Genes and disorders included in the Anemia ID next-generation sequencing genetic panel

This is not an exhaustive list of disorders that contribute to chronic anemia, nor of the genes that can cause these disorders.

Congenital dyserythropoietic anemias	
Disorder	**Gene**
Congenital dyserythropoietic anemia (CDA)	*C15ORF41, CDAN1, SEC23B, KIF23*
GATA1-related thrombocytopenia	*GATA1*
Majeed syndrome	*LPIN2*
Sideroblastic anemia	*ALAS2*

Diamond-Blackfan anemia
Genes
RPL5, RPL11, RPL35A, RPS7, RPS10, RPS19, RPS24, RPS26

Enzymopathies (RBC enzyme disorders)	
Disorder	**Gene**
Adenylate kinase deficiency	*AK1*
Aldolase A deficiency	*ALDOA*
G6PD deficiency	*G6PD*
Gamma-glutamylcysteine synthetase deficiency	*GCLC*
Glucose phosphate isomerase deficiency	*GPI*
Glutathione peroxidase deficiency	*GPX1*
Glutathione reductase deficiency	*GSR*
Glutathione synthetase deficiency	*GSS*
Glycogen storage disease VII	*PFKM*
Hexokinase deficiency	*HK1*
Methemoglobinemia due to deficiency of methemoglobin reductase	*CYB5R3*
Phosphoglycerate kinase 1 deficiency	*PGK1*
Pyruvate kinase deficiency	*PKLR*
Triosephosphate isomerase deficiency	*TPI1*
Uridine 5-prime monophosphate hydrolase deficiency	*NT5C3A*

Membranopathies (RBC membrane disorders)	
Disorder	**Gene**
Congenital X-linked hemolytic anemia	*ATP11C*
GLUT1 deficiency	*SLC2A1*
Hereditary elliptocytosis	*EPB41, GYPC, SPTA1, SPTB*
Hereditary pyropoikilocytosis	*SPTA1, SPTB*
Hereditary spherocytosis	*ANK1, EPB42, SLC4A1, SPTA1, SPTB*
Hereditary stomatocytosis	*ABCG5, ABCG8, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1*
McLeod neuroacanthocytosis syndrome	*XK*
Ovalocytosis	*SLC4A1*
Porencephaly	*COL4A1*

Hyperbilirubinemias	
Gilbert syndrome	*UGT1A1, UGT1A6, UGT1A7*
Hyperbilirubinemia, rotor type	*SLCO1B1, SLCO1B3*

agios