

1. De Tomasi L, David P, Humbert C, Silbermann F, Arrondel C, Tores F, Fouquet S, Desgrange A, Niel O, Bole-Feysot C, Nitschké P, Roume J, Cordier MP, Pietrement C, Isidor B, Khau Van Kien P, Gonzales M, Saint-Frison MH, Martinovic J, Novo R, Piard J, Cabrol C, Verma IC, Puri R, Journal H, Aziza J, Gavard L, Said-Menthon MH, Heidet L, Saunier S, Jeanpierre C. Mutations in GREB1L Cause Bilateral Kidney agenesis in Humans and Mice. *Am J Hum Genet.* 2017; 101(5):803-814.
2. Heidet L, Morinière V, Henry C, De Tomasi L, Reilly ML, Humbert C, Alibeu O, Fourrage C, Bole-Feysot C, Nitschké P, Tores F, Bras M, Jeanpierre M, Pietrement C, Gaillard D, Gonzales M, Novo R, Schaefer E, Roume J, Martinovic J, Malan V, Salomon R, Saunier S, Antignac C, Jeanpierre C. Targeted Exome Sequencing Identifies PBX1 as Involved in Monogenic Congenital Anomalies of the Kidney and Urinary Tract. *J Am Soc Nephrol.* 2017 28(10):2901-2914
3. Ryan R, Failler M, Reilly ML, Garfa-Traore M, Delous M, Filhol E, Reboul T, Bole-Feysot C, Nitschké P, Baudouin V, Amselem S, Escudier E, Legendre M, Benmerah A, Saunier S. Functional characterization of tektin-1 in motile cilia and evidence for TEKT1 as a new candidate gene for motile ciliopathies. *Hum Mol Genet.* 2017
4. Boyer O, Dorval G, Servais A. Hereditary Podocytopathies in Adults: The Next Generation. *Kidney Dis (Basel).* 2017; 3(2):50-56.
5. Dorval G, Gribouval O, Martinez-Barquero V, Machuca E, Tête MJ, Baudouin V, Benoit S, Chabchoub I, Champion G, Chauveau D, Chehade H, Chouchane C, Cloarec S, Cochat P, Dahan K, Dantal J, Delmas Y, Deschênes G, Dolhem P, Durand D, Ekinci Z, El Karoui K, Fischbach M, Grunfeld JP, Guignon V, Hachicha M, Hogan J, Hourmant M, Hummel A, Kamar N, Krummel T, Lacombe D, Llanas B, Mesnard L, Mohsin N, Niaudet P, Nivet H, Parvex P, Pietrement C, de Pontual L, Noble CP, Ribes D, Ronco P, Rondeau E, Sallee M, Tsimaratos M, Ulinski T, Salomon R, Antignac C, Boyer O. Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. *Pediatr Nephrol.* 2017
6. Dorval G, Lion M, Guérin S, Krid S, Galmiche-Rolland L, Salomon R, Boyer O. Immunoabsorption in Anti-GBM Glomerulonephritis: Case Report in a Child and Literature Review. *Pediatrics.* 2017; 140(5) pii: e20161733.
7. Gondra L, Décramer S, Chalouhi GE, Muller F, Salomon R, Heidet L. Hyperechogenic kidneys and polyhydramnios associated with HNF1B gene mutation. *Pediatr Nephrol.* 2016 (10):1705-8.

8. Lerner J, Bagattin A, Verdeguer F, Makinistoglu MP, Garbay S, Felix T, Heidet L, Pontoglio M. Human mutations affect the epigenetic/bookmarking function of HNF1B. *Nucleic Acids Res.* 2016;44(17):8097-111. Gross O, Kashtan CE, Rheault MN, Flinter F, Savige J, Miner JH, Torra R, Ars E, Deltas C, Savva I, Perin L, Renieri A, Ariani F, Mari F, Baigent C, Judge P, Knebelman B, Heidet L, Lagas S, Blatt D, Ding J, Zhang Y, Gale DP, Prunotto M, Xue Y, Schachter AD, Morton LCG, Blem J, Huang M, Liu S, Vallee S, Renault D, Schifter J, Skelding J, Gear S, Friede T, Turner AN, Lennon R. Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. *Nephrol Dial Transplant.* 2017;32(6):916-924.
9. Audrézet MP, Corbiere C, Lebbah S, Morinière V, Broux F, Louillet F, Fischbach M, Zaloszc A, Cloarec S, Merieau E, Baudouin V, Deschênes G, Roussey G, Maestri S, Visconti C, Boyer O, Abel C, Lahoche A, Randrianaivo H, Bessenay L, Mekahli D, Ouertani I, Decramer S, Ryckenwaert A, Cornec-Le Gall E, Salomon R, Ferec C, Heidet L. Comprehensive PKD1 and PKD2 Mutation Analysis in Prenatal Autosomal Dominant Polycystic Kidney Disease. *J Am Soc Nephrol.* 2016; 27(3):722-9.
10. Girard M, Bizet AA, Lachaux A, Gonzales E, Filhol E, Collardeau-Frachon S, Jeanpierre C, Henry C, Fabre M, Viremouneix L, Galmiche L, Debray D, Bole-Feysot C, Nitschke P, Pariente D, Guettier C, Lyonnet S, Heidet L, Bertholet A, Jacquemin E, Henrion-Caude A, Saunier S. DCDC2 Mutations Cause Neonatal Sclerosing Cholangitis. *Hum Mutat.* 2016 37(10):1025-9.
11. Grampa V, Delous M, Zaidan M, Ody G, Thomas S, Elkhartoufi N, Filhol E, Niel O, Silbermann F, Lebreton C, Collardeau-Frachon S, Rouvet I, Alessandri JL, Devisme L, Dieux-Coeslier A, Cordier MP, Capri Y, Khung-Savatovsky S, Sigaudy S, Salomon R, Antignac C, Gubler MC, Benmerah A, Terzi F, Attié-Bitach T, Jeanpierre C, Saunier S. Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. *PLoS Genet.* 2016; 12(3):e1005894.
12. Mencarelli MA, Heidet L, Storey H, van Geel M, Knebelmann B, Fallerini C, Miglietti N, Antonucci MF, Cetta F, Sayer JA, van den Wijngaard A, Yau S, Mari F, Bruttini M, Ariani F, Dahan K, Smeets B, Antignac C, Flinter F, Renieri A. Evidence of digenic inheritance in Alport syndrome. *J Med Genet.* 2015; 52(3):163-74.
13. Bizet AA, Becker-Heck A, Ryan R, Weber K, Filhol E, Krug P, Halbritter J, Delous M, Lasbennes MC, Linghu B, Oakeley EJ, Zarhrate M, Nitschké P, Garfa-Traore M, Serluca F, Yang F, Bouwmeester T, Pinson L, Cassuto E, Dubot P, Elshakhs NA, Sahel JA, Salomon R, Drummond IA, Gubler MC, Antignac C, Chibout S, Szustakowski JD, Hildebrandt F, Lorentzen E, Sailer AW, Benmerah A, Saint-Mezard P, Saunier S. Mutations in TRAF3IP1/IFT54 reveal a new role for IFT proteins in microtubule stabilization. *Nat Commun.* 2015; 6:8666.