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Originalarbeiten:

1. Hildebrandt F, Pohlmann A, **Omran H**. LODVIEW: A computer program for the graphicalevaluation of lod score results in exclusion mapping of human disease genes. *Comput Biomed Res* 26:592–599, 1993
2. Hildebrandt F, Singh-Sawhney I, Schnieders B, Centofante L, **Omran H**, Pohlmann A, SchmaltzC, Wedekind H, Schubotz C, Antignac C, Weber JL, Brandis M.and Members of the APN Study Group. Mapping of a gene for familial juvenile nephronophthisis: Refining the map and definition of flanking markers on chromosome 2. *Am J Hum Genet* 53:1256–1261, 1993 (IF 11,2)
3. Kaiser R, Kern A, Fressle R, Steinbrecher A, **Omran H**, Malzacher V, Kügler D, Kampa D,Batsford S. Zeckenvermittelte Erkrankungen in Baden-Württemberg. *Münch med Wschr* 138:647-652, 1996
4. **Omran H**, Hildebrandt F, Korinthenberg R, Brandis M. Probable Opitz trigonocephaly Csyndrome with medulloblastoma. *Am J Med Genetics* 69:395-399, 1997 (IF 2,3)
5. Vollmer M, Koehrer M, Topaloglu R, Strahm B, **Omran H**, Hildebrandt F. Two novel mutations of the gene for Kir 1.1 (ROMK) in neonatal Bartter syndrome. *Pediatr Nephrol* 12:69-71, 1998 (IF2,9)
6. **Omran H**, Ketelsen UP, Heinen F, Sauer M, Rudnik-Schöneborn S, Wirth B, Zerres K, KratzerW, Korithenberg R. Axonal neuropathy and predominance of type II myofibres in SMA I. *J ChildNeurol* , 13:327-331, 1998 (IF 1,9)
7. **Omran H**, Häffner K, Vollmer M, Pigulla J, Wagner G, Caridi G, Hildebrandt F. Exclusion of the candidate genes *ACE* and *Bcl-2* for six families with nephronophthisis not linked to the *NPH1* locus. *Nephrol Dial Transplant* 14:2328-2331, 1999 (IF 3,3)
8. **Omran H**, Fernandez C, Jung M, Häffner K, Fargier B, Waldherr R, Gretz N, Brandis M, Rüschendorf F, Reis A, Hildebrandt F. Identification of a new gene locus for adolescent nephronophthisis, on chromosome 3q22 in a large Venzuelan pedigree. *Am J Hum Genet* 66:118-127, 2000 (IF 11,2)
9. **Omran H**, Uhl M, Brandis M, Wolff G. Survival and dominant transmission of "lethal" platyspondylic dwarfism of the "Westcoast-types,,. *J Ped* 136:411-413, 2000 (IF 4,0)
10. **Omran H**, Häffner K, Völkel A, Kuehr J, Ketelsen UP, Ross UH, Konietzko N, Wienker T, BrandisM, Hildebrandt F. Homozygosity mapping of a gene locus for primary ciliary dyskinesia on chromosome 5p and identification of the heavy dynein chain *DNAH5* as a candidate gene. *Am J Resp Cell Mol Biol* 23:669-702, 2000 (IF 4,1)
11. **Omran H**, Häffner K, Burth S, Fernandez C, Fargier B, Villaquiran A, Nothwang HG, Schnittger S, Lehrach H, Woo D, Brandis M, Sudbrak R, Hildebrandt F. Human adolescent nephronophthisis: Gene locus synteny with polycystic kidney disease in pcy mice. *J Am Soc Nephrol* 12:107-113, 2001 (IF 9,0)
12. Hildebrandt F, Rensing C, Betz RC, Sommer U, Birnbaum S, Imm A, **Omran H**, Leipholdt M, Otto E, and Members of the APN Study Group. Establishing an Algorithm for Molecular Genetic Diagnostics in 127 Families with Juvenile Nephronophthisis. *Kidney Int* 59:434-445, 2001 (IF 7,9)
13. **Omran H**, Häffner K, Burth S, Ala-Mello S, Antignac C, Hildebrandt F. Evidence for further genetic heterogeneity in nephronophthisis. *Nephrol Dial Transplant* 16:755-758, 2001 (IF 3,3)
