ORPHA	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
57146				**Rare hepatic disease**										
101940		E74.0			**Metabolic liver disease**									
220489						**Rare hereditary hemochromatosis**									
79230	602390-613313	E83.1					Hemochromatosis type 2							
139491	606069	E83.1					Hemochromatosis type 4							
225123	604250	E83.1					Hemochromatosis type 3							
97992				**Rare hematologic disease**										
108997					**Rare anemia**									
1047						**Sideroblastic anemia**								
75564		D64.3					Acquired idiopathic sideroblastic anemia							
98362							**Constitutional sideroblastic anemia**							
699	557000	D64.0	237985009					Pearson syndrome						
2598	0462-613561	G71.3						Mitochondrial myopathy and sideroblastic anemia						
2802	301310	D64.0						X-linked sideroblastic anemia - ataxia						
49827	249270	D53.1	237617006					Thiamine-responsive megaloblastic anemia syndrome						
75563	300751	D64.0						X-linked sideroblastic anemia						
255132	205950	D64.0						Autosomal recessive pyridoxine-refractory sideroblastic anemia						
260305	205950	D64.0						Autosomal recessive congenital sideroblastic anemia						
300298	615234	D64.0						Severe congenital hypochromic anemia with ringed sideroblasts						
68364						**Hemoglobinopathy**								
621	790-250800	D74.0						**Hereditary methemoglobinemia**						
139373	250800	D74.0						Recessive hereditary methemoglobinemia type 1						
139380	250800	D74.0						Recessive hereditary methemoglobinemia type 2						
330041		D74.0						Autosomal dominant methemoglobinemia						
2132		D58.2	51053007				Hemoglobin C disease							
2133		D58.2	25065001				Hemoglobin E disease							
90039		D58.2	66729008				Hemoglobin D disease							
99139							Unstable hemoglobin disease							
275745								**Alpha-thalassemia and related diseases**						
846	604131	D56.0	3-68913001					**Alpha-thalassemia**						
93616	613978	D56.0	48553001						Hemoglobin H disease					
163596	236750	D56.0							Hb Bart's hydrops fetalis					
232288								**Alpha-thalassemia-related diseases**						
847	301040	D56.0							Alpha-thalassemia - X-linked intellectual deficit syndrome					
98791	141750	D56.0	277918006						Alpha-thalassemia - intellectual deficit syndrome linked to chromosome 16					
231401	300448	D46.7 D56.0							Alpha-thalassemia - myelodysplastic syndrome					
275749								**Beta-thalassemia and related diseases**						
848	613985	D56.1	6-65959000					**Beta-thalassemia**						
231214	613985	D56.1							Beta-thalassemia major					
231222	613985	D56.1	191189009						Beta-thalassemia intermedia					
231226	603902	D56.1							Dominant beta-thalassemia					
231230		D58.2						**Beta-thalassemia associated with another hemoglobin anomaly**						
46532	35-613566	D56.4							Hereditary persistence of fetal hemoglobin - beta-thalassemia					
231237	141749	D56.2	16360009						Delta-beta-thalassemia					
231242		D58.2							Hemoglobin C - beta-thalassemia					
231249		D58.2	234392002						Hemoglobin E - beta-thalassemia					
330032		D56.1							Hemoglobin Lepore - beta-thalassemia					
231386		D58.2						**Beta-thalassemia with other manifestations**						
231256									Beta-thalassemia - trichothiodystrophy					

ORPHA	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
231393	314050	D56.1 D69.4							Beta-thalassemia - X-linked thrombocytopenia					
275752				**Sickle cell disease and related diseases**										
232	603903	D57.1 D57.2	-417357006					Sickle cell anemia						
251355								**Sickle cell disease associated with an other hemoglobin anomaly**						
251359		D57.2							Sickle cell - beta-thalassemia disease					
251365		D57.2							Sickle cell - hemoglobin C disease					
251370		D57.2							Sickle cell - hemoglobin D disease					
251375		D57.2							Sickle cell - hemoglobin E disease					
251380		D57.2							Hereditary persistence of fetal hemoglobin - sickle cell disease					
280615	613977	D58.2					Hemoglobinopathy Toms River							
98363						**Rare hemolytic anemia**									
182043								**Rare constitutional hemolytic anemia**						
1046	600461	D58.8						Lethal hemolytic anemia - genital anomalies						
2134	615008	D58.8						**Atypical hemolytic uremic syndrome**						
93575	612925	D58.8							Atypical hemolytic uremic syndrome with C3 anomaly					
93576	612922	D58.8							Atypical hemolytic uremic syndrome with MCP/CD46 anomaly					
93578	612924	D58.8							Atypical hemolytic uremic syndrome with B factor anomaly					
