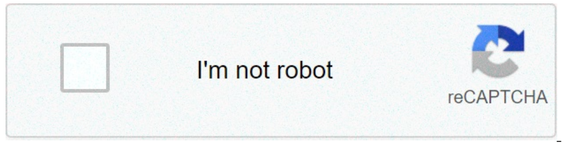

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Intrahepatic cholestasis of pregnancy (icp) icd 10

81222, 81223, 81403, 81404 x2, 81405 x2, 81406 x4, 81407, 81479 Cholestasis is characterized by jaundice and pruritus. It can present as the hallmark feature in progressive familial intrahepatic cholestasis (PFIC) or as a feature in other inherited disorders such as Alagille syndrome where cholestasis occur in 95% of cases in the neonatal period. PFIC is a group of autosomal recessive liver disorders caused by defects in bile secretion and is characterized by intrahepatic cholestasis with disease onset usually in infancy and childhood. PFIC patients usually develop fibrosis and end-stage liver disease before adulthood. Defects in PFIC-associated genes ATP8B1 and ABCB11 may also cause a milder disease called benign recurrent intrahepatic cholestasis. There are several other inherited disorders where cholestasis is frequent such as Alagille syndrome (JAG1 and NOTCH2), arthrogryposis, renal dysfunction, and cholestasis syndrome (ARC syndrome; VPS33B and VIPAS39), alpha-1-antitrypsin deficiency (SERPINA1), citrullinemia (SLC25A13), congenital defects of bile acid synthesis (HSD3B7 and AKR1D1), familial hypercholanemia (TJP2 and BAAT) and neonatal ichthyosis-sclerosing cholangitis syndrome (CLDN1). The prevalence of PFIC is unknown while the prevalence is as follows for other causes of cholestasis: Dubin-Johnson 1:1,300 in Iranian or Moroccan Jews, Alagille syndrome 1:30,000, Crigler-Najjar syndrome 1:1,000,000. The Gilbert syndrome prevalence is 3-7% but it does cause only abnormal laboratory findings but no clinical symptoms. Gene Associated phenotypes Inheritance ClinVar HGMD ABCB11 Cholestasis, Cholestasis, benign recurrent intrahepatic, 2 AD/AR 35 299 ABCB4 Gallbladder disease, Low phospholipid-associated cholelithiasis, Cholestasis AD/AR 27 224 ABCC2 Dubin-Johnson syndrome AD/AR 29 46 AKR1D1 Bile acid synthesis defect, congenital, 2 AR 7 14 ATP8B1 Intrahepatic cholestasis of pregnancy, Familial intrahepatic cholestasis, recurrent, Cholestasis, progressive familial intrahepatic, Benign recurrent intrahepatic cholestasis AD/AR 18 131 BAAT Hypercholanemia, familial AR 3 7 CFTR Cystic fibrosis, Congenital bilateral absence of the vas deferens AD/AR 518 1803 CREB3L3 Hypertriglyceridaemia AD 9 CYP7B1 Bile acid synthesis defect, Spastic paraplegia 5A, autosomal recessive AR 18 60 DCDC2 Deafness, Nephronophthisis, Sclerosing cholangitis, neonatal AR 13 9 DGUOK Mitochondrial DNA depletion syndrome, Portal hypertension, noncirrhotic, Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4 AR 23 62 EPCAM Diarrhea 5, with tufting enteropathy, congenital, Colorectal cancer, hereditary nonpolyposis AD/AR 38 80 FAH Tyrosinemia AR 53 102 HSD3B7 Bile acid synthesis defect, congenital, 1 AR 8 25 JAG1 Alagille syndrome AD 131 610 LCT Lactase deficiency AR 11 15 LMF1 Combined lipase deficiency AR 4 14 MKS1 Bardet-Biedl syndrome, Meckel syndrome AR 50 52 MYO5B* Diarrhea, with microvillus atrophy AR 14 80 NEUROG3 Diarrhea, malabsorptive, congenital AR 3 8 NOTCH2* Alagille syndrome, Hajdu-Cheney syndrome AD 37 70 NPC1 Niemann-Pick disease AR 164 472 NPC2 Niemann-pick disease AR 21 27 NPHP1 Nephronophthisis, Joubert syndrome, Senior-Loken syndrome AR 19 76 NPHP3 