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Name of Journal: World Journal of Gastroenterology

ESPS Manuscript NO: 24356

Manuscript Type: ORIGINAL ARTICLE

Retrospective Study

Clinical and *ABCB11* profiles in Korean infants with progressive familial
intrahepatic cholestasis

Ji Sook Park, Jae Sung Ko, Jeong Kee Seo, Jin Soo Moon, Sung Sup Park

Abstract

AIM: To investigate clinical profiles and mutations of *ABCB11* in Koreans with

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... Genome, GEO DataSets, GEO Profiles, GSS, GTR, HomoloGene, MedGen, MeSH ...

Twelve patients with clinical PFIC2 and ABCB11 mutations were identified, and 22 ...

Progressive familial intrahepatic cholestasis, type 2 (PFIC2) is a rare form of ... All patients were infants at the time of symptom onset, and had normal or ...

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23 Nov 2013 ... Progressive familial intrahepatic cholestasis (PFIC) is a group of rare ... present with intrahepatic cholestasis, usually in infancy and childhood. ... Based on clinical presentation, laboratory findings, liver histology and In patients with negative BSEP staining one should first test for ABCB 11 whereas in ...

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