UMC Utrecht expertisecentrum voor genetische ziekten van de darm en lever

Meest relevante wetenschappelijke publicaties

1. Meer van S, de Man RA, van den Berg AP, Houwen RHJ, Linn FHH, van Oijen MGH, Siersema PD, van Erpecum KJ. No increased risk of hepatocellular carcinoma in cirrhosis due to Wilson disease during long-term follow-up. J Gastroenterol Hepatol 2015;30:535-9.

2. Woerd WL van der, Wichers CG, Vestergaard AL, Andersen JP, Paulusma CC, Houwen RHJ, van der Graaf SF. Rescue of defective ATP8B1 trafficking by CF correctors as a therapeutic strategy for familial intrahepatic cholestasis. J Hepatol 2016;64:1339-47.

3. Houwen RHJ. Bis-choline tetrathiomolybdate for Wilson's disease. Lancet Gastroenterol Hepatol 2017;2:839-40.

4. Kamath BM, Baker A, Houwen R, Todorova L, Kerkar N. Systematic review: the epidemiology, natural history and burden of Alagille syndrome. J Pediatr Gastroenterol Nutr 2018;67:148-156.

5. Van Rijn JM, Ardy RC, Kuloğlu Z, Härter B, van Haaften-Visser DY, van der Doef HPJ, van Hoesel M, Kansu A, van Vugt AHM, Thian M, Kokke FTM, Krolo A, Keçeli Başaran M, Gurcan Kaya N, Ünlüsoy Aksu A, Dalgıç B, Ozcay F, Baris Z, Kain R, Stigter ECA, Lichtenbelt KD, Massink MPG, Duran KJ, Verheij JBGM, Lugtenberg D, Nikkels PJG, Brouwer HGF, Verkade HJ, Scheenstra R, Spee B, Nieuwenhuis EES, Coffer PJ, Janecke AR, van Haaften G, Houwen RHJ, Müller T, Middendorp S, Boztug K. Intestinal failure and aberrant lipid metabolism in patients with DGAT1 deficiency. Gastroenterology 2018;155:130-143.

6. Schene IF, Joore IP, Oka R, Mokry M, van Vugt AHM, van Boxtel R, van der Doef HPJ, van der Laan LJW, Verstegen MMA, van Hasselt PM, Nieuwenhuis EES, Fuchs SA. Prime editing for functional repair in patient-derived disease models. Nat Commun 2020;11:5352.

7. Kruitwagen HS, Oosterhoff LA, van Wolferen ME, Chen C, Nantasanti Assawarachan S, Schneeberger K, Kummeling A, van Straten G, Akkerdaas IC, Vinke CR, van Steenbeek FG, van Bruggen LWL, Wolfswinkel J, Grinwis GCM, Fuchs SA, Gehart H, Geijsen N, Vries RG, Clevers H, Rothuizen J, Schotanus BA, Penning LC, Spee B. Long-Term Survival of Transplanted Autologous Canine Liver Organoids in a *COMMD1*-Deficient Dog Model of Metabolic Liver Disease. Cells 2020;11:410.

8. Kamath BM, Stein P, Houwen R, Verkade HJ. Potential of ileal bile acid transporter inhibition as a therapeutic target in Alagille syndrome an progressive familial intrahepatic cholestasis. Liver Int 2020;40:1812-22

9. Large-Scale Production of LGR5-Positive Bipotential Human Liver Stem Cells. Schneeberger K, Sánchez-Romero N, Ye S, van Steenbeek FG, Oosterhoff LA, Pla Palacin I, Chen C, van Wolferen ME, van Tienderen G, Lieshout R, Colemonts-Vroninks H, Schene I, Hoekstra R, Verstegen MMA, van der Laan LJW, Penning LC, Fuchs SA, Clevers H, De Kock J, Baptista PM, Spee B. Hepatology 2020;72:257-270.

10. Knisely AS, Houwen RHJ. Liver Steatosis and Diarrhea After Liver Transplantation for Progressive Familial Intrahepatic Cholestasis Type 1: Can Biliary Diversion Solve These Problems? J Pediatr Gastroenterol Nutr 2021;72:341-342.

WILSON DISEASE: ORPHA-905

1. Meer van S, de Man RA, van den Berg AP, Houwen RHJ, Linn FHH, van Oijen MGH, Siersema PD, van Erpecum KJ. No increased risk of hepatocellular carcinoma in cirrhosis due to Wilson disease during long-term follow-up. J Gastroenterol Hepatol 2015;30:535-9.

2. Houwen RHJ. Bis-choline tetrathiomolybdate for Wilson's disease. Lancet Gastroenterol Hepatol 2017;2:839-40.

3. Schene IF, Joore IP, Oka R, Mokry M, van Vugt AHM, van Boxtel R, van der Doef HPJ, van der Laan LJW, Verstegen MMA, van Hasselt PM, Nieuwenhuis EES, Fuchs SA. Prime editing for functional repair in patient-derived disease models. Nat Commun. 2020;11:5352.

PFIC & BRIC: ORPHA-172 & ORPHA-65682

1. Woerd WL van der, Wichers CG, Vestergaard AL, Andersen JP, Paulusma CC, Houwen RHJ, van der Graaf SF. Rescue of defective ATP8B1 trafficking by CF correctors as a therapeutic strategy for familial intrahepatic cholestasis. J Hepatol 2016;64:1339-47.

2. Woerd WL van der, Houwen RHJ, van der Graaf SFJ. Current and future therapies for inherited cholestatic liver diseases. World J Gastroenterol 2017;23:763-75.

3. Knisely AS, Houwen RHJ. Liver Steatosis and Diarrhea After Liver Transplantation for Progressive Familial Intrahepatic Cholestasis Type 1: Can Biliary Diversion Solve These Problems? J Pediatr Gastroenterol Nutr 2021;72:341-342.

ALAGILLE SYNDROME: ORPHA-52

1. Kamath BM, Baker A, Houwen R, Todorova L, Kerkar N. Systematic review: the epidemiology, natural history and burden of Alagille syndrome. J Pediatr Gastroenterol Nutr 2018;67:148-156.

2. Kamath BM, Stein P, Houwen R, Verkade HJ. Potential of ileal bile acid transporter inhibition as a therapeutic target in Alagille syndrome an progressive familial intrahepatic cholestasis. Liver Int 2020;40:1812-22

CONGENITAL DIARRHEA: ORPHA-363300

1. Wiegerinck CL, Janecke AR, Schneeberger K, Vogel GF, van Haaften-Visser DY, Escher JC, Adam R, Thoeni CE, Pfaller K, Jordan AJ, Weis CA, Nijman IJ, Monroe GR, van Hasselt PM, Cutz E, Klumperman J, Clevers H, Nieuwenhuis EE, Houwen RHJ, van Haaften G, Hess MW, Huber LA, Stapelbroek JM, Mueller T, Middendorp S. Loss of syntaxin 3 causes variant microvillus inclusion disease. Gastroenterology 2014;147:65-8.

2. Haaften-Visser DY van, Harakalova M, Mocholi E, van Montfrans JM, Elkadri A, Rieter E, Fiedler K, van Hasselt PM, Triffaux EMM, van Haelst MM, Nijman IJ, Kloosterman WP, Nieuwenhuis EES, Muise AM, Cuppen E, Houwen RHJ, Coffer PJ. Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. J Biol Chem 2017;292:7904-20.

3. Van Rijn JM, Ardy RC, Kuloğlu Z, Härter B, van Haaften-Visser DY, van der Doef HPJ, van Hoesel M, Kansu A, van Vugt AHM, Thian M, Kokke FTM, Krolo A, Keçeli Başaran M, Gurcan Kaya N, Ünlüsoy Aksu A, Dalgıç B, Ozcay F, Baris Z, Kain R, Stigter ECA, Lichtenbelt KD, Massink MPG, Duran KJ, Verheij JBGM, Lugtenberg D, Nikkels PJG, Brouwer HGF, Verkade HJ, Scheenstra R, Spee B, Nieuwenhuis EES, Coffer PJ, Janecke AR, van Haaften G, Houwen RHJ, Müller T, Middendorp S, Boztug K. Intestinal failure and aberrant lipid metabolism in patients with DGAT1 deficiency. Gastroenterology 2018;155:130-143.