Nephronophthisis, Renal-hepatic-pancreatic dysplasia, Meckel syndrome AR 38 75 NPHP4 Nephronophthisis, Senior-Loken syndrome AR 20 113 NR1H4 Cholestasis, progressive familial intrahepatic 5 AR 6 5 PEX1 Heimler syndrome, Peroxisome biogenesis factor disorder 1A, Peroxisome biogenesis factor disorder 1B AR 112 134 PEX10 Adrenoleukodystrophy, neonatal, Zellweger syndrome, Peroxisome biogenesis disorder, Ataxia AR 34 29 PEX12 Zellweger syndrome, Peroxisome biogenesis disorder AR 43 37 PEX2 Zellweger syndrome, Peroxisome biogenesis disorder AR 16 18 PEX26 Adrenoleukodystrophy, neonatal, Zellweger syndrome, Peroxisome biogenesis disorder AR 13 27 PEX5 Adrenoleukodystrophy, neonatal, Rhizomelic chondrodysplasia punctata, Zellweger syndrome, Peroxisome biogenesis disorder AR 8 14 PEX6 Heimler syndrome, Peroxisome biogenesis disorder 4A, Peroxisome biogenesis disorder 4B AR 58 107 SCYL1 Spinocerebellar ataxia, autosomal recessive 21 AR 12 6 SERPINA1 Alpha-1-antitrypsin deficiency AR 49 80 SLC25A13 Citrin deficiency AR 24 113 SLC26A3 Diarrhea, secretory chloride, congenital AR 55 88 SMPD1 Niemann-Pick disease AR 110 249 SPINT2 Diarrhea, secretory sodium, congenital AR 6 12 TJP2 Cholestasis, progressive familial intrahepatic, Hypercholanemia, familial, Deafness, autosomal dominant 51 AD/AR 25 27 TMEM216 Joubert syndrome, Meckel syndrome AR 17 8 TRMU Liver failure, infantile, Reversible infantile respiratory chain deficiency AR 20 21 TTC37 Trichohepatoenteric syndrome, Primary immunodeficiency AR 12 64 UGT1A1 Crigler-Najjar syndrome, Gilbert syndrome, Breast milk jaundice AD/AR 29 144 VIPAS39 Arthrogryposis, renal dysfunction, and cholestasis 2 AR 8 13 VPS33B Arthrogryposis - renal dysfunction - cholestasis AD/AR 17 58 * Some, or all, of the gene is duplicated in the genome. Read more. # The gene has suboptimal coverage (means 20x with mapping quality score (MQ>20) reads), and/or the gene has exons listed under Test limitations section that are not included in the panel as they are not sufficiently covered with high quality sequence reads. The sensitivity to detect variants may be limited in genes marked with an asterisk (*) or number sign (#). Due to possible limitations these genes may not be available as single gene tests. Non-coding variants covered by Cholestasis Panel Gene Genomic location HG19 HGVS RefSeq RS-number ABCB11 Chr2:169873328 c.77-19T>A NM_003742.2 CFTR Chr7:117119654 c.-495C>T NM_000492.3 rs397507565 CFTR Chr7:117119797 NM_000492.3 CFTR Chr7:117119900 c.-249G>C NM_000492.3 CFTR Chr7:117119984 c.-165G>A NM_000492.3 rs145483167 CFTR Chr7:117120064 c.-85C>G NM_000492.3 CFTR Chr7:117120115 c.-34C>T NM_000492.3 rs756314710 CFTR Chr7:117120325 c.53+124T>C NM_000492.3 CFTR Chr7:117179040 c.870-1113_870-1110delGAAT NM_000492.3 rs397508809 CFTR Chr7:117182041 c.1117-26_1117-25delAT NM_000492.3 rs397508159 CFTR Chr7:117199500 c.1393-18G>A NM_000492.3 rs397508199 CFTR Chr7:117218381 c.1585-9412A>G NM_000492.3 rs397508229 CFTR Chr7:117227774 c.1585-19T>C NM_000492.3 rs778457306 CFTR Chr7:117227921 c.1679+34G>T NM_000492.3 rs767901668 CFTR Chr7:117229521 c.1680-886A>G NM_000492.3 rs397508266 CFTR Chr7:117229524 c.1680-883A>G NM_000492.3 CFTR Chr7:117229530 c.1680-877G>T NM_000492.3 rs397508261 CFTR Chr7:117243855 c.2908+19G>C NM_000492.3 rs370683572 CFTR Chr7:117246713 c.2909-15T>G NM_000492.3 rs397508455 CFTR Chr7:117246840 c.2988+33G>T NM_000492.3 CFTR Chr7:117251609 c.3140-26A>G NM_000492.3 rs76151804 CFTR Chr7:117251619 c.3140-16T>A NM_000492.3 rs767232138 CFTR Chr7:117251624 c.3140-11A>G NM_000492.3 CFTR Chr7:117266272 c.3469-1304C>G NM_000492.3 CFTR Chr7:117267864 c.3717+40A>G NM_000492.3 rs397508595 CFTR Chr7:117280015 c.3718-2477C>T NM_000492.3 rs75039782 CFTR Chr7:117282680 c.3873+33A>G NM_000492.3 rs397508622 CFTR Chr7:117288374 c.3874-4522A>G NM_000492.3 CFTR Chr7:117308395 c.