93579	5400-609814	D58.8							Atypical hemolytic uremic syndrome with H factor anomaly					
93580	612923	D58.8							Atypical hemolytic uremic syndrome with I factor anomaly					
93581	235400	D58.8							Atypical hemolytic uremic syndrome with anti-factor H antibodies					
217023	612926	D58.8							Atypical hemolytic uremic syndrome with thrombomodulin anomaly					
79293	245900	E78.6						Familial LCAT deficiency						
86818	300194	Q87.8						Alport syndrome - intellectual deficit - midface hypoplasia - elliptocytosis						
90044	5020-609153	D58.8						**Familial pseudohyperkalemia**						
100039									Familial pseudohyperkalemia type 1					
100040									Familial pseudohyperkalemia type 2					
100041									Familial pseudohyperkalemia, Cardiff type					
98364								**Rare constitutional hemolytic anemia due to a red cell membrane anomaly**						
822	70-612653	D58.0	55995005						Hereditary spherocytosis					
288		D58.1	191169008						**Hereditary elliptocytosis**					
98864										Common hereditary elliptocytosis				
98865										Homozygous hereditary elliptocytosis				
98866										Spherocytic elliptocytosis				
98867			9434008							Hereditary pyropoikilocytosis				
98868		D58.1								Southeast Asian ovalocytosis				
98365									**Stomatocytosis**					
3203	185000	D58.8								Overhydrated hereditary stomatocytosis				
3202	194380	D58.8								Dehydrated hereditary stomatocytosis				
71275	268150	D58.8	37272000							Rh deficiency syndrome				
99134										Intermediate stomatocytosis syndrome				
168577	608885	D58.8								Hereditary cryohydrocytosis with reduced stomatin				
98366									**Constitutional hemolytic anemia due to acanthocytosis**					
14	200100	E78.6	190787008							Abetalipoproteinemia				
59306	300842									McLeod neuroacanthocytosis syndrome				
169464	612300	D84.1							Primary CD59 deficiency					
98369								**Rare constitutional hemolytic anemia due to an enzyme disorder**						
79277	263700	E80.0	22935002						Congenital erythropoietic porphyria					
98370									**Hemolytic anemia due to hexose monophosphate shunt and glutathione metabolism anomalies**					

ORPHA	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
32	1900-266130	D55.1	234589002							**Glutathione synthetase deficiency**				
289846	266130	D55.1								Glutathione synthetase deficiency with 5-oxoprolinuria				
289849	231900	D55.1									Glutathione synthetase deficiency without 5-oxoprolinuria			
33574	230450	D55.1								Gamma-glutamylcysteine synthetase deficiency				
90030		D55.1								Hemolytic anemia due to glutathione reductase deficiency				
99135		D55.1								6-phosphogluconate dehydrogenase deficiency				
98372									**Hemolytic anemia due to a disorder of glycolytic enzymes**					
371	232800	E74.0	234406005							Glycogen storage disease due to muscle phosphofructokinase deficiency				
868	615512	D55.2	234405009							Triose phosphate-isomerase deficiency				
57	611881	E74.0	111578003							Glycogen storage disease due to aldolase A deficiency				
713	300653	D55.2	124335006							Glycogen storage disease due to phosphoglycerate kinase 1 deficiency				
766	266200	D55.2								Hemolytic anemia due to red cell pyruvate kinase deficiency				
712	613470	D55.2								Hemolytic anemia due to glucophosphate isomerase deficiency				
714	222800	D55.2								Hemolytic anemia due to diphosphoglycerate mutase deficiency				
90031	235700	D55.2								Non-spherocytic hemolytic anemia due to hexokinase deficiency				
248305		D55.2								Hemolytic anemia due to glyceraldehyde-3-phosphate dehydrogenase deficiency				
98374									**Hemolytic anemia due to an erythrocyte nucleotide metabolism disorder**					
1044	102800	D55.3								Anemia due to adenosine triphosphatase deficiency				
35120	266120	D55.3								Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency				
86817	612631	D55.3								Hemolytic anemia due to adenylate kinase deficiency				
99138		D55.3								Hemolytic anemia due to erythrocyte adenosine deaminase overproduction				
178330	140700	D58.2						Heinz body anemia						
182047							**Rare acquired hemolytic anemia**							
447	0818-615399	D59.5	1963002					Paroxysmal nocturnal hemoglobinuria						
90038	235400	D59.3						Typical hemolytic uremic syndrome						
98375								**Autoimmune hemolytic anemia**						
1959		D69.3	75331009						Evans syndrome					
90033		D59.1	3978000						Autoimmune hemolytic anemia, warm type					
90036		D59.1							Mixed-type autoimmune hemolytic anemia					
90037		D59.0	309742004						Drug-induced autoimmune hemolytic anemia					
228312									**Autoimmune hemolytic anemia, cold type**					
56425		D59.1								Cold agglutinin disease				
90035		D59.6	4-62871001							Paroxysmal cold hemoglobinuria				
551882								Hemolytic dise Hemolytic disease of the newborn with Kell allo-immunization						
182040						**Medullar aplasia**								
68383							**Rare constitutional medullar aplasia**							
124	900	D61.0	88854002					Blackfan-Diamond anemia						
1775	613988	Q82.8	74911008					Dyskeratosis congenita						
84	2	D61.0	30575002					Fanconi anemia						
3088	268130							Retinopathy - anemia- central nervous system anomalies						
3322	305000	D61.0						Hoyeraal-Hreidarsson syndrome						
3466	194350	D61.0						WT limb-blood syndrome						
3319	604498	D61.0	234482009					Congenital amegakaryocytic thrombocytopenia						
811	260400	D61.0						Shwachman-Diamond syndrome						
314399	614675							Autosomal dominant aplasia and myelodysplasia						
164823							**Rare acquired medullar aplasia**							
447	0818-615399	D59.5	1963002					Paroxysmal nocturnal hemoglobinuria						
824	254450	D47.4						Myelofibrosis with myeloid metaplasia						

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88	742-614743	D61.0	191256002					Idiopathic aplastic anemia						
98421								**Red cell aplasia**						
98871	227050	D60.1	234375006						Transient erythroblastopenia of childhood					
98872		D60.0							Adult pure red cell aplasia					
248293						**Rare deficiency anemia**									
248296							**Constitutional deficiency anemia**							
98360								**Constitutional anemia due to iron metabolism disorder**						
1195	209300	D50.8	111571009						Congenital atransferrinemia					
48818	604290	G23.0							Aceruloplasminemia					
83642	206100	D50.8							Microcytic anemia with liver iron overload					
209981	206200	D50.8							IRIDA syndrome					
98396								**Constitutional megaloblastic anemia due to vitamin B12 metabolism disorder**						
26	10-614857	E72.1							**Methylmalonic acidemia with homocystinuria**					
79282	277400	E72.1								Methylmalonic acidemia with homocystinuria, type cblC				
79283	277410	E72.1								Methylmalonic acidemia with homocystinuria, type cblD				
79284	277380	E72.1								Methylmalonic acidemia with homocystinuria, type cblF				
859	275350	D51.2	237934001						Transcobalamin II deficiency					
332	3320-261000	D51.0	234361004						Congenital intrinsic factor deficiency					
622	940-277410	E72.1							**Homocystinuria without methylmalonic aciduria**					
2169	236270	E72.1								Methylcobalamin deficiency type cblE				
2170	250940	E72.1								Methylcobalamin deficiency type cblG				
308380	277410	E72.1								Methylcobalamin deficiency type cblDv1				
35858	261100	D51.1							Gräsbeck-Imerslund disease					
98408								**Constitutional megaloblastic anemia due to folate metabolism disorder**						
51208	229100	E70.8							Formiminoglutamic aciduria					
90045	229050	D52.8							Hereditary folate malabsorption					
319651	613839	D52.8							Constitutional megaloblastic anemia with severe neurologic disease					
98415								**Vitamin B12- and folate-independent constitutional megaloblastic anemia**						
30	258900	D53.0	7-69525003						Hereditary orotic aciduria					
49827	249270	D53.1	237617006						Thiamine-responsive megaloblastic anemia syndrome					
206428									**Hypoxanthine-guanine phosphoribosyltransferase deficiency**					
510	0322-308950	E79.1	10406007							Lesch-Nyhan syndrome				
79233	300323	E79.8								Kelley-Seegmiller syndrome				
302330							Rare acquired (	Plummer-Vinson syndrome						
293830		D64.4				**Constitutional dyserythropoietic anemia**									
85		D64.4	52951008					**Congenital dyserythropoietic anemia**						
67044	300367	D69.4						Congenital dyserythropoietic anemia with thombocytopenia						
98869	5631-224120	D64.4	59548005					Congenital dyserythropoietic anemia type 1						
98870	105600	D64.4	26409005					Congenital dyserythropoietic anemia type 3						
98873	224100	D64.4	68870007					Congenital dyserythropoietic anemia type 2						
293825	613673	D64.4						Congenital dyserythropoietic anemia type 4						
363727	300835	D64.4						X-linked dyserythropoetic anemia with abnormal platelets and neutropenia						
77297	609628						Majeed syndrome							
199337	612714						Pancreatic insufficiency - anemia - hyperostosis							