14. Birkenhäger R, Otto E, Schürmann MJ, Ruf EM, Maier-Lutz I, Beekmann F, Fekete A, **Omran H**, Feldmann D, Milford DV, Jeck N, Konrad M, Landau D, Knoers N, Antignac C, Sudbrak R,Kispert A, Hildebrandt F. Mutation of BSND causes Bartter syndrome with sensorineural deafness and kidney failure. *Nat Genet* 29:310-314, 2001 (IF 35,2)
15. **Omran H**, Sasmaz G, Häffner K, Volz A, Olbrich H, Otto E, Wienker TF, Korinthenberg R,Brandis M, Antignac C, Hildebrandt F. Identification of a gene locus for Senior-Løken syndrome in the region of the nephronophthisis type 3 gene. *J Am Soc Nephrol* 13:75-79, 2002 (IF 9,0)
16. Olbrich H, Häffner K, Kispert A, Völkel A, Volz A, Sasmaz G, Reinhardt R, Hennig S, Lehrach H,Konietzko N, Zariwala M, Noone PG, Knowles M, Mitchison HM, Meeks M, Chung EMK,Hildebrandt F, Sudbrak R, **Omran H**. Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of left-right asymmetry. *Nat Genet* 30:143-144, 2002 (IF 35,2)
17. Schuermann MJ, Otto E, Becker A, Saar K, Ruschendorf F, Polak BC, Ala-Mello S, Hoefele J, Wiedensohler A, Haller M, **Omran H**, Nurnberg P, Hildebrandt F. Mapping of Gene Loci for

Nephronophthisis Type 4 and Senior-Loken Syndrome, to Chromosome 1p36. *Am J Hum Genet*70:1240-1246, 2002 (IF 11,2)

18. Volz A, Melkaoui R, Hildebrandt F, **Omran H**. Candidate gene analysis of KIAA0678 encoding a DnaJ-like protein for adolescent nephronophthisis and Senior-Loken syndrome type 3. *Cytogenet Genome Res* 97:163-166, 2002 (IF 2,0)
19. Neesen J, Drenkhahn J, Tiede S, Burfeind P, Grzmil M, Konietzko J, Dixkens C, Kreutzberger J,Laccone F, Engel W, **Omran H**. Identification of the Human Ortholog of the t-Complex-Encoded Protein TCTEX2 and Evaluation as a Candidate Gene for Primary Ciliary Dyskinesia. *Cytogenet Genome Res* 98:38-44, 2002 (IF 2,0)
20. Rudnik-Schöneborn S, Goebel H, Schlote W, Molaian S, **Omran H**., Ketelsen U., KorinthenbergR., Wenzel D., Lauffer H., Kreiss-Nachtsheim M., Wirth B., Zerres K. Classical infantile spinalmuscular atrophy with SMN deficiency causes sensory neuronopathy. *Neurology* 60:983-7, 2003 (IF 8,2)
21. Fliegauf M, Fröhlich C, Horvath J, Olbrich H, Hildebrandt F, **Omran H**. Identification of the human CYS1 gene and candidate gene analysis in Boichis disease. *Ped Nephrol* 18:498-505, 2003 (IF2,0)
22. Kispert A, Olbrich H, Volz A, Ketelsen UP, Horvath J, Melkaoui R, Petry M, Zariwala M, NoonePG, Knowles M, **Omran H**. Evidence for Genotype-Phenotype Correlations in *DNAH5* Mutations of Patients with Primary Ciliary Dyskinesia - Expression of *DNAH5*'s Mouse Ortholog in the Upper and Lower Airway. *Thorax* 58:552-4, 2003 (IF 8,3)
23. Horvath J, Ketelsen UP, Geibel-Zehender A, Korinthenberg R, **Omran H**. Identification of a novel *LAMP2* mutation responsible for X chromosomal dominant Danon disease. *Neuropediatrics* 34:270-3, 2003 (IF 1,2)
24. Olbrich H, Fliegauf M, Hoefele J,Kispert A, Otto E, Volz A, Wolf MT, Sasmaz G, Trauer U,Reinhardt R, Sudbrak R, Antignac C, Gretz N, Walz G, Schermer B, Benzing T, Hildebrandt F, **Omran H**. Mutations in a novel gene, *NPHP3*, cause adolescent nephronophthisis, tapeto-retinal degeneration and hepatic fibrosis. *Nat Genet* 34:455-9, 2003 (IF 35,2)
