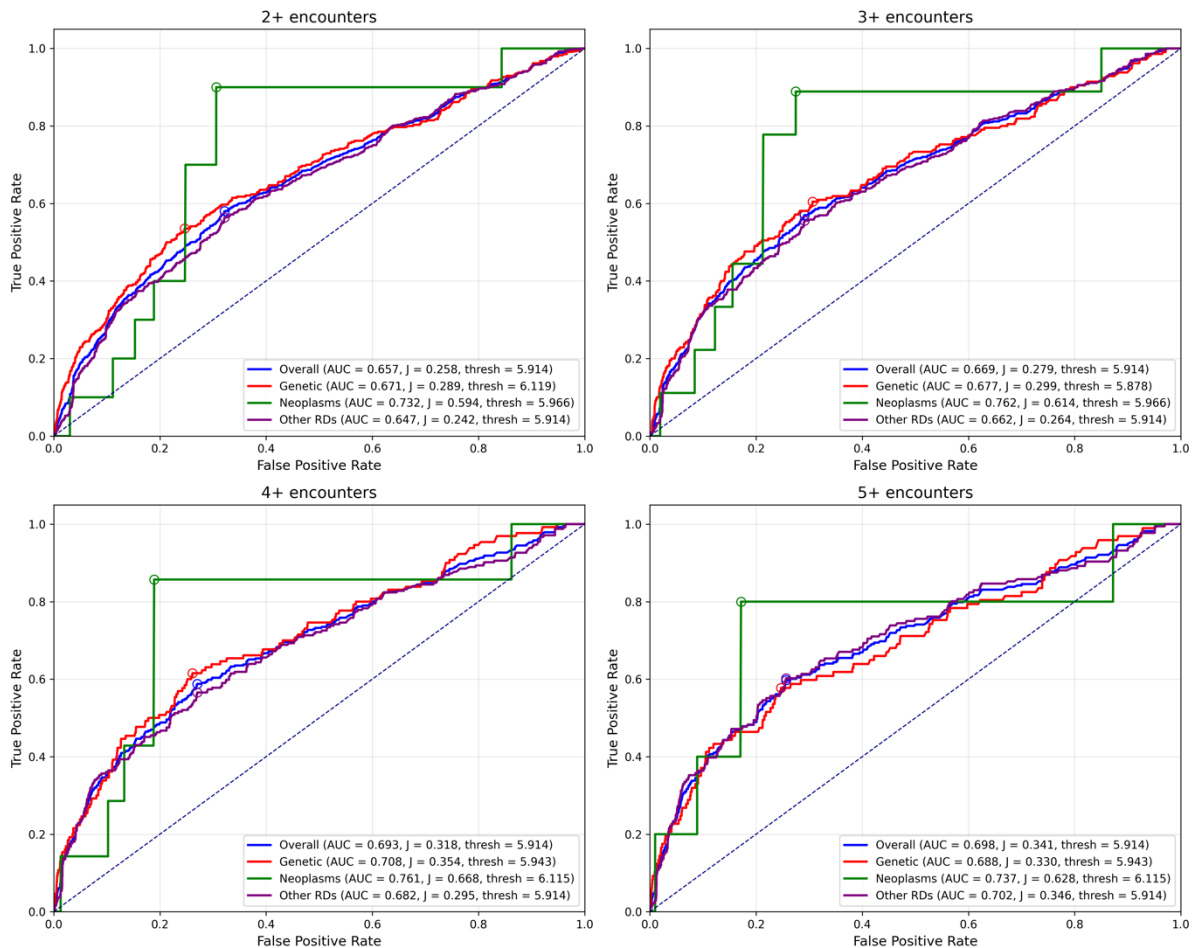
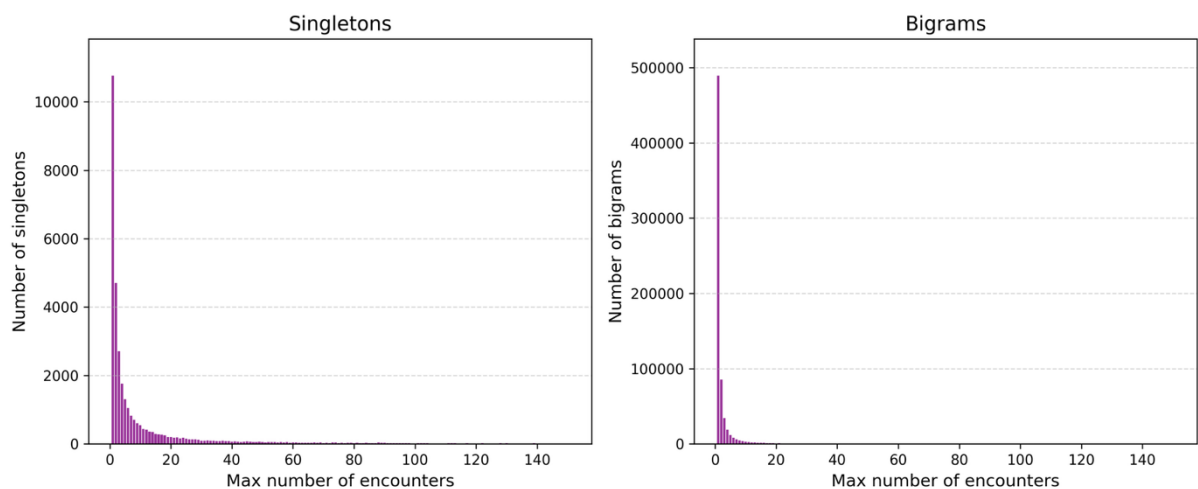


Figure S23. Singletons and bigrams term histograms across all encounters



Supplementary note 1: Deep dive into the first 2 encounters

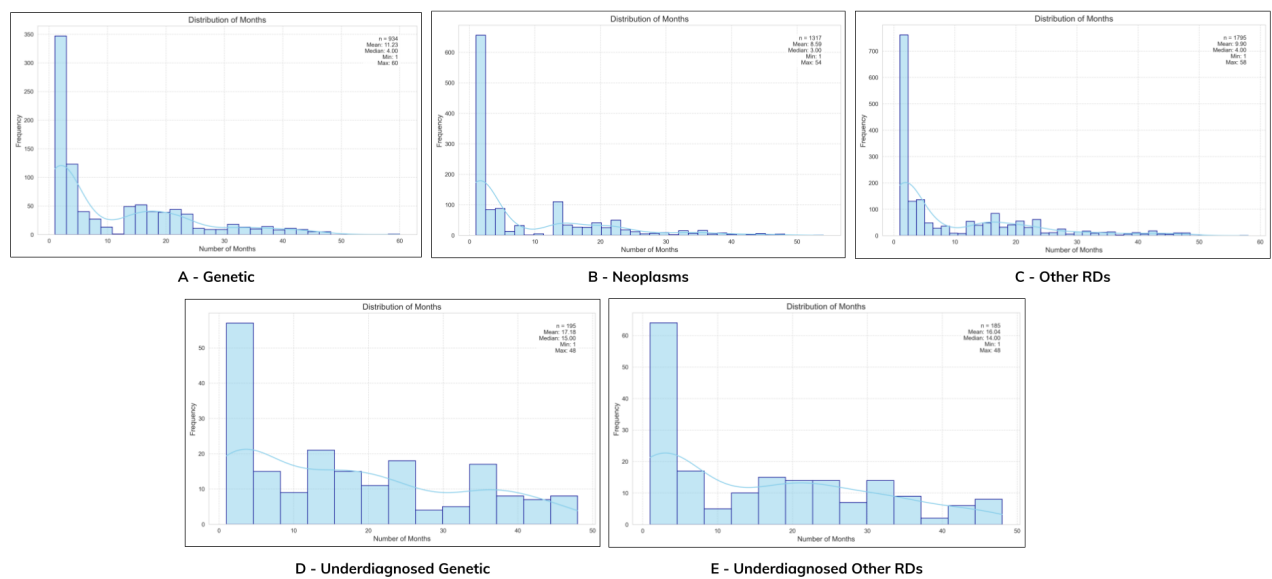


Figure 1. Distribution of months between the first and the second encounter for patients with a diagnosis present in the second encounter. A - Genetic diseases; B - Neoplasms; C - Other RDs; D - Underdiagnosed Genetic; E - Underdiagnosed Other RDs. The number of patients with underdiagnosed neoplasms was insignificant.

The individual-level analysis revealed a large number of patients who had their rare disease diagnosis recorded in the second encounter in our dataset. We therefore examined this subset more closely, specifically looking at the distribution of months between the first and second encounter (as shown in Fig. 1). The interval between the first two encounters varies notably across the three major disease classes. Patients with neoplasms (Fig. 1A) have the shortest typical gap between first and second visits – their mean and median intervals are the lowest, indicating that most cancer patients return for follow-up very quickly. Genetic rare disease patients (Fig. 1B) show intermediate intervals (with a moderate mean and median), while Other RDs (Fig. 1C) have, on average, the longest gap between the first and second visits.

These patterns likely stem from the nature and urgency of each condition. Neoplasms often require urgent evaluation and treatment, so follow-up tends to be scheduled very soon after the initial visit (hence the short intervals observed). Genetic disorders typically involve diagnostic testing and specialist consultations that, while important, may not be as immediately time-sensitive as cancer treatment. This leads to moderately timed follow-ups and more variation in interval length (for example, waiting for genetic test results might delay the second encounter). Other RDs encompass a diverse array of conditions, some of which progress slowly or have less clearly defined care pathways. Consequently, follow-up timing in this group is more irregular: a portion of patients return quickly if symptoms worsen or if a management plan is in place, whereas others experience long delays before the next encounter, possibly due to milder disease progression, limited treatment options, or challenges in accessing specialized care. This results in the Other RDs group having the highest average interval and the most dispersed (skewed) distribution of follow-up times.

The time between the first and second encounters is much longer on average for underdiagnosed genetic disease patients (Fig. 1D) compared to the overall genetic disease

cohort. In the entire genetic cohort, the distribution is heavily skewed toward short intervals – the median gap is only about 4 months, and many patients return for a second visit within a few months. In contrast, under-diagnosed genetic cases show a distribution shifted far to the right, with a median around 15 months (over a year). The mean interval is also higher for underdiagnosed patients (~17 months, versus ~11 months in the full genetic cohort), indicating that lengthy gaps are common rather than rare outliers in this group. In fact, whereas most patients in the overall genetic cohort had their second encounter within a few months, a typical underdiagnosed genetic patient did not return for over a year after the first visit. This suggests significant initial delays for the underdiagnosed group.

A similar pattern emerges for rare diseases that are not primarily genetic (e.g. certain rare autoimmune, metabolic, or other conditions) – the Other RDs group. The overall cohort of Other RD patients (Fig. 1C) shows a short typical interval between the first two visits: the median is about 4 months (identical to the genetic cohort's median), and the mean gap is around 10 months (indicating a long-tail distribution where some patients have much longer gaps). However, in the under-diagnosed subset of these other rare diseases (Fig. 1E), we again see a pronounced shift toward longer intervals. The median time to the second encounter is roughly 14 months (over a year), very close to that of the underdiagnosed genetic group, and the average (mean) is about 16 months (compared to ~9–10 months for the full cohort). This tells us that underdiagnosed non-genetic rare disease patients also experience substantially delayed second encounters relative to their peers. In practical terms, a typical patient with an underdiagnosed rare condition in this category waited more than a year for a follow-up, whereas most patients in the overall cohort were seen again within a few months.