

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11		
250908				Rare oncologic disease												
98057					Rare tumor											
3399				Germ cell tumor												
363579									Extragenadal germ cell tumor							
99913				Extragenadal non-dysgerminomatous germ cell tumor												
876	273300		404081005						Yolk sac tumor							
883		D44.5-C56-C62	55818009	Teratoma												
141077		D10.5	31248004							Epignathus						
141107		D10.6								Nasopharyngeal teratoma						
252018										Teratoma of the central nervous system						
99926		C58		Gestational choriocarcinoma												
180226			28047004						Embryonal carcinoma							
48736										Intracranial embryonal carcinoma						
289362										Extracranial embryonal carcinoma						
180229			28325008	Polyembryoma												
314613									Growing teratoma syndrome							
180234			32844007	Mixed germ cell tumor												
182127			28307001						Extragenadal germinoma							
91352				Germinoma of the central nervous system												
99912		C56								Malignant dysgerminomatous germ cell tumor						
363582				Gonadal germ cell tumor												
363504	273300	C62							Testicular germ cell tumor							
842	273300	C62		Testicular seminomatous germ cell tumor												
363494	273300	C62		Testicular non seminomatous germ cell tumor												
35807	603737	C56		Ovarian germ cell malignant tumor												
99912		C56							Malignant dysgerminomatous germ cell tumor							
206538		C56		Malignant nondysgerminomatous germ cell tumor												
289356		C56		Non-gestational ovarian choriocarcinoma												
68347				Tumor of hematopoietic and lymphoid tissues												
125	210900	Q99.8 Q87.1	4434006					Bloom syndrome								
3226	614038	Q82.0		Deafness - lymphedema - leukemia												
98292	154800	Q82.2-C96.2	04-414653009					Mastocytosis								
2467		C96.2	397016004	Systemic mastocytosis												
98848		D47.0	03-397356009						Indolent systemic mastocytosis							
158775				Smouldering systemic mastocytosis												
158778				Isolated bone marrow mastocytosis												
98849		C96.2 00-397357000		Systemic mastocytosis with an associated clonal hematologic non-mast cell lineage disease												
98850		C96.2 08-397358005		Aggressive systemic mastocytosis												
158793				Lymphadenopathic mastocytosis with eosinophilia												
98851		C94.3	02-128924002						Mast cell leukemia							
158796				Classic mast cell leukemia												
158799				Aleukemic mast cell leukemia												
66646		Q82.2	397012002	Cutaneous mastocytosis												
79455		Q82.2		Cutaneous mastocytoma												
79456		Q82.2		Diffuse cutaneous mastocytosis												
280785		Q82.2		Bullous diffuse cutaneous mastocytosis												
280794		Q82.2		Pseudoxanthomatous diffuse cutaneous mastocytosis												
79457	154800	Q82.2	78745000						Maculopapular cutaneous mastocytosis							
90389		Q82.2	8214000	Telangiectasia macularis eruptiva perstans												
158766				Typical urticaria pigmentosa												
158769				Plaque-form urticaria pigmentosa												
158772				Nodular urticaria pigmentosa												
66661		C96.2 02-188660004		Mast cell sarcoma												
66662		C96.2	63175003						Extracutaneous mastocytoma							

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171895				Myeloid hemopathy											
519	52270-602439	C92.0	009-91861009						Acute myeloid leukemia						
86845		C92.0	128827005						Acute myeloid leukemia with multilineage dysplasia						
86846		C92.0	128830003						Therapy related acute myeloid leukemia and myelodysplastic syndrome						
102379									Acute myeloid leukemia and myelodysplastic syndromes related to alkylating agent						
102381		C92.0							Acute myeloid leukemia and myelodysplastic syndromes related to topoisomerase type 2 inhibitor						
164726									Acute myeloid leukemia and myelodysplastic syndromes related to radiation						
86851		C95.0	07-128818009						Acute leukemia of ambiguous lineage						
98835		C95.0							Acute undifferentiated leukemia						
98836		C95.0	397346005						Bilineal acute leukemia						
98837		C95.0	09-278453007						Acute biphenotypic leukemia						
98797		Q98.6							Acute myeloid	Acute promyelocytic leukemia					
98829		C92.7							Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)						
98831		C92.6	128829008						Acute myeloid leukemia with 11q23 abnormalities						
102724		C92.7							Acute myeloid leukemia with t(8;21)(q22;q22) translocation						
319480		C92.0							Acute myeloid leukemia with CEBPA somatic mutations						
168231									Unclassified acute myelomonocytic leukemia						
514		C92.7	09-413441006						Acute monoblastic leukemia						
318	133180	C94.0							Acute erythroid leukemia						
518		C94.2							Acute megakaryoblastic leukemia						
99887		C94.2							Acute megakaryoblastic leukemia in Down syndrome						
329469		C94.2							Acute megakaryoblastic leukemia without Down syndrome						
86843		C94.5	003-80570006						Acute panmyelosis with myelofibrosis						
86849		C94.7	69077002						Acute basophilic leukemia						
86850		C92.3	007-35287006						Myeloid sarcoma						
98832		C92.0							Minimally differentiated acute myeloblastic leukemia						
98833		C92.0							Acute myeloblastic leukemia without maturation						
98834		C92.0							Acute myeloblastic leukemia with maturation						
319465		C92.0							Inherited acute myeloid leukemia						
52688	614286	D46	06-188736006						Myelodysplastic syndromes						
75564		D64.3							Acquired idiopathic sideroblastic anemia						
185662									Refractory cyt						
98827		D46.7							Refractory anemia						
101096		D46.7	89112009						Unclassified myelodysplastic syndrome						
186858									Aregenerative anemia						