25. Ibanez-Tallon I, Pagenstecher A, Fliegauf M, Olbrich H, Kispert A, Ketelsen UP, North A, HeintzN, **Omran H**. Dysfunction of axonemal dynein heavy chain Mdnah5 inhibits ependymal flow and reveals a novel mechanism for hydrocephalus formation. *Hum Mol Genet* 13:2133-41, 2004 (IF7,7)
26. Zankl A, Neumann L, Ignatius J, Nikkels P, Schrander-Stumpel C, Mortier G, **Omran H**, Wright M, Hilbert K, Bonafe L, Spranger J, Zabel B, Superti-Furga A. Dominant negative mutations in the C-propeptide of COL2A1 cause platyspondylic lethal skeletal dysplasia, torrance type, and define a novel subfamily within the type 2 collagenopathies. *Am J Med Genet* 133:61-67, 2005 (IF 2,3)
27. Kubisch C, Ketelsen UP, Goebel I, **Omran H**. Autosomal recessive rippling muscle disease with homozygous CAV3 mutations. *Annals Neurol* 57:303-304, 2005 (IF 11,2)
28. Otto EA, Loeys B, Khanna H, Hellemans J, Sudbrak R, Fan S, Muerb U, O'Toole JF, Helou J, Attanasio M, Utsch B, Sayer JA, Lillo C, Jimeno D, Coucke P, De Paepe A, Reinhardt R, Klages S, Tsuda M, Kawakami I, Kusakabe T, Omran H, Imm A, Tippens M, Raymond PA, Hill J, Beales P, He S, Kispert A, Margolis B, Williams DS, Swaroop A, Hildebrandt F. Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. *Nat Genet*. 37:282-8. 2005 (IF 35,2)
29. Fliegauf M., Olbrich H., Horvath J., Wildhaber J.H., Zariwala M.A., Kennedy M., Knowles M.R.,**Omran H**. Mis-localization of DNAH5 and DNAH9 in respiratory cells from primary ciliary dyskinesia patients. *Am J Respir Crit Care Med* 171:1343-1349, 2005 (IF 11)
30. Tacke U., Olbrich H., Sass J.O., Fekete A., Horvath J., Ziyeh S., Kleijer W.J., Rolland M.O.Payne S., Vargiami E., Zafeiriou D.I., **Omran H**. Possible genotype-phenotype correlations in children with mild clinical course of Canavan disease. *Neuropediatrics* 36:252-255, 2005 (IF 1,2)
31. Ketelsen UP, Brand-Saberi B., Uhlenberg B., Wagner M, Laberke HG, **Omran H**. Congenital myopathy with arrest of myogenesis prior to formation of myotubes. *Neuropediatrics* 36:246-251,2005 (IF 1,2)
32. Horváth J., Olbrich H., Fliegauf M., Kispert A., King S.M., Mitchison H., Zariwala M.A, KnowlesM.R., Sudbrack R., Reinhardt R., **Omran H**. Identification of the human axonemal dynein light chain 1 (DNAL1) gene and candidate gene analysis in patients with primary ciliary dyskinesia. *Am J Resp Cell Mol Biol* 33:41-47, 2005 (IF 4,1)
33. *Schermer B., *Hopker K., ***Omran H.**,Ghenoiu C., Fliegauf M., Fekete A., Horvath J., KottgenM., Hackl M., Zschiedrich S., Huber T.B., Kramer-Zucker A., Zentgraf H., Blaukat A., Walz G.,Benzing T. Phosphorylation by casein kinase 2 induces PACS-1 binding of nephrocystin and targeting to cilia. *EMBO J* 24:4415-24, 2005 (*shared first co-authorship) (IF 9,8)

34. Vougioukas V.I., **Omran H**., Gläsker S., van Velthoven V. Far lateral supracerebellarinfratentorial approach for the treatment of pontomesencephalic gliomas: clinical experience withpediatric patients. *Childs Nerv Syst* 22:1149-53, 2006 (IF 1,2)