*1233T>A NM_000492.3 DGUOK Chr2:74177650 c.444-62C>A NM_080916.2 DGUOK Chr2:74177701 c.444-11C>G NM_080916.2 rs536746349 EPCAM Chr2:47606078 c.556-14A>G NM_002354.2 rs376155665 JAG1 Chr20:10629767 c.1349-12T>G NM_000214.2 MYO5B Chr18:47365503 c.4852+11A>G NM_001080467.2 NPC1 Chr18:21132700 c.1554-1009G>A NM_000271.4 NPC1 Chr18:21137182 c.882-28A>G/T NM_000271.4 NPC1 Chr18:21137182 c.882-28A>G NM_000271.4 NPC1 Chr18:21137182 c.882-28A>T NM_000271.4 PEX6 Chr6:42933858 c.2301-15C>G NM_000287.3 rs267608236 PEX6 Chr6:42933952 c.2300+28G>A NM_000287.3 rs267608237 SERPINA1 Chr14:94854894 c.-5+2dupT NM_000295.4 SERPINA1 Chr14:94854896 c.-5+1G>A NM_000295.4 rs775786225 SMPD1 Chr11:6415102 c.1341-21_1341-18delAATG NM_000543.4 rs1312743513 UGT1A1 Chr2:234668848 c.-85_-83dupCAT NM_000463.2 UGT1A1 Chr2:234668879 c.-41_-40dupTA NM_000463.2 rs34983651 VPS33B Chr15:91550814 c.499-11G>A NM_018668.3 The strengths of this test include: CAP accredited laboratory CLIA-certified personnel performing clinical testing in a CLIA-certified laboratory Powerful sequencing technologies, advanced target enrichment methods and precision bioinformatics pipelines ensure superior analytical performance Careful construction of clinically effective and scientifically justified gene panels Some of the panels include the whole mitochondrial genome (please see the Panel Content section) Our Nucleus online portal providing transparent and easy access to quality and performance data at the patient level Our publicly available analytic validation demonstrating complete details of test performance ~2,000 non-coding disease causing variants in our clinical grade NGS assay for panels (please see 'Non-coding disease causing variants covered by this panel' in the Panel Content section) Our rigorous variant classification scheme Our systematic clinical interpretation workflow using proprietary software enabling accurate and traceable processing of NGS data Our comprehensive clinical statements Genes with partial, or whole gene, segmental duplications in the human genome are marked with an asterisk (*) if they overlap with the UCSC pseudogene regions. The technology may have limited sensitivity to detect variants in genes marked with these symbols (please see the Panel content table above). This test does not detect the following: Complex inversions Gene conversions Balanced translocations Some of the panels include the whole mitochondrial genome but not all (please see the Panel Content section) Repeat expansion disorders unless specifically mentioned Non-coding variants deeper than ±20 base pairs from exon-intron boundary unless otherwise indicated (please see above Panel Content / non-coding variants covered by the panel). This test may not reliably detect the following: Low level mosaicism in nuclear genes (variant with a minor allele fraction of 14.6% is detected with 90% probability) Stretches of mononucleotide repeats Low level heteroplasmy in mtDNA (>90% are detected at 5% level) Indels larger than 50bp Single exon deletions or duplications Variants within pseudogene regions/duplicated segments Some disease causing variants present in mtDNA are not detectable from blood, thus post-mitotic tissue such as skeletal muscle may be required for establishing molecular diagnosis. The sensitivity of this test may be reduced if DNA is extracted by a laboratory other than Blueprint Genetics. For additional information, please refer to the Test performance section and see our Analytic Validation. The genes on the panel have been carefully selected based on scientific literature, mutation databases and our experience. Our panels are sectioned from our high-quality, clinical grade NGS assay. Please see our sequencing and detection performance table for details regarding our ability to detect different types of alterations (Table). Assays have been validated for various sample types including EDTA-blood, isolated DNA (excluding from formalin fixed paraffin embedded tissue), saliva and dry blood spots (filter cards). These sample types were selected in order to maximize the likelihood for high-quality DNA yield. The diagnostic yield varies depending on the assay used, referring healthcare professional, hospital and country. Plus analysis increases the likelihood of finding a genetic diagnosis for your patient, as large deletions and duplications cannot be detected using sequence analysis alone. Blueprint Genetics' Plus Analysis is a combination of both sequencing and deletion/duplication (copy number variant (CNV)) analysis. The performance metrics listed below are from an initial validation performed at our main laboratory in Finland. The performance metrics of our laboratory in Seattle, WA, are equivalent. Performance of Blueprint Genetics high-quality, clinical grade NGS sequencing assay for panels. Sensitivity % (TP/(TP+FN) Specificity % Single nucleotide variants 99.89% (99,153/99,266) >99.9999% Insertions, deletions and indels by sequence analysis 1-10 bps 99.2% (7,745/7,806) >99.9999% 11-50 bps 99.13% (2,524/2,546) >99.9999% Copy number variants (exon level dels/dups) 1 exon level deletion (heterozygous) 100% (20/20) NA 1 exon level deletion (homozygous) 100% (5/5) NA 1 exon level deletion (het or homo) 100% (25/25) NA 2-7 exon level deletion (het or homo) 100% (44/44) NA 1-9 exon level duplication (het or homo) 75% (6/8) NA Simulated CNV detection 5 exons level deletion/duplication 98.7% 100.00% Microdeletion/-duplication sdrs (large CNVs, n=37)) Size range (0.1-47 Mb) 100% (25/25) The performance presented above reached by Blueprint Genetics high-quality, clinical grade NGS sequencing assay with the following coverage metrics Mean sequencing depth 143X Nucleotides with >20x sequencing coverage (%) 99.86% Performance of Blueprint Genetics Mitochondrial Sequencing Assay. Sensitivity % Specificity % ANALYTIC VALIDATION (NA samples; n=4) Single nucleotide variants Heteroplasmic (45-100%) 100.0% (50/50) 100.0% Heteroplasmic (35-45%) 100.0% (87/87) 100.0% Heteroplasmic (25-35%) 100.0% (73/73) 100.0% Heteroplasmic (15-25%) 100.0% (77/77) 100.0% Heteroplasmic (10-15%) 100.0% (74/74) 100.0% Heteroplasmic (5-10%) 100.0% (3/3) 100.0% Heteroplasmic (

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