100020		D46.2	01-415284008						Refractory ane						
86841	153550	D46.7							Refractory anemia with excess blasts-1						
168960		D46.3	05-128848007						Refractory anemia with excess blasts-2						
98274									Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality						
3318	01977-614521	D47.3	09-109994006						Refractory anemia with excess blasts in transformation						
521	608232	C92.1	005-92818009						Myeloproliferative neoplasm						
824	254450	D47.4							Essential thrombocythemia						
729	263300	D45	05-128841001						Chronic myeloid leukemia						
86829		C92.1	07-188734009						Myelofibrosis with myeloid metaplasia						
86830		D47.1							Polycythemia vera						
168940		D47.5	03-413836008						Chronic neutrophilic leukemia						
172216									Unclassified chronic myeloproliferative disease						
314950		D47.5							Chronic eosinophilic leukemia						
629932									Hyper eosinop						
98275									Idiopathic hypereosinophilic syndrome						
86834	607785	C93.3	01-445227008						Primary hypereosinophilic syndrome						
98823		C93.1	06-128831004						Secondary hyp Lymphoid hypereosinophilic syndrome						
98824		C92.2	06-277589003						Myelodysplastic/myeloproliferative disease						
									Juvenile myelomonocytic leukemia						
									Chronic myelomonocytic leukemia						
									Atypical chronic myeloid leukemia						

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98825									Unclassified myelodysplastic/myeloproliferative disease					
168943									Myeloid neoplasms associated with eosinophilia and abnormality of PDGFRA, PDGFRB or FGFR1					
168947									Myeloid neoplasm associated with PDGFRA rearrangement					
168950									Myeloid neoplasm associated with PDGFRB rearrangement					
168953	613523								Myeloid neoplasm associated with FGFR1 rearrangement					
171898									Lymphoid hemopathy					
3261	03909-614470	D72.8							Autoimmune lymphoproliferative syndrome					
98282									Plasma cell tumor					
2905		D47.7	001-79268002						POEMS syndrome					
29073	254500	C90.0	006-55921005						Multiple myeloma					
400144									AL amyloidosis					
314709		E85.4							Primary systemic amyloidosis					
186876									Primary localized amyloidosis					
100022		C90.2							Plasmacytoma					
180417									Primary plasmacytoma of the bone					
93557		D89.8							Extramedullary soft tissue plasmacytoma					
93558		D89.8	373604002						Non-amyloid					
186888									Heavy chain deposition disease					
100025		C88.3	109982002						Light and heavy chain deposition disease					
100026		C88.2	109984001						Light chain deposition disease					
98287									Heavy chain di					
185184									Mu heavy-chain disease					
389	604856	C96.5	C96.6	000-65399007					Alpha heavy-chain disease					
198159									Gamma heavy-chain disease					
99871		C96.6	129000002						Histiocytic and dendritic cell tumor					
99872		C96.5	404160007						Macrophage o					
99873		C96.5							Histiocytic sarcoma					
99874		C96.5							Langerhans cell histiocytosis					
86897		C96.4	128814006						Letterer-Siwe disease					
86900	267730	C96.4	07-373168002						Eosinophilic granuloma					
86902		C96.4	128816008						Hashimoto-Pritzker syndrome					
86903		C96.4	397355008						Hand-Schüller-Christian disease					
98290									Adult pulmonary Langerhans cell histiocytosis					
70568		D47.9	254290004						Langerhans cell sarcoma					
86904									Interdigitating dendritic cell sarcoma					
98291									Follicular dendritic cell sarcoma					
223735									Dendritic cell sarcoma not otherwise specified					
513	13067-247640	C91.0							Immunodeficiency-associated lymphoproliferative disease					
100407									Posttransplant lymphoproliferative disease					
99861		C91.0							Methotrexate-associated lymphoproliferative disorders					
171915									Lymphoproliferative disease associated with primary immune disease					
300842									Lymphoma					
545	613024	C82	000-55150002						Acute lymphoblastic leukemia					
33226	53600-610430	C88.0	004-35562000						Non-Hodgkin lymphoma					
58017		C91.4	001-54087003						Precursor B-cell acute lymphoblastic leukemia					
67038	12558-612559	C91.1	000-92814006						Precursor T-cell acute lymphoblastic leukemia					
300878		C91.4							B-cell non-Hodgkin lymphoma					
300912									Indolent B-cell non-Hodgkin lymphoma					
52417		C88.4							Follicular lymphoma					
86854		C83.0							Waldenström macroglobulinemia					
86867		C83.0	397349003						Hairy cell leukemia					
300869		C83.0							B-cell chronic lymphocytic leukemia					
300846									Hairy cell leukemia variant					
									Marginal zone lymphoma					
									MALT lymphoma					
									Splenic marginal zone lymphoma					
									Nodal marginal zone B-cell lymphoma					
									Splenic diffuse red pulp lymphoma					
									Aggressive B-cell non-Hodgkin lymphoma					

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543	113970	C83.7	006-77381001											Burkitt lymphoma
544		C83.3	005-46732000											Diffuse large B-cell lymphoma
86869		C83.8	239940004											Lymphomatoid granulomatosis
98838		C83.3	05-444910004											Primary mediastinal large B-cell lymphoma
98839		C83.3	399648005											Intravascular large B-cell lymphoma
364043		C83.3												ALK-positive large B-cell lymphoma
289661		C83.3												Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly
300849		C83.3												Diffuse large B-cell lymphoma of the central nervous system
300857		C83.3												T-cell/histiocyte rich large B cell lymphoma
300888		C83.3												Diffuse large B-cell lymphoma with chronic inflammation
48686		C83.8	128800006											Primary effusion lymphoma
52416		C83.1	006-74654000											Mantle cell lymphoma
86852		C91.3	07-277619001											B-cell prolymphocytic leukemia