35. Olbrich H., Horváth J., Fekete A., Loges N.T., Storm van's Gravesande K., Blum A., Hörmann K.,**Omran H**. Axonemal localization of the dynein component DNAH5 is not altered in secondary ciliary dyskinesia. *Ped. Research* 59:418-22, 2006 (IF 2,7)
36. Sass J.O., Mohr V., Olbrich H., Engelke U., Horvath J., Fliegauf M., Loges N.T., Schweitzer-Krantz S., Moebus R., Weiler P., Kispert A., Superti-Furga A., Wevers R.A., **Omran H.** Mutations in *ACY1*, the gene encoding aminoacylase 1, cause a novel inborn error of metabolism. *Am J Hum Genet* 78:401-9, 2006 (IF 11,2)
37. Mills P.B., Struys E., Jakobs C., Plecko B., Baxter P., Baumgartner M., Willemsen M.A., **Omran H.**, Tacke U., Uhlenberg B., Weschke B., Clayton P.T. Mutations in antiquitin in individuals with pyridoxine-dependent seizures. *Nat Med.* 12:307-9, 2006 (IF 22,8)
38. Hornef N, Olbrich H, Horvath J, Zariwala MA, Fliegauf M, Loges NT, Wildhaber J, Noone PG,Kennedy M, Antonarakis SE, Blouin JL, Bartoloni L, Nublein T, Ahrens P, Griese M, Kuhl H,Sudbrak R, Knowles MR, Reinhardt R, **Omran H**. *DNAH5* Mutations are a Common Cause of Primary Ciliary Dyskinesia with Outer Dynein Arm Defects. *Am J Respir Crit Care Med*. 174:120-126; 2006 (IF 11)
39. Budny B, Chen W, **Omran H**, Fliegauf M, Tzschach A, Wisniewska M, Jensen LR, Raynaud M,Shoichet SA, Badura M, Lenzner S, Latos-Bielenska A, Ropers HH. A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oralfacial-digital type I syndrome. *Hum Genet.* 120:171-8, 2006 (IF 5,0)
40. Zariwala MA, Leigh MW, Ceppa F, Kennedy MP, Noone PG, Carson JL, Hazucha MJ, Lori A,Horvath J, Olbrich H, Loges NT, Bridoux AM, Pennarun G, Duriez B, Escudier E, Mitchison HM, Chodhari R, Chung EM, Morgan LC, de Iongh RU, Rutland J, Pradal U, **Omran H**, Amselem S,Knowles MR. Mutations of DNAI1 in Primary Ciliary Dyskinesia: Evidence of Founder Effect in a Common Mutation. *Am J Respir Crit Care Med*. 174:858-66, 2006 (IF 11)
41. Fliegauf M, Horvath J, von Schnakenburg C, Olbrich H, Muller D, Thumfart J, Schermer B,Pazour GJ, Neumann HP, Zentgraf H, Benzing T, **Omran H**. Nephrocystin specifically localizes to the transition zone of renal and respiratory cilia and photoreceptor connecting cilia. *J Am SocNephrol.* 17:2424-33, 2006 (IF 9,0)
42. Schermer B, Ghenoiu C, Bartram M, Muller RU, Kotsis F, Hohne M, Kuhn W, Rapka M, NitschkeR, Zentgraf H, Fliegauf M, **Omran H**, Walz G, Benzing T. The von Hippel-Lindau tumor suppressor protein controls ciliogenesis by orienting microtubule growth. *J Cell Biol.* 175:547-54,2006 (IF 10,8)
43. Liebau MC, Gal A, Superti-Furga A, **Omran H**, Pohl M. L1CAM mutation in a boy with hydrocephalus and duplex kidneys. *Pediatr Nephrol.* 22:1058-61, 2007 (IF 2,9)
44. Driller K, Pagenstecher A, Uhl M, **Omran H**, Berlis A, Grunder A, Sippel AE. Nuclear Factor I X deficiency causes brain malformation and severe skeletal defects. *Mol Cell Biol*. 27:3855-3867,2007 (IF 5,4)
45. Kennedy MP, **Omran H**, Leigh MW, Dell S, Morgan L, Molina PL, Robinson BV, Minnix SL, Olbrich H, Severin T, Ahrens P, Lange L, Morillas HN, Noone PG, Zariwala MA, Knowles MR. Congenital Heart Disease and other Heterotaxic Defects in a Large Cohort of Patients with Primary Ciliary Dyskinesia. *Circulation* 115:2814-21, 2007 (IF 15,2)
