The latest signed off version for the GMS is v1.21. The current version, shown here, may differ from the signed-off version.

Cholestasis (Version 1.110)

Relevant disorders: R171
Panel types: GMS Rare Disease Virtual, GMS Rare Disease, GMS signed-off
Latest signed off version: v1.21 (20 Aug 2020)

78 Entities

78 reviewed, 61 green

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**ABCB11**	3 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Familial Intrahepatic Cholestasis • Cholestasis, progressive familial intrahepatic 2, 601847 • Cholestasis, Progressive Familial Intrahepatic 2 • PFIC2 • Cholestasis, benign recurrent intrahepatic, 2, 605479

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**ABCB4**	4 reviews 3 green	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Progressive Familial Intrahepatic Cholestasis • modifier in other forms of genetic cholestasis • Familial Intrahepatic Cholestasis • gallstones • cholelithiasis • PFIC • PFIC3 • Cholestasis, progressive familial intrahepatic 3, 602347 • Cholestasis, intrahepatic, of pregnancy, 3, 614972 • Neonatal and Adult Cholestasis • Cholestasis, Progressive Familial Intrahepatic 3 **Tags**
GreenGreen List (high evidence)	**ABCC2**	4 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
		2 green		• Expert Review Green • NHS GMS • Other • Victorian Clinical Genetics Services **Phenotypes** • modifier in biliary atresia • Dubin Johnson syndrome • Cholestasis • intrahepatic cholestasis of pregnancy • Dubin-Johnson syndrome, 237500 **Tags**
GreenGreen List (high evidence)	**ADK**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Hypermethioninemia due to adenosine kinase deficiency, OMIM:614300, MONDO:0013676 **Tags**
GreenGreen List (high evidence)	**AKR1D1**	4 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert Review Green • NHS GMS • Other

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• UKGTN • Victorian Clinical Genetics Services **Phenotypes** • Bile acid synthesis defect, congenital, 2 235555 • fat soluble vitamin deficiency • liver failure • bile salt synthesis defect • Bile acid synthesis defect, congenital, 2 • Neonatal and Adult Cholestasis • cholestasis **Tags**
GreenGreen List (high evidence)	**ALDOB**	4 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert Review Green • NHS GMS • Other • UKGTN • Victorian Clinical Genetics Services **Phenotypes** • acute liver failure • Neonatal and Adult Cholestasis • Fructose intolerance, hereditary **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**AMACR**	4 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Neonatal and Adult Cholestasis • Bile acid synthesis defect, congenital, 4 214950 **Tags**
GreenGreen List (high evidence)	**ATP7B**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Wilson disease, 277900 **Tags**
GreenGreen List (high evidence)	**ATP8B1**	4 reviews 3 green	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Familial Intrahepatic Cholestasis • Cholestasis, intrahepatic, of pregnancy, 1, 147480

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Cholestasis, Progressive Familial Intrahepatic 1 • Cholestasis, benign recurrent intrahepatic, 243300 • Cholestasis, progressive familial intrahepatic 1, 211600 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**BAAT**	3 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Hypercholanemia, Familial • fat soluble vitamin deficiency • Hypercholanemia, familial, 607748 • cholestasis • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**BCS1L**	4 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green • NHS GMS • Other **Phenotypes**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Cholestasis • GRACILE syndrome **Tags**
GreenGreen List (high evidence)	**CFTR**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Cholestasis • Neonatal and Adult Cholestasis • Cystic fibrosis, OMIM:219700, MONDO:0009061 • {Pancreatitis, hereditary}, OMIM:167800 **Tags**
GreenGreen List (high evidence)	**CLDN1**	3 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • ichthyosis-hypotrichosis-sclerosing cholangitis • Ichthyosis, leukocyte vacuoles, alopecia and sclerosing cholangitis, 607626 • Neonatal and Adult Cholestasis

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• NISCH syndrome • Neonatal ichthyosis sclerosing cholangitis (NISCH) syndrome **Tags**
GreenGreen List (high evidence)	**COG7**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Congenital disorder of glycosylation, type IIe , 608779 **Tags**
GreenGreen List (high evidence)	**CYP27A1**	4 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert Review Green • NHS GMS • Other • UKGTN • Victorian Clinical Genetics Services **Phenotypes** • Severe neonatal cholestasis • Cerebrotendinous xanthomatosis, 213700 **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**CYP7A1**	5 reviews 1 green 2 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Bile acid synthesis defect, congenital, 3 • Neonatal and Adult Cholestasis **Tags** • for-review • gene-checked
GreenGreen List (high evidence)	**CYP7B1**	5 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert Review Green • NHS GMS • Other • UKGTN • Victorian Clinical Genetics Services **Phenotypes** • Bile acid synthesis defect, congenital, 3, 613812 • Neonatal and Adult Cholestasis