99860	615545	C91.0												Precursor B-cell acute lymphoblastic leukemia
289666		C83.3												Plasmablastic lymphoma
171918														T-cell non-Hodgkin lymphoma
70568		D47.9	254290004											Posttransplant lymphoproliferative disease
86870		C86.4												CD4+/CD56+ hematodermic neoplasm
86871		C91.3	06-277567002											T-cell prolymphocytic leukemia
86872		C91.5	001-55081009											T-cell large granular lymphocyte leukemia
86873		C91.7	128833001											Aggressive NK-cell leukemia
86875		C91.5	005-77430005											Adult T-cell leukemia/lymphoma
86879		C84.4	414166008											Extranodal nasal NK/T cell lymphoma
86880		C84.4	08-277654008											Enteropathy-associated T-cell lymphoma
86882		C84.4	445406001											Hepatosplenic T-cell lymphoma
98841		C83.3	277637000											Anaplastic large cell lymphoma
300895		C83.3												ALK-positive anaplastic large cell lymphoma
300903		C83.3												ALK-negative anaplastic large cell lymphoma
86886		C84.4	37009-835009											Angioimmunoblastic T-cell lymphoma
364033														Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood
364039														Hydroa vacciniforme-like lymphoma
391	00021-236000	C81												Hodgkin lymphoma, classical
197138														Hodgkin lymphoma Lymphocyte-rich classical Hodgkin lymphoma
98846		C81.3												Lymphocyte-depleted classical Hodgkin lymphoma
98843		C81.1												Nodular sclerosis classical Hodgkin lymphoma
98844		C81.2												Mixed cellularity classical Hodgkin lymphoma
86893		C81.1	004-70600005											Nodular lymphocyte predominant Hodgkin lymphoma
168966			128798004											Composite lymphoma
282331														Primary organ Primary pulmonary lymphoma
542														Primary cutaneous lymphoma
171901														Primary cutaneous T-cell lymphoma
178548														Indolent primary cutaneous T-cell lymphoma
541		C86.6	00-397352006											Primary cutaneous CD30+ T-cell lymphoproliferative disease
98842		C86.6	03-397353001											Lymphomatoid papulosis
300865		C86.6												Primary cutaneous anaplastic large cell lymphoma
86884		C84.4	05-404133000											Subcutaneous panniculitis-like T-cell lymphoma
178522		C84.4												Primary cutaneous CD4+ small/medium-sized pleomorphic T-cell lymphoma
178566														Mycosis fungoides and variants
2584	254400	C84.0												Classical mycosis fungoides
33111		C84.0	277796003											Granulomatous slack skin
178512		C84.0												Folliculotropic mycosis fungoides
178517		C84.0	404120006											Localized pagetoid reticulosis
178551														Aggressive primary cutaneous T-cell lymphoma

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178540		C82.6								Primary cutaneous follicle center lymphoma				
2591	28550-615293	D21	254146000							Infantile myofibromatosis				
2415		D18.1	234095009							Lymphatic malformation				
79489		D18.1								Macrocystic lymphatic malformation				
141209		D18.1								Diffuse lymphatic malformation				
217410		D18.1								Circumscribed lymphatic malformation				
79490		D18.1								Microcystic lymphatic malformation				
217409										Cervicofacial ly Cystic hygroma				
168984	613089	Q87.3								CLAPO syndrome				
618	15134-155700	C43								Familial melanoma				
492		L72.1								Proliferating trichilemmal cyst				
840		D23.9	239121009							Syringocystadenoma papilliferum				
864		274899008-33059009								Trichofolliculoma				
122	135150		110985001							Birt-Hogg-Dube syndrome				
338										Familial multiple fibrofolliculoma				
493		L85.8								Familial keratoacanthoma				
523	150800	D23								Familial leiomyomatosis				
31112	607907	9004-238863004-128742004								Dermatofibrosarcoma protuberans				
53715	11900-610455	M11.2	61778004							Tumoral calcinosis				
306658	610455	M11.2								Normocalcemic tumoral calcinosis				
306661	211900	M11.2								Hypercalcemic tumoral calcinosis				
65748	132800	D23								Multiple keratoacanthoma, Ferguson-Smith type				
79140		C44								Cutaneous neuroendocrine carcinoma				
79493	01606-605041									Brooke-Spiegler syndrome				
211	132700		211710004							Familial cylindromatosis				
867	01606-612099	D23.3	403825008							Familial multiple trichoepithelioma				
79665	175100	D12.6	60876000							Gardner syndrome				
91414	132600	D23	004-44155009							Pilomatrixoma				
168632	605827	Q82.5								Generalized basaloid follicular hamartoma syndrome				
168999										Malignant melanoma of the mucosa				
199257										Superficial fibromatosis				
79142	126900	M72.0								Familial Dupuytren contracture				
199251		M72.2	13370002							Ledderhose disease				
199260		M72.8								Calcified aponeurotic fibroma				
199267		M72.8	399903008							Infantile digital fibromatosis				
263435										Congenital smooth muscle hamartoma				
276280		Q87.3								Hemihyperplasia-multiple lipomatosis syndrome				
294057										Rare nevus				
626	137550	D22								Large congenital melanocytic nevus				
2612	163200	Q85.8								Linear nevus sebaceus syndrome				
2611		Q82.5								Linear verrucous nevus syndrome				
79466		Q82.5	399995006							Inflammatory linear verrucous epidermal nevus				
79467		Q82.5								Verrucous nevus				
79468		Q82.5								Acanthokeratolytic verrucous nevus				
35125		Q85.8	007-25201003							Epidermal nevus syndrome				
139414										Congenital panfollicular nevus				
166286		Q82.5	239118007							Porokeratotic eccrine ostial and dermal duct nevus				
171723	193900	Q38.6	389203001							White sponge nevus				
263425		D22.3								Nevus of Ota				
263432		D22.6								Nevus of Ito				
313936										PENS syndrome				
300515										Rare nail tumor				
300504										Onychocytic matricoma				

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329977									Classic endocrine tumor of the appendix					
329984									Goblet cell carcinoma					
100080								Colon endocrine tumor						
100081								Rectal endocrine tumor						
100082								Anal endocrine tumor						
100093	114900	E34.0						Carcinoid tumor and carcinoid syndrome						
104010								Intestinal polyposis						
80398								Familial adenc Gardner syndrome						
99818	175100	D12.6						Turcot syndrome with polyposis						
261584		D12.6						Familial adenomatous polyposis due to 5q22.2 microdeletion						
2869	175200	Q85.8	54411001					Peutz-Jeghers syndrome						
201 15108-615109		Q85.8	58037000					Cowden syndrome						
82005								Juvenile polyp Juvenile polyposis of infancy						
329971	174900	D12.6						Generalized juvenile polyposis/juvenile polyposis coli						
109	153480	Q87.8	005-46011003					Bannayan-Riley-Ruvalcaba syndrome						
2930	175500		76304001					Cronkhite-Canada syndrome						
157794	01228-610069	D12.6						Hereditary mixed polyposis syndrome						
157798	175020	D12.6						Hyperplastic polyposis syndrome						
160148		D12.6						Cap polyposis						
468258								Attenuated fa MUTYH-related attenuated familial adenomatous polyposis						
247806	175100	D12.6						APC-related attenuated familial adenomatous polyposis						
104075		D01.4						Small bowel adenocarcinoma						
104076		C17						Small bowel leiomyosarcoma						
180821								Gastro-esophageal tumor						
63443	613659	C16						Gastric cancer						
26106	137215	C16						Familial gastric cancer						
36273		C16.2						Gastric linitis plastica						
313920		C16						Epstein-Barr virus-associated gastric carcinoma						
70482	133239	C15	372138000					Esophageal carcinoma						
99976	614266	C15						Esophageal adenocarcinoma						
99977	133239	C15						Esophageal squamous cell carcinoma						
100075								Gastric endocrine tumor						
180824								Pancreatic tumor						
677		C25.1	53618008					Pancreatoblastoma						
51013	606719							Melanoma-pancreatic cancer syndrome						
97253		E16.8						Pancreatic endocrine tumor						
913		E16.4	004-53132006					Zollinger-Ellison syndrome						
97261		E16.8						GRFoma						
97278		E16.8						PPoma						
97279		E16.8 08-302822000						Insulinoma						
97280		E16.8 00-302823005						Glucagonoma						
97282		E16.8						VIPoma						
97283		E16.8 05-253006001						Somatostatinoma						
218407								Pancreatic carcinoma	Familial pancreatic carcinoma					
276142								Salivary glands tumor						
276145								Malignant epithelial tumor of the salivary glands						
276148								Benign epithelial tumor of the salivary glands						
314022								Gastric adenocarcinoma and proximal polyposis of the stomach						
98060								Rare respiratory tumor						
150 61550-607107		C11						Nasopharyngeal carcinoma						
101945								Rare bronchopulmonary tumor						
2420		C85.9						Primary pulmonary lymphoma						
50251	156240	C45	62064005					Mesothelioma						

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64741		C34							Pulmonary blastoma					
64742	601200	C34							Pleuropulmonary blastoma					
99933		C34							Pleuropulmonary blastoma type 1					
99934		C34							Pleuropulmonary blastoma type 2					
99935		C34							Pleuropulmonary blastoma type 3					
284343	601200								Pleuropulmonary blastoma family tumor susceptibility syndrome					
284362									Fetal lung interstitial tumor					
70573	182280	C34							Small cell lung cancer					
97287		D38.1							Bronchial endocrine tumor					
284395		C34							Well-differentiated fetal adenocarcinoma of the lung					
98061									Rare otorhinolaryngologic tumor					
150	61550-607107	C11							Nasopharyngeal carcinoma					
100083									Laryngeal endocrine tumor					
100084									Middle ear endocrine tumor					
141112									Nasal glial heterotopia					
141115									Nasal ganglioglioma					
98062									Rare nervous system tumor					
46135									Primary central nervous system lymphoma					
54595		D44.4 009-4009002							Craniopharyngioma					
251558									Tumor of the neuroepithelial tissue					
182067									Glial tumor					
251633									Low grade ependymoma					
251937									Ependymal tumor					
251639									Ependymoma					
251643									Subependymoma					
251646									Myxopapillary ependymoma					
251561									Anaplastic ependymoma					
360	37800-613029	C71.8 007-63634009							High-grade astrocytoma					
251670									Glioblastoma					
251579									Astrocytoma					
251582									Gliosarcoma					
251589									Giant cell glioblastoma					
251592									Gliomatosis cerebri					
251595									Anaplastic astrocytoma					
251598									Low-grade astrocytoma					
251601									Diffuse astrocytoma					
251604									Protoplasmic astrocytoma					
251607									Fibrillary astrocytoma					
503227									Gemistocytic astrocytoma					
251618									Pleomorphic xanthoastrocytoma					
251623									Pilocytic astrocytoma					
298111									Piloxyoid astrocytoma					
251630									Subependymal giant cell astrocytoma					
503307									Pituicytoma					
251663									Oligodendroglioma					
503339									Anaplastic oligodendroglioma					
251674									Oligoastrocytoma					
251679									Anaplastic oligoastrocytoma					
251852									Glioma					
2677	612219	253096008-55045006							Angiocentric glioma					
252471									Chordoid glioma					
251858	155255								Astroblastoma					
251863	155255								Embryonal tumor of the neuroepithelial tissue					
									Neuroepithelioma					
									Medulloblastoma					
									Anaplastic/large cell medulloblastoma					
									Medulloblastoma with extensive nodularity					
									Desmoplastic/nodular medulloblastoma					