46. Engelke UF, Sass JO, Van Coster RN, Gerlo E, Olbrich H, Krywawych S, Calvin J, Hart C, **Omran H**, Wevers RA. NMR spectroscopy of aminoacylase 1 deficiency, a novel inborn error of metabolism. *NMR Biomed*. 21:138-47. 2008 (IF 3,4)
47. Janzarik WG, Kratz CP, Loges NT, Olbrich H, Klein C, Schafer T, Scheurlen W, Roggendorf W, Weiller C, Niemeyer C, Korinthenberg R, Pfister S, **Omran H**. Further Evidence for a Somatic KRAS Mutation in a Pilocytic Astrocytoma. *Neuropediatrics*. 38: 61-3, 2007 (IF 1,2)
48. Sass JO, Olbrich H, Mohr V, Hart C, Woldseth B, Krywawych S, Bjurulf B, Lakhani PK, Buchdahl RM, **Omran H**. Neurological findings in aminoacylase 1 deficiency. *Neurology* 68:2151-3, 2007 (IF 8,2)
49. Helou J, Otto EA, Attanasio M, Allen SJ, Parisi M, Glass I, Utsch B, Hashmi S, Fazzi E, **Omran H**, O' Toole J, Sayer J, Hildebrandt F. Mutation analysis of NPHP6/CEP290 in patients with Joubert-Syndrome and Senior-Loken-Syndrome. *J Med Genet*. 44:657-63, 2007 (IF 5,7)

50. Tan SY, Rosenthal J, Zhao XQ, Francis RJ, Chatterjee B, Sabol SL, Linask KL, Bracero L, Connelly PS, Daniels MP, Yu Q, **Omran H**, Leatherbury L, Lo CW. Heterotaxy and complex structural heart defects in a mutant mouse model of primary ciliary dyskinesia. *J Clin Invest*. 117:3742-3752, 2007 (IF 12,8)
51. Schwabe GC, Hoffmann K, Loges NT, Birker D, Rossier C, de Santi MM, Olbrich H, Fliegauf M,Failly M, Liebers U, Collura M, Gaedicke G, Mundlos S, Wahn U, Blouin JL, Niggemann B, **Omran H**, Antonarakis SE, Bartoloni L. Primary ciliary dyskinesia associated with normal axoneme ultrastructure is caused by DNAH11 Mutations. *Hum Mutat*. 29:289-98, 2008 (IF 5,2)
52. Bergmann C, Fliegauf M, Brüchle NO, Frank V, Olbrich H, Kirschner J, Schermer B, SchmeddingI, Kispert A, Kränzlin B, Nürnberg G, Becker C, Grimm T, Girschick G, Lynch SA, Kelehan P,Senderek J, Neuhaus TJ, Stallmach T, Zentgraf H, Nürnberg P, Gretz N, Lo C, Lienkamp S,Schäfer T, Walz G, Benzing T, Zerres K, **Omran H**. Loss of nephrocystin-3 function can cause embryonic lethality, Meckel-Gruber-like syndrome, situs inversus, and renal-hepatic-pancreatic dysplasia. *Am J Hum Genet*. 82:959-70, 2008 (IF 11,2)
53. Pfister S, Janzarik WG, Remke M, Ernst A, Werft W, Becker N, Toedt G, Wittmann A, Kratz C, Olbrich H, Ahmadi R, Thieme B, Joos S, Radlwimmer B, Kulozik A, Pietsch T, Herold-Mende C, Gnekow A, Reifenberger G, Korshunov A, Scheurlen W, **Omran H**, Lichter P. BRAF gene duplication constitutes a mechanism of MAPK pathway activation in low-grade astrocytomas. *J Clin Invest*. 118:1739-49, 2008 (IF 12,8)
54. Wessels MW, Avital A, Failly M, Munoz A, **Omran H**, Blouin JL, Willems PJ. Candidate gene analysis in three families with acilia syndrome. Am J Med Genet A. 146:1765-7, 2008 (IF 2,3)
55. Sass JO, Jobard F, Topçu M, Mahfoud A, Werlé E, Cure S, Al-Sannaa N, Alshahwan SA, Bataillard M, Cimbalistiene L, Grolik C, Kemmerich V, **Omran H**, Sztriha L, Tabache M, Fischer J. L: -2-Hydroxyglutaric aciduria: Identification of ten novel mutations in the L2HGDH gene. *J Inherit Metab Dis*. Short Report 31(2):S275-279, 2008 (IF 4,0)