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				Tags
GreenGreen List (high evidence)	**DCDC2**	4 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert Review Green • NHS GMS • Other • Victorian Clinical Genetics Services **Phenotypes** • Sclerosing cholangitis, neonatal, 617394 • PFIC type 5 • Neonatal sclerosis cholangitis • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**DGUOK**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880 **Tags**
GreenGreen List (high evidence)	**FAH**	3 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
		1 green		• NHS GMS **Phenotypes** • Neonatal and Adult Cholestasis • Tyrosinaemia, Type 1, 276700 • Cholestasis **Tags**
GreenGreen List (high evidence)	**GALE**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review • Expert Review Green **Phenotypes** • Galactose epimerase deficiency, OMIM:230350 • MONDO:0009257 **Tags**
GreenGreen List (high evidence)	**GALK1**	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review • Expert Review Green **Phenotypes** • Galactokinase deficiency with cataracts, OMIM:230200 • MONDO:0009255 Edit **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Q1_22_NHS_review • Q2_22_expert_review • Q2_22_rating
GreenGreen List (high evidence)	**GALM**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • Literature **Phenotypes** • Galactosemia IV, OMIM:618881 • MONDO:0030105 **Tags**
GreenGreen List (high evidence)	**GALT**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Galactosemia, OMIM:230400 • MONDO:0018116 **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**GBA**	3 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Gaucher disease, perinatal lethal 608013 • Gaucher disease, type I 230800 • Gaucher disease, type II 230900 • Gaucher disease, type III 231000 • Gaucher disease, type IIIC 231005 **Tags** • new-gene-name
GreenGreen List (high evidence)	**GBE1**	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Glycogen storage disease IV, OMIM:232500 **Tags** • Q1_22_NHS_review

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Q2_22_expert_review • Q2_22_rating
GreenGreen List (high evidence)	**HADHA**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • LCHAD deficiency, OMIM:609016, MONDO:0012173 **Tags**
GreenGreen List (high evidence)	**HNF1B**	2 reviews 1 green	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	**Sources** • Expert list • Expert Review Green **Phenotypes** • Renal cysts and diabetes syndrome, 137920 **Tags**
GreenGreen List (high evidence)	**HSD3B7**	3 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				Phenotypes • Bile acid sythesis defect, congenital, 1 607765 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**JAG1**	6 reviews 4 green	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Alagille syndrome 1, OMIM:118450 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**KIF12**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • Literature **Phenotypes** • Cholestasis • High Gamma-Glutamyltransferase (GGT) **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**LIPA**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • lysosomal acid lipase deficiency • Wolman disease, OMIM:278000, MONDO:0019148 • Cholesteryl ester storage disease, OMIM:278000, MONDO:0019149 • Neonatal and Adult Cholestasis • cholestasis **Tags**
GreenGreen List (high evidence)	**MPI**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Congenital disorder of glycosylation, type Ib, OMIM:602579 • MPI-CDG, MONDO:0011257 **Tags**
GreenGreen List (high evidence)	**MPV17**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green

List	Entity	Reviews	Mode of inheritance	Details
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				Phenotypes • Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 **Tags**
GreenGreen List (high evidence)	**MVK**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Mevalonic aciduria, OMIM:610377 **Tags**
GreenGreen List (high evidence)	**MYO5B**	4 reviews 2 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green • NHS GMS **Phenotypes** • Diarrhea 2, with microvillus atrophy, OMIM:251850 **Tags**
GreenGreen List (high evidence)	**NBAS**	3 reviews 1 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • Literature **Phenotypes**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Infantile liver failure syndrome 2, OMIM:616483 **Tags**
GreenGreen List (high evidence)	**NOTCH2**	5 reviews 3 green	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Alagille syndrome 2 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**NPC1**	4 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Niemann-Pick disease, type D, 257220 • Niemann-Pick disease type C1, 257220 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**NPC2**	4 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
		3 green		• Other **Phenotypes** • Neonatal and Adult Cholestasis • Niemann-Pick disease type C2, 607625 **Tags**
GreenGreen List (high evidence)	NR1H4	4 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • ciliopathy • Cholestasis, Progressive Familial Intrahepatic 5 • modifier of other genetic cholestatic conditions • Neonatal and Adult Cholestasis • Cholestasis, progressive familial intrahepatic 5, 617049 **Tags**
GreenGreen List (high evidence)	PEX1	5 reviews 1 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Peroxisome Biogenesis Disorder 1A (Zellweger), 214100 • Zellweger syndrome • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**PEX12**	3 reviews 1 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Peroxisome biogenesis disorder 3B 266510 • Peroxisome biogenesis disorder 3A (Zellweger) 614859 **Tags**
GreenGreen List (high evidence)	**PEX26**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Peroxisome biogenesis disorder 7A (Zellweger)614872 **Tags**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
GreenGreen List (high evidence)	**PEX6**	3 reviews 1 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Peroxisome biogenesis disorder 4A (Zellweger) 614862 **Tags**
GreenGreen List (high evidence)	**PKHD1**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Polycystic kidney disease 4, with or without hepatic disease, OMIM:263200 • MONDO:0044327 **Tags**
GreenGreen List (high evidence)	**POLG**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Mitochondrial DNA depletion syndrome 4A (Alpers type),

List	Entity	Reviews	Mode of inheritance	Details
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				OMIM:203700, MONDO:0008758 **Tags**
GreenGreen List (high evidence)	**RINT1**	3 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • Literature **Phenotypes** • Infantile liver failure syndrome 3 OMIM:618641 • infantile liver failure syndrome 3 MONDO:0032844 **Tags**
GreenGreen List (high evidence)	**SERPINA1**	5 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Alpha-1 Antitrypsin Deficiency • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	**SLC25A13**	4 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS

List	Entity	Reviews	Mode of inheritance	Details
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				• Other **Phenotypes** • CHOLESTASIS, NEONATAL INTRAHEPATIC, CAUSED BY CITRIN DEFICIENCY • NICCD • Citrullinemia type 2, neonatal onset • Citrullinemia type 2, adult onset • Citrullinemia, adult-onset type II 603471 • Citrullinemia, type II, neonatal-onset 605814 • Neonatal and Adult Cholestasis **Tags**
GreenGreen List (high evidence)	SMPD1	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Niemann-Pick disease, type A, OMIM:257200, MONDO:0009756 **Tags**
GreenGreen List (high evidence)	TALDO1	4 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS

List	Entity	Reviews	Mode of inheritance	Details
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				• Other **Phenotypes** • Transaldolase deficiency, 606003 **Tags**
GreenGreen List (high evidence)	**TJP2**	3 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Cholestasis, Progressive Familial Intrahepatic 4 • Neonatal and Adult Cholestasis • Cholestasis, progressive familial intrahepatic 4, 615878 **Tags**
GreenGreen List (high evidence)	**TRMU**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Liver failure, transient infantile, OMIM:613070 **Tags**

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GreenGreen List (high evidence)	**UGT1A1**	3 reviews 2 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • [Gilbert syndrome] 143500 • Crigler-Najjar syndrome, type I 218800 • Neonatal and Adult Cholestasis • Crigler-Najjar syndrome, type II 606785 • unconjugated jaundice **Tags**
GreenGreen List (high evidence)	**UNC45A**	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Green **Phenotypes** • Cholestasis • Diarrhoea • Bone fragility • Impaired hearing **Tags**
GreenGreen List (high evidence)	**USP53**	3 reviews 2 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				Phenotypes • Paediatric cholestatic liver disease • Cholestasis • deafness **Tags**
GreenGreen List (high evidence)	**VIPAS39**	3 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • Arthrogryposis, Renal Dysfunction, and Cholestasis 2 • ARC syndrome • Arthrogryposis-renal-cholestasis syndrome • Neonatal and Adult Cholestasis • Arthrogryposis, renal dysfunction, and cholestasis 2, 613404 **Tags**
GreenGreen List (high evidence)	**VPS33B**	4 reviews 3 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • NHS GMS • Other **Phenotypes** • arthrogryposis-renal-cholestasis syndrome

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 • Arthrogryposis, Renal Dysfunction, and Cholestasis 1 • Arthrogryposis, Renal Dysfunction, and Cholestasis Syndrome • ARC syndrome • Neonatal and Adult Cholestasis • Arthrogryposis, Renal Dysfunction, And Cholestasis 1 **Tags**
GreenGreen List (high evidence)	**YARS**	3 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green • Literature **Phenotypes** • Charcot-Marie-Tooth disease, dominant intermediate C 608323 • Intellectual disability • deafness • nystagmus • liver dysfunction **Tags** • new-gene-name
GreenGreen List (high evidence)	**ZFYVE19**	3 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Green

List	Entity	Reviews	Mode of inheritance	Details
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		2 green		• Literature **Phenotypes** • Cholestasis, progressive familial intrahepatic, 9, OMIM:619849 **Tags** • gene-checked
AmberAmber List (moderate evidence)	FARSA	2 reviews 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber • Literature **Phenotypes** • ?Rajab interstitial lung disease with brain calcifications 2, OMIM:619013 **Tags** • watchlist
AmberAmber List (moderate evidence)	FARSB	1 review	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber • Literature **Phenotypes**

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Rajab interstitial lung disease with brain calcifications, 613658 **Tags**
AmberAmber List (moderate evidence)	GNAS	3 reviews 1 green	MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted	**Sources** • Expert list • Expert Review Amber • NHS GMS • Other **Phenotypes** • McCune-Albright syndrome • Cholestasis **Tags**
AmberAmber List (moderate evidence)	IARS	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Amber **Phenotypes** • Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 **Tags** • new-gene-name

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Filter Entities 78 Entities				
AmberAmber List (moderate evidence)	LSR	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber • Literature **Phenotypes** • transient neonatal cholestasis • intellectual disability • short stature **Tags** • watchlist
AmberAmber List (moderate evidence)	MMP15	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber • Literature **Phenotypes** • Cholestasis, MONDO:0001751 • congenital heart disease, MONDO:0005453 **Tags** • watchlist
AmberAmber List (moderate evidence)	NPHP3	2 reviews 1 green	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Amber

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				Phenotypes • Renal-hepatic-pancreatic dysplasia 1, 208540 **Tags**
AmberAmber List (moderate evidence)	PEX14	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert list • Expert Review Amber **Phenotypes** • Peroxisome biogenesis disorder 13A (Zellweger), 614887 **Tags**
AmberAmber List (moderate evidence)	PEX2	3 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber • NHS GMS • Other **Phenotypes** • Neonatal and Adult Cholestasis • Peroxisome Biogenesis Disorder 5A (Zellweger), 614866 **Tags**
AmberAmber List (moderate evidence)	VPS50	2 reviews	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Amber

List	Entity	Reviews	Mode of inheritance	Details
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				• Literature **Phenotypes** • Neonatal cholestatic liver disease • Failure to thrive • Profound global developmental delay • Postnatal microcephaly • Seizures • Abnormality of the corpus callosum **Tags** • watchlist
RedRed List (low evidence)	AP1S1	2 reviews 1 green 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Red • Literature **Phenotypes** • Non-syndromic congenital intestinal failure • MEDNIK syndrome, OMIM:609313 **Tags**
RedRed List (low evidence)	PPM1F	2 reviews 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Red • Literature **Phenotypes**

List	Entity	Reviews	Mode of inheritance	Details
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				• sclerosing cholangitis • short stature • hypothyroidism • abnormal tongue pigmentation **Tags**
RedRed List (low evidence)	**SLC51A**	2 reviews 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Red • Literature **Phenotypes** • ?Cholestasis, progressive familial intrahepatic, 6, OMIM:619484 **Tags**
RedRed List (low evidence)	**WDR83OS**	2 reviews 1 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Red • Literature **Phenotypes** • Cholestasis **Tags**
No listNo list	CC2D2A	5 reviews 1 green 2 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Expert Review Removed • NHS GMS • Other

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				Phenotypes • COACH syndrome 216360 • Meckel syndrome 6 612284 • Joubert syndrome 9 612285 • Congenital hepatic fibrosis • Ciliopathy **Tags** • curated_removed
No listNo list	RPGRIP1L	4 reviews 1 green 2 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert list • Expert Review Removed • Illumina TruGenome Clinical Sequencing Services • NHS GMS • Radboud University Medical Center, Nijmegen • UKGTN **Phenotypes** • Meckel syndrome 5 (611561) • Joubert syndrome 7 (611560) • COACH syndrome (216360)

List	Entity	Reviews	Mode of inheritance	Details
Filter Entities 78 Entities				
				• Congenital hepatic fibrosis **Tags** • curated_removed
No listNo list	TMEM67	4 reviews 2 green 2 red	BIALLELIC, autosomal or pseudoautosomal	**Sources** • Emory Genetics Laboratory • Expert list • Expert Review Removed • Illumina TruGenome Clinical Sequencing Services • NHS GMS • Radboud University Medical Center, Nijmegen • UKGTN **Phenotypes** • COACH syndrome (216360) • {Bardet-Biedl syndrome 14, modifier of} (615991) • Nephronophthisis 11 (613550) • Meckel syndrome 3 (607361) • Joubert syndrome 6 (310688) • congenital hepatic fibrosis **Tags**

https://panelapp.genomicsengland.co.uk/panels/544/

List	Entity	Reviews	Mode of inheritance	Details
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				• curated_removed