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11
251867	155255									Classic medulloblastoma				
252505										Primitive neur Neuroblastoma				
1957			76060004							Esthesioneuroblastoma				
251877										Ganglioneuroblastoma				
251880										Ependymoblastoma				
251883										Medulloepithelioma				
251891										Atypical teratoid/rhabdoid tumor				
251896										Choroid plexus tumor				
2807	260500	D33.0	18021007							Papilloma of choroid plexus				
251899	260500									Choroid plexus carcinoma				
251902										Atypical papilloma of choroid plexus				
251905										Pineal tumor of the neuroepithelial tissue				
251909										Pineoblastoma				
251912										Pineocytoma				
251915										Papillary tumour of the pineal region				
251919										Pineal parenchymal tumor of intermediate differentiation				
251924										Neuronal tumor				
73256			128858006							Central neurocytoma				
251927										Extraventricular neurocytoma				
251931										Cerebellar liponeurocytoma				
251934										Mixed neuronal-gliar tumor				
65285	158350	Q04.8	128791005-							Lhermitte-Duclos disease				
251937										Gangliocytoma				
251940										Desmoplastic infantile astrocytoma/ganglioglioma				
251946										Dysembryoplastic neuroepithelial tumor				
393064										Ganglioglioma Nasal ganglioglioma				
251957										Anaplastic ganglioglioma				
251962										Papillary glioneuronal tumor				
251975										Rosette-forming glioneuronal tumour of the fourth ventricle				
251992										Ganglioneuroma				
251995										Primary germ cell tumor of the central nervous system				
48736										Intracranial embryonal carcinoma				
91352										Germinoma of the central nervous system				
252006										Yolk sac tumor of the central nervous system				
252015										Choriocarcinoma of the central nervous system				
252018										Teratoma of the central nervous system				
252021										Mixed germ cell tumor of the central nervous system				
252025										Tumor of the meninges				
2495	606190	D32.9	19453003							Meningioma				
252028										Primary melanocytic tumor of the central nervous system				
252031										Diffuse leptomeningeal melanocytosis				
252046										Meningeal melanocytoma				
252050										Primary melanoma of the central nervous system				
263662	607174									Familial multiple meningioma				
252054										Hemangioblastoma				
252057										Tumor of cranial and spinal nerves				
2086		C72.3								Optic pathway glioma				
255360										Malignant peri Malignant triton tumor				
85102		C47.9	01-404036006							Perineurioma				
200002										Extraneural pe Reticular perineurioma				
100001										Sclerosing perineurioma				
100003										Intraneural perineurioma				
252128										Malignant perineurioma				

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11
252131				Benign peripheral nerve sheath tumor										
504339									Benign schwannoma	Vestibular schwannoma				
252183				Neurofibroma										
300501				Painful orbital and systemic neurofibromas-marfanoid habitus syndrome										
279897				Primary oculocerebral lymphoma										
98063						Rare gynecological tumor								
2800	167300	C44 001-71447003		Extramammary Paget disease										
180220						Rare uterine adnexal tumor								
97293		D27		Rare benign ovarian tumor										
206470		D27							Serous or mucinous cystadenoma of childhood					
314451		D27		Meigs syndrome										
314459		D27							Pseudo-Meigs syndrome					
314466		D27		Atypical Meigs syndrome										
314473		D27		Ovarian fibroma										
314478		D27		Ovarian fibrothecoma										
180237		D28.2							Benign tumor of fallopian tubes					
180242		C57.0	363444001	Malignant tumor of fallopian tubes										
213500									Rare ovarian cancer					
135719				Ovarian germ cell tumor										
206538										Malignant dysgerminomatous germ cell tumor				
289356		C56		Malignant nondysgerminomatous germ cell tumor										
135722				Non-gestational ovarian choriocarcinoma										
99915		C56	18861007	Ovarian malignant germ cell tumor										
99916		C56		Granulosa cell malignant tumor										
99917		C56		Ovarian malignant Sertoli-Leydig cell tumor										
206473		C56		Theca steroid-producing cell malignant tumor, not further specified										
206484	424500	C56		Borderline ovarian epithelial tumor										
213504		C56		Ovarian gonadoblastoma										
213512		C56		Ovarian adenocarcinoma										
14414350-614385		C18	315058005	Malignant mixed epithelial mesenchymal tumor of the ovary										
214104				Hereditary nonpolyposis colon cancer										
99817	276300	C18		Familial ovarian cancer										
14514291-604370		C50 C56		Muir-Torre syndrome										
213524		C56		Non-polyposis Turcot syndrome										
180250				Hereditary breast and ovarian cancer syndrome										
180253				Hereditary site-specific ovarian cancer syndrome										
50920	615554	D24		Rare breast tumor										
180261		D24		Rare benign breast tumor										
180267		D24		Mammary polyadenomatosis										
180257				Phyllode tumor										
14514291-604370		C50 C56		Giant adenofibroma of the breast										
180275		C50.0	05-403946000	Rare malignant breast tumor										
213528		C50		Hereditary breast and ovarian cancer syndrome										
213531		C50		Paget disease of the nipple										
213557		C50		Rare adenocarcinoma of the breast										
227535	05365-604370	C50		Metaplastic carcinoma of the breast										
180284				Salivary gland type cancer of the breast										
180312				Hereditary breast cancer										
137583				Benign ductal tumor of breast										
180247		C52	254893005	Rare vulvovaginal tumor										
206489		C52		Vulvar intraepithelial neoplasia										
206492		C52		Vaginal carcinoma										
213564				Vaginal germ cell malignant tumor										
				Vulvovaginal rhabdomyosarcoma										
				Rare uterine cancer										

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11	
213569									Rare cancer of the corpus uteri						
213574		C54							Rare adenocarcinoma of the corpus uteri						
427189									Malignant mixed Adenosarcoma of the corpus uteri						
213605		C54							Carcinofibroma of the corpus uteri						
213610		C54							Malignant mixed müllerian tumor of the corpus uteri						
427235									Sarcoma of the Rhabdomyosarcoma of the corpus uteri						
213625		C54							Leiomyosarcoma of the corpus uteri						
213630		C54							Malignant peripheral neuroectodermal tumor of the corpus uteri						
213711		C50							Endometrial stromal sarcoma						
213716		C50							Squamous cell carcinoma of the corpus uteri						
213721		C50							Undifferentiated carcinoma of the corpus uteri						
213726		C54							Papillary carcinoma of the corpus uteri						
213731		C54							Poorly differentiated endocrine carcinoma of the corpus uteri						
213736		C54							Well-differentiated endocrine neoplasm of the corpus uteri						
213741		C54							Adenoid cystic carcinoma of the corpus uteri						
213746		C54							Transitional cell carcinoma of the corpus uteri						
213751		C54							Malignant germ cell tumor of the corpus uteri						
213761									Rare cancer of the cervix uteri						
213767		C53							Rare squamous cell carcinoma of the cervix uteri						
213772		C53							Adenocarcinoma of the cervix uteri						
213777		C53							Poorly differentiated endocrine carcinoma of the cervix uteri						
213782									Malignant mixed epithelial and mesenchymal tumor of the cervix uteri						
213787		C53							Malignant müllerian mixed tumor of the cervix uteri						
213792		C53							Adenosarcoma of the cervix uteri						
213797									Sarcoma of the cervix uteri						
213802		C53							Rhabdomyosarcoma of the cervix uteri						
213807		C53							Leiomyosarcoma of the cervix uteri						
213812		C53							Malignant peripheral neuroectodermal tumor of the cervix uteri						
213817		C53							Papillary carcinoma of the cervix uteri						
213823		C53							Adenoid cystic carcinoma of the cervix uteri						
213828		C53							Adenoid basal carcinoma of the cervix uteri						
213833		C53							Glassy cell carcinoma of the cervix uteri						
213837		C53							Malignant germ cell tumor of the cervix uteri						
254685									Gestational trophoblastic disease						
59305		416441004-417475006							Gestational trophoblastic neoplasm						
99925		D39.2							Invasive mole						
99926		C58							Gestational choriocarcinoma						
99928		D39.2 008-75320001							Placental site trophoblastic tumor						
254698		D39.2							Epithelioid trophoblastic tumor						
99927 14293-231090 001.9 001.1 48430004									Hydatidiform mole						
254688 31090-614293		O01.0							Complete hydatidiform mole						
254693		O01.1							Partial hydatidiform mole						
100100									Thymic tumor						
3398		C37 D15.0 128621008							Thymic epithelial neoplasm						
99867	274230	C37 D15.0 05-444231005							Thymoma						
263310		C37 D15.0							Thymoma type A						
263317		C37 D15.0							Thymoma type B						
263324		C37 D15.0							Thymoma type AB						
99868		C37 06-444596001							Thymic carcinoma						
99869		C37							Thymic neuroendocrine carcinoma						
263331		C37							Well-differentiated thymic neuroendocrine carcinoma						
263335		C37							Moderately-differentiated thymic neuroendocrine carcinoma						
263339		C37							Poorly-differentiated thymic neuroendocrine carcinoma						

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11
100078									Ileal endocrine tumor					
100079									Endocrine tumor of the appendix					
329977									Classic endocrine tumor of the appendix					
329984									Goblet cell carcinoma					
100080									Colon endocrine tumor					
100081									Rectal endocrine tumor					
100082									Anal endocrine tumor					
100093	114900	E34.0							Carcinoid tumor and carcinoid syndrome					
100101									Endocrine tumor with other location					
79140		C44							Cutaneous neuroendocrine carcinoma					
97287		D38.1							Bronchial endocrine tumor					
100083									Laryngeal endocrine tumor					
100084									Middle ear endocrine tumor					
100085									Hepatic endocrine tumor					
100086		C23							Gall-bladder endocrine tumor					
53715	11900-610455	M11.2	61778004						Tumoral calcinosis					
306658	610455	M11.2							Normocalcemic tumoral calcinosis					
306661	211900	M11.2							Hypercalcemic tumoral calcinosis					
195186									Non-secreting Non-secreting chemodectoma					
100087									Thyroid tumor					
97285									Thyroid lymphoma					
100088									Thyroid carcinoma					
146	88550-607464	C73							Papillary or follicular thyroid carcinoma					
653	62300-171400	D44.8	61808009						Multiple endocrine neoplasia type 2					
99361	155240	C73							Familial medullary thyroid carcinoma					
247698	171400	D44.8							Multiple endocrine neoplasia type 2A					
247709	162300	D44.8							Multiple endocrine neoplasia type 2B					
142		C73	255031003						Anaplastic thyroid carcinoma					
1332		C73	255032005						Medullary thyroid carcinoma					
97290	605642	C73							Familial papillary thyroid carcinoma with renal papillary neoplasia					
319494									Familial nonmedullary thyroid carcinoma					
97290	605642	C73							Familial papillary thyroid carcinoma with renal papillary neoplasia					
319487	03744-606240	C73							Familial papillary or follicular thyroid carcinoma					
100090									Rare parathyroid tumor					
2207		E21.0							Familial primary hyperparathyroidism					
652	131100	D44.8	30664006						Multiple endocrine neoplasia type 1					
99877	145000	E21.0							Familial parathyroid adenoma					
99879	45000-610071	E21.0							Familial isolated hyperparathyroidism					
99880	145001	E21.0							Hyperparathyroidism - jaw tumor syndrome					
143	608266	C75.0	255037004						Parathyroid carcinoma					
100091									Adrenal/paraganglial tumor					
652	131100	D44.8	30664006						Multiple endocrine neoplasia type 1					
717									Catecholamine-producing tumor					
892	193300	Q85.8	46659004						Von Hippel-Lindau disease					
29072	01650-605373	D35.0 D35.6							Hereditary pheochromocytoma-paraganglioma					
247698	171400	D44.8							Multiple endocrine neoplasia type 2A					
247709	162300	D44.8							Multiple endocrine neoplasia type 2B					
276621									Sporadic pheochromocytoma/secretory paraganglioma					
276624									Sporadic pheochromocytoma					
276627									Sporadic secreting paraganglioma					
324299									Multiple paragangliomas associated with polycythemia					
1359	60980-605244	D44.8	239132009						Carney complex					
1501	202300	C74.0 E24.8	2227007						Adrenocortical carcinoma					

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463		D35.0	127021009												Adrenal incidentaloma
404	605635	E26.0													Familial hyperaldosteronism type 2
91351															Pituitary dermoid and epidermoid cysts
99888															Adrenocortical adenoma
100094															Multiple polyglandular tumor
892	193300	Q85.8	46659004												Von Hippel-Lindau disease
1359 60980-605244		D44.8	239132009												Carney complex
97286	606864	D44.8													Carney-Stratakis syndrome
139411	604287	D44.8													Carney triad
276152	610755	D44.8													Multiple endocrine neoplasia type 4
276161															Multiple endocrine neoplasia
652	131100	D44.8	30664006												Multiple endocrine neoplasia type 1
653 62300-171400		D44.8	61808009												Multiple endocrine neoplasia type 2
99361	155240	C73													Familial medullary thyroid carcinoma
247698	171400	D44.8													Multiple endocrine neoplasia type 2A
247709	162300	D44.8													Multiple endocrine neoplasia type 2B
304055															Pituitary tumor
99408															Pituitary adenoma
91349		D35.2													Non-functioning pituitary adenoma
314786		D35.2													Silent pituitary adenoma
314790		D35.2													Null pituitary adenoma
314753															Functioning pituitary adenoma
2965		D35.2 E22.1	002-34337008												Prolactinoma
91347		D35.2	254959007												TSH-secreting pituitary adenoma
91348		D35.2	254960002												Functioning gonadotroph adenoma
96253	219090	E24.0	190502001												Cushing disease
96256															Somatotroph adenoma
963	102200	E22.0	74107003												Acromegaly
99725	102200	E22.0	80849007												Gigantism
199244		E24.1	43019009												Nelson syndrome
629528															Mixed function Somatomammotropinoma
314777	00634-102200	D35.2													Familial isolated pituitary adenoma
300385		C75.1													Pituitary carcinoma
289635															Rare virus induced tumor
102024															HHV-8 related disorders
160	148000	D36.0	03-238809002												Castleman disease
93682		D36.0													Pediatric Castleman disease
93685		D36.0													Localized Castleman disease
93686	148000	D36.0													Multicentric Castleman disease
33276	148000	C46.9 C46.7	007-49937004												Kaposi's sarcoma
48686		C83.8	128800006												Primary effusion lymphoma
289638															Epstein-Barr Virus-related tumor
289644															Epstein-Barr virus-associated malignant lymphoproliferative disorder
364033															Systemic Epstein-Barr virus-positive T-cell lymphoproliferative disease of childhood
364039															Hydroa vacciniforme-like lymphoma
543	113970	C83.7	006-77381001												Burkitt lymphoma
46135															Primary central nervous system lymphoma
48686		C83.8	128800006												Primary effusion lymphoma
70568		D47.9	254290004												Posttransplant lymphoproliferative disease
86869		C83.8	239940004												Lymphomatoid granulomatosis
86879		C84.4	414166008												Extranodal nasal NK/T cell lymphoma
98293															Hodgkin lymphoma
391 00021-236000		C81													Hodgkin lymphoma, classical

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11
98845		C81.0	05-128799007											Lymphocyte-rich classical Hodgkin lymphoma
98846		C81.3												Lymphocyte-depleted classical Hodgkin lymphoma
98843		C81.1												Nodular sclerosis classical Hodgkin lymphoma
98844		C81.2												Mixed cellularity classical Hodgkin lymphoma
86893		C81.1	004-70600005											Nodular lymphocyte predominant Hodgkin lymphoma
289661		C83.3												Epstein-Barr virus-positive diffuse large B-cell lymphoma of the elderly
289666		C83.3												Plasmablastic lymphoma
300888		C83.3												Diffuse large B-cell lymphoma with chronic inflammation
289651														Epstein-Barr Virus-associated carcinoma
150 61550-607107		C11												Nasopharyngeal carcinoma
289682														Lymphoepithelial-like carcinoma
313920		C16												Epstein-Barr virus-associated gastric carcinoma
289656														Epstein-Barr Virus-associated mesenchymal tumor
64720		443719001-51549004												Leiomyosarcoma
86902		C96.4	128816008											Follicular dendritic cell sarcoma
289685														Myopericytoma
290849														Rare head and neck tumor
67037	275355													Squamous cell carcinoma of head and neck
289596		D10.6												Juvenile nasopharyngeal angiofibroma
314425														Rare odontologic tumor
314419														Ameloblastoma
314422														Ameloblastic carcinoma
140162														Inherited cancer-predisposing syndrome
357027		C69.2												Familial retinoblastoma
892	193300	Q85.8	46659004											Von Hippel-Lindau disease
733	175100	D12.6												Familial adenomatous polyposis
79665	175100	D12.6	60876000											Gardner syndrome
99818	175100	D12.6												Turcot syndrome with polyposis
261584		D12.6												Familial adenomatous polyposis due to 5q22.2 microdeletion
652	131100	D44.8	30664006											Multiple endocrine neoplasia type 1
637	101000	Q85.0	007-92503002											Neurofibromatosis type 2
636 62210-613675		Q85.0	92824003											Neurofibromatosis type 1
97685	613675	Q85.0												17q11 microdeletion syndrome
363700	162200	Q85.0												Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion
524 09265-609266		D48.9	428850001											Li-Fraumeni syndrome
377	109400	Q87.8	69408002											Gorlin syndrome
1059	112200	Q27.8	254784002											Blue rubber bleb nevus
1775 13987-613988		Q82.8	74911008											Dyskeratosis congenita
587	158320	D23 C44	403824007											Muir-Torre syndrome
2929 75050-612242		D12.6												Juvenile polyposis syndrome
79076	612242	D12.6												Juvenile polyposis of infancy
329971	174900	D12.6												Generalized juvenile polyposis/juvenile polyposis coli
653 62300-171400		D44.8	61808009											Multiple endocrine neoplasia type 2
99361	155240	C73												Familial medullary thyroid carcinoma
247698	171400	D44.8												Multiple endocrine neoplasia type 2A
247709	162300	D44.8												Multiple endocrine neoplasia type 2B
296	166000	Q78.4	268274005											Enchondromatosis
2608	310465	Q87.8												N syndrome
144 14350-614385		C18	315058005											Hereditary nonpolyposis colon cancer
587	158320	D23 C44	403824007											Muir-Torre syndrome
99817	276300	C18												Non-polyposis Turcot syndrome
1572 14700-615577	D83.8 D83.9		23238000											Common variable immunodeficiency
71290	601399	D69.4												Familial platelet syndrome with predisposition to acute myelogenous leukemia

ORPHAcode	OMIM	ICD10	SNOMED	Level 1	Level 2	Level 3	Level 4	Level 5	Level 6	Level 7	Level 8	Level 9	Level 10	Level 11
2128	235000	Q87.3					Hemihypertrophy							
798	269150	Q87.0					Schinzel-Giedion syndrome							
647	251260		234638009				Nijmegen breakage syndrome							
910	78780-610651	Q82.1	44600005				Xeroderma pigmentosum							
90342	278750	Q82.1	88877002				Xeroderma pigmentosum variant							
220295	78780-610651	Q87.1	Q82.1				Xeroderma pigmentosum/Cockayne syndrome complex							
276249	278700	Q82.1					Xeroderma pigmentosum complementation group A							
276252	610651	Q82.1					Xeroderma pigmentosum complementation group B							
276255	278720	Q82.1					Xeroderma pigmentosum complementation group C							
276258	278730	Q82.1					Xeroderma pigmentosum complementation group D							
276261	278740	Q82.1					Xeroderma pigmentosum complementation group E							
276264	278760	Q82.1					Xeroderma pigmentosum complementation group F							
276267	278780	Q82.1					Xeroderma pigmentosum complementation group G							
2849	267000	Q87.3					Perlman syndrome							
882	276700	E70.2	410056006				Tyrosinemia type 1							
220	194080	N04.1	236385009				Denys-Drash syndrome							
347	136680	N04.1	445431000				Frasier syndrome							
811	260400	D61.0					Shwachman-Diamond syndrome							
50944	224750	Q82.8					Schöpf-Schulz-Passarge syndrome							
60040	602501	Q87.3					Megalencephaly-capillary malformation-polymicrogyria syndrome							
99812	606593	D81.1					LIG4 syndrome							
166113							Bazex syndrome							
169079	611291	D81.1					Cernunnos-XLF deficiency							
221016		Q82.8					Rothmund-Thomson syndrome type 2							
240760	613078						Nijmegen breakage syndrome-like disorder							
251347	604391						Ataxia-telangiectasia-like disorder							
280558	613398						Warsaw breakage syndrome							
300576	608615						Oligodontia - cancer predisposition syndrome							
306498							PTEN hamartoma tumor syndrome							
137608							Segmental outgrowth - lipomatosis - arteriovenous malformation - epidermal nevus							
201	15108-615109	Q85.8	58037000				Cowden syndrome							
744	176920	Q87.3	23150001				Proteus syndrome							
109	153480	Q87.8	005-46011003				Bannayan-Riley-Ruvalcaba syndrome							
2969	158350	Q87.3					Proteus-like syndrome							
363972	613563	Q87.1					Noonan syndrome-like disorder with juvenile myelomonocytic leukemia							
220460	75100-608456	D12.6					Attenuated familial adenomatous polyposis							
247798	608456	D12.6					MUTYH-related attenuated familial adenomatous polyposis							
247806	175100	D12.6					APC-related attenuated familial adenomatous polyposis							
238510							Lymphoproliferative syndrome							
2442	00635-308240	D82.3	77121009				X-linked lymphoproliferative disease							
238505	13011-615122	D72.8					Autosomal recessive lymphoproliferative disease							
275517	607271	D72.8					Autoimmune lymphoproliferative syndrome with recurrent infections							
3261	03909-614470	D72.8					Autoimmune lymphoproliferative syndrome							
275523	605233	D72.8					Dianzani autoimmune lymphoproliferative disease							
268114		D72.8					RAS-associated autoimmune lymphoproliferative disease							
252202	276300						Constitutional mismatch repair deficiency syndrome							
276399	138800	E04.2					Familial multinodular goiter							
289539	614327						BAP1-related tumor predisposition syndrome							
293822	614456						MITF-related melanoma and renal cell carcinoma predisposition syndrome							
313846	614564						Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome							
319328							Inherited renal cell cancer-predisposing syndrome							
892	193300	Q85.8	46659004				Von Hippel-Lindau disease							
122	135150		110985001				Birt-Hogg-Dube syndrome							