56. Morris-Rosendahl DJ, Najm J, Lachmeijer AM, Sztriha L, Martins M, Kuechler A, Haug V, Zeschnigk C, Martin P, Santos M, Vasconcelos C, **Omran H**, Kraus U, Van der Knaap MS, Schuierer G, Kutsche K, Uyanik G. Refining the phenotype of alpha-1a Tubulin (TUBA1A) mutation in patients with classical lissencephaly. Clin Genet. 74:425-33, 2008 (IF 3,9)
57. Loges NT, Olbrich H, Fenske L, Mussaffi H, Horvath J, Fliegauf M, Kuhl H, Baktai G, Peterffy E,Chodhari R, Chung EM, Rutman A, O'Callaghan C, Blau H, Tiszlavicz L, Voelkel K, Witt M, Zietkiewicz E, Neesen J, Reinhardt R, Mitchison HM, **Omran H**. DNAI2 mutations cause primary ciliary dyskinesia with defects in the outer dynein arm. *Am J Hum Genet*. 83:547-58, 2008 (IF11,2)
58. **Omran H**, Kobayashi D, Olbrich H, Tsukahara T, Loges NT, Hagiwara H, Zhang Q, Leblond G,O'Toole E, Hara C, Mizuno H, Kawano H, Fliegauf M, Yagi T, Koshida S, Miyawaki A, Zentgraf H, Seithe H, Reinhardt R, Watanabe Y, Kamiya R, Mitchell DR, Takeda H. Ktu/PF13 is required for cytoplasmic pre-assembly of axonemal dyneins. *Nature* 456:611-6, 2008 (IF 38,5)
59. Francis RJ, Chatterjee B, Loges NT, Zentgraf H, **Omran H**, Lo CW. The initiation and maturation of cilia generated flow in the newborn and postnatal mouse airway. Am J Physiol Lung Cell Mol Physiol. 296(6):L1067-1075, 2009 (IF 3,7)
60. Stölting T, **Omran H**, Erlekotte A, Denecke J, Reunert J, Marquardt T. Novel ALG8 mutations expand the clinical spectrum of congenital disorder of glycosylation type Ih. *Mol Genet Metab*. 98:305-9, 2009 (IF 2,8)
61. Osten L, Kubitza M, Gallagher AR, Kastner J, Olbrich H, de Vries U, Kees F, Lelongt B, Somlo S, **Omran H**, Witzgall R. Doxycycline accelerates renal cyst growth and fibrosis in the pcy/pcy mouse model of type 3 nephronophthisis, a form of recessive polycystic kidney disease. *Histochem Cell Biol*. 132:199-210, 2009 (IF 2,6)
62. Loges NT, Olbrich H, Becker-Heck A, Häffner K, Heer A, Reinhard C, Schmidts M, Kispert A, Zariwala MA, Leigh MW, Knowles MR, Zentgraf H, Seithe H, Nürnberg G, Nürnberg P, Reinhardt R, **Omran H**. Deletions and point mutations of LRRC50 cause primary ciliary dyskinesia due to dynein arm defects. *Am J Hum Genet*. 85:883-9, 2009 (11,2)
63. Jurk K, Schulz AS, Kehrel BE, Räpple D, Schulze H, Möbest D, Friedrich WW, **Omran H**, Deak E, Henschler R, Scheele JS, Zieger B. Novel integrin-dependent platelet malfunction in siblings with leukocyte adhesion deficiency-III (LAD-III) caused by a point mutation in FERMT3. *Thromb Haemost*. 103:1053-64, 2010 (IF 6,0)

64. Valstar MJ, Bertoli-Avella AM, Wessels MW, Ruijter GJ, de Graaf B, Olmer R, Elfferich P, NeijsS, Kariminejad R, Suheyl Ezgü F, Tokatli A, Czartoryska B, Bosschaart AN, van den Bos-Terpstra F, Puissant H, Bürger F, **Omran H**, Eckert D, Filocamo M, Simeonov E, Willems PJ,Wevers RA, Niermeijer MF, Halley DJ, Poorthuis BJ, van Diggelen OP. Mucopolysaccharidosistype IIID: 12 new patients and 15 novel mutations. *Hum Mutat*. 31E1348-60. 2010 (IF 5,2)
65. Tylki-Szymanska A, Gradowska W, Sommer A, Heer A, Walter M, Reinhard C, **Omran H**, SassJO, Jurecka A. Aminoacylase 1 deficiency associated with autistic behavior. *J Inherit Metab Dis*.33(3):211-214, 2010 (IF 4,0)
66. Kuehni CE, Frischer T, Strippoli MP, Maurer E, Bush A, Nielsen KG, Escribano A, Lucas JS,Yiallouros P, **Omran H**, Eber E, O'Callaghan C, Snijders D, Barbato A; ERS Task Force on Primary Ciliary Dyskinesia in Children. Factors influencing age at diagnosis of primary ciliary dyskinesia in European children. *Eur Respir J*. 36(6):1248-58, 2010 (IF 6,3)
67. **Omran H,** Olbrich H. Zilienkrankheiten unter besonderer Berücksichtigung der Primären Ziliären Dyskinesie*. Medizinische Genetik* 2010, 22(3):315-321
68. Eisenhardt AE, Olbrich H, Röring M, Janzarik W, Van Anh TN, Cin H, Remke M, Witt H, Korshunov A, Pfister SM, Omran H, Brummer T. Functional characterization of a BRAF insertion mutant associated with pilocytic astrocytoma. *Int J Cancer*. 129(9):2297-2303, 2011 (IF 6,2)
69. Berg JS, Evans JP, Leigh MW, **Omran H**, Bizon C, Mane K, Knowles MR, Weck KE, Zariwala MA. Next generation massively parallel sequencing of targeted exomes to identify genetic mutations in primary ciliary dyskinesia: implications for application to clinical testing. *Genet Med*.13:218-29, 2011 (IF 5,6)
70. Schindler G, Capper D, Meyer J, Janzarik W, **Omran H**, Herold-Mende C, Schmieder K, Wesseling P, Mawrin C, Hasselblatt M, Louis DN, Korshunov A, Pfister S, Hartmann C, Paulus W, Reifenberger G, von Deimling A. Analysis of BRAF V600E mutation in 1,320 nervous system tumors reveals high mutation frequencies in pleomorphic xanthoastrocytoma, ganglioglioma and extra-cerebellar pilocytic astrocytoma. *Acta Neuropathol*. 121:397-405, 2011 (IF 9,7)
71. Sommer JU, Schäfer K, **Omran H**, Olbrich H, Wallmeier J, Blum A, Hörmann K, Stuck BA. ENT manifestations in patients with primary ciliary dyskinesia: prevalence and significance of otorhinolaryngologic co-morbidities. *Eur Arch Otorhinolaryngol*. 268:383-8, 2011 (IF 1,4)
72. Merveille AC, Davis EE, Becker-Heck A, Legendre M, Amirav I, Bataille G, Belmont J, Beydon N, Billen F, Clément A, Clercx C, Coste A, Crosbie R, de Blic J, Deleuze S, Duquesnoy P, Escalier D, Escudier E, Fliegauf M, Horvath J, Hill K, Jorissen M, Just J, Kispert A, Lathrop M, Loges NT, Marthin JK, Momozawa Y, Montantin G, Nielsen KG, Olbrich H, Papon JF, Rayet I, Roger G, Schmidts M, Tenreiro H, Towbin JA, Zelenika D, Zentgraf H, Georges M, Lequarré AS, Katsanis N, **Omran H**, Amselem S. CCDC39 is required for assembly of inner dynein arms and the dynein regulatory complex and for normal ciliary motility in humans and dogs. *Nat Genet*. 43(1):72-78,2011 (IF 35,2)
73. Becker-Heck A, Zohn IE, Okabe N, Pollock A, Lenhart KB, Sullivan-Brown J, McSheene J, LogesNT, Olbrich H, Haeffner K, Fliegauf M, Horvath J, Reinhardt R, Nielsen KG, Marthin JK, Baktai G, Anderson KV, Geisler R, Niswander L, **Omran H**, Burdine RD. The coiled-coil domain containing protein CCDC40 is essential for motile cilia function and left-right axis formation. *Nat Genet.* 43:79-84, 2011 (IF 35,2)
74. Cin H, Meyer C, Herr R, Janzarik WG, Lambert S, Jones DT, Jacob K, Benner A, Witt H, Remke M, Bender S, Falkenstein F, Van Anh TN, Olbrich H, von Deimling A, Pekrun A, Kulozik AE, Gnekow A, Scheurlen W, Witt O, **Omran H**, Jabado N, Collins VP, Brummer T, Marschalek R, Lichter P, Korshunov A, Pfister SM. Oncogenic FAM131B-BRAF fusion resulting from 7q34 deletion comprises an alternative mechanism of MAPK pathway activation in pilocytic astrocytoma. *Acta Neuropathol.* 121(6):763-74, 2011 (IF 9,7)
75. Sommer A, Christensen E, Schwenger S, Seul R, Haas D, Olbrich H, **Omran H**, Sass JO. The molecular basis of aminoacylase 1 deficiency. *Biochim Biophys Acta.* 1812:685-90, 2011 (IF 4,6)
76. Schmid F, Glaus E, Barthelmes D, Fliegauf M, Gaspar H, Nürnberg G, Nürnberg P, **Omran H**, Berger W, Neidhardt J. U1 snRNA-mediated gene therapeutic correction of splice defects caused by an exceptionally mild BBS mutation. *Hum Mutat.* 32:815-24, 2011 (IF 5,2)
77. Röseler S, Sandrock K, Bartsch I, Busse A, **Omran H**, Loges NT, Zieger B. Lethal phenotype of mice carrying a Sept11 null mutation. *Biol Chem.* 392:779-81, 2011 (IF 2,7)
78. Zariwala MA, **Omran H**, Ferkol TW. The emerging genetics of primary ciliary dyskinesia. *Proc Am Thorac Soc.* 8:430-3, 2011

79. Knowles MR, Leigh MW, Carson JL, Davis SD, Dell SD, Ferkol TW, Olivier KN, Sagel SD, Rosenfeld M, Burns KA, Minnix SL, Armstrong MC, Lori A, Hazucha MJ, Loges NT, Olbrich H, Becker-Heck A, Schmidts M, Werner C, **Omran H**, Zariwala MA. Genetic Disorders of Mucociliary Clearance Consortium. Mutations of DNAH11 in patients with primary ciliary dyskinesia with normal ciliary ultrastructure. *Thorax.* 67:433-41, 2012 (IF 8,3)
80. Mitchison HM, Schmidts M, Loges NT, Freshour J, Dritsoula A, Hirst RA, O'Callaghan C, Blau H, Al Dabbagh M, Olbrich H, Beales PL, Yagi T, Mussaffi H, Chung EM, **Omran H**, Mitchell DR. Mutations in axonemal dynein assembly factor DNAAF3 cause primary ciliary dyskinesia. *Nat Genet*. 44:381-9, 2012 (IF 35,2)
81. Nakhleh N, Francis R, Giese RA, Tian X, Li Y, Zariwala MA, Yagi H, Khalifa O, Kureshi S, Chatterjee B, Sabol SL, Swisher M, Connelly PS, Daniels MP, Srinivasan A, Kuehl K, Kravitz N, Burns K, Sami I, **Omran H**, Barmada M, Olivier K, Chawla KK, Leigh M, Jonas R, Knowles M, Leatherbury L, Lo CW. High prevalence of respiratory ciliary dysfunction in congenital heart disease patients with heterotaxy. *Circulation*. 2232-42, 2012 (IF 15,2)
82. Panizzi JR, Becker-Heck A, Castleman VH, Al-Mutairi DA, Liu Y, Loges NT, Pathak N, Austin-Tse C, Sheridan E, Schmidts M, Olbrich H, Werner C, Häffner K, Hellman N, Chodhari R, Gupta A, Kramer-Zucker A, Olale F, Burdine RD, Schier AF, O'Callaghan C, Chung EM, Reinhardt R, Mitchison HM, King SM, **Omran H**, Drummond IA. CCDC103 mutations cause primary ciliary dyskinesia by disrupting assembly of ciliary dynein arms. *Nat Genet*. 44:714-9, 2012 (IF 35,2)
83. Chaki M, Airik R, Ghosh AK, Giles RH, Chen R, Slaats GG, Wang H, Hurd TW, Zhou W, Cluckey A, Gee HY, Ramaswami G, Hong CJ, Hamilton BA, Cervenka I, Ganji RS, Bryja V, Arts HH, vanReeuwijk J, Oud MM, Letteboer SJ, Roepman R, Husson H, Ibraghimov-Beskrovnaya O, Yasunaga T, Walz G, Eley L, Sayer JA, Schermer B, Liebau MC, Benzing T, Le Corre S, