

10 PUBLICATIONS H. DOLLFUS

1. *Mutations in TUBGCP4 alter microtubule organization via the γ -tubulin ring complex in autosomal-recessive microcephaly with chorioretinopathy.* Scheidecker S, Etard C, Haren L, Stoetzel C, Hull S, Arno G, Plagnol V, Drunat S, Passemard S, Toutain A, Obringer C, Koob M, Geoffroy V, Marion V, Strähle U, Ostergaard P, Verloes A, Merdes A, Moore AT, Dollfus H. *Am J Hum Genet.* 2015 Apr 2;96(4):666-74
2. *Predominantly Cone-System Dysfunction as Rare Form of Retinal Degeneration in Patients With Molecularly Confirmed Bardet-Biedl Syndrome.* Scheidecker S, Hull S, Perdomo Y, Studer F, Pelletier V, Muller J, Stoetzel C, Schaefer E, Defoort-Dhellemmes S, Drumare I, Holder GE, Hamel CP, Webster AR, Moore AT, Puech B, Dollfus HJ. *Am J Ophthalmol.* 2015 Aug;160(2):364-372
3. *Comparing the Bbs10 complete knockout phenotype with a specific renal epithelial knockout one highlights the link between renal defects and systemic inactivation in mice.* Cognard N, Scerbo MJ, Obringer C, Yu X, Costa F, Haser E, Le D, Stoetzel C, Roux MJ, Moulin B, Dollfus H, Marion V. *Cilia.* 2015 Aug 13
4. *BBS-induced ciliary defect enhances adipogenesis, causing paradoxical higher-insulin sensitivity, glucose usage, and decreased inflammatory response.* Marion V, Mockel A, De Melo C, Obringer C, Claussmann A, Simon A, Messaddeq N, Durand M, Dupuis L, Loeffler JP, King P, Mutter-Schmidt C, Petrovsky N, Stoetzel C, Dollfus H. *Cell Metab.* 2012; 16(3):363-77.
5. *Pharmacological modulation of the retinal unfolded protein response in bardet-biedl syndrome reduces apoptosis and preserves light detection ability.* Mockel A, Obringer C, Hakvoort TB, Seeliger M, Lamers WH, Stoetzel C, Dollfus H, Marion V. *J Biol Chem.* 2012; 287(44):37483-94.
6. *Exome sequencing identifies mutations in LZTFL1, a BBSome and smoothed trafficking regulator, in a family with Bardet-Biedl syndrome with situs inversus and insertional polydactyly.* Marion V, Stutzmann F, Gérard M, De Melo C, Schaefer E, Claussmann A, Hellé S, Delague V, Souied E, Barrey C, Verloes A, Stoetzel C, Dollfus H. *J Med Genet.* 2012; 49(5):317-21.
7. *Retinal dystrophy in Bardet-Biedl syndrome and related syndromic ciliopathies.* Mockel A, Perdomo Y, Stutzmann F, Letsch J, Marion V, Dollfus H *Prog Retin Eye Res.* 2011; 30(4):258-74.
8. *Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy.* Otto EA, Hurd TW, Airik R, Chaki M, Zhou W, Stoetzel C, Patil SB, Levy S, Ghosh AK, Murga-Zamalloa CA, van Reeuwijk J, Letteboer SJ, Sang L, Giles RH, Liu Q, Coene KL, Estrada-Cuzcano A, Collin RW, McLaughlin HM, Held S, Kasanuki JM, Ramaswami G, Conte J, Lopez I, Washburn J, Macdonald J, Hu J, Yamashita Y, Maher ER, Guay-Woodford LM, Neumann HP, Obermüller N, Koenekoop RK, Bergmann C, Bei X, Lewis RA, Katsanis N, Lopes V, Williams DS, Lyons RH, Dang CV, Brito DA, Dias MB, Zhang X, Cavalcoli JD, Nürnberg G, Nürnberg P, Pierce EA, Jackson PK, Antignac C, Saunier S, Roepman R, Dollfus H, Khanna H, Hildebrandt *Nat Genet.* 2010 Oct;42(10):840-50.
9. *Transient ciliogenesis involving Bardet-Biedl syndrome proteins is a fundamental characteristic of adipogenic differentiation.* Marion V, Stoetzel C, Schlicht D, Messaddeq N, Koch M, Flori E, Danse JM, Mandel JL, Dollfus H. *Proc Natl Acad Sci U S A.* 2009; 106(6):1820-5.
10. *BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus.* Stoetzel C, Laurier V, Davis EE, Muller J, Rix S, Badano JL, Leitch CC, Salem N, Chouery E, Corbani S, Jalk N, Vicaire S, Sarda P, Hamel C, Lacombe D, Holder M, Odent S, Holder S, Brooks AS, Elcioglu NH, Silva ED, Rossillion B, Sigaudy S, de Ravel TJ, Lewis RA, Leheup B, Verloes A, Amati-Bonneau P, Mégarbané A, Poch O, Bonneau D, Beales PL, Mandel JL, Katsanis N, Dollfus H. *Nat Genet.* 2006; 38(5):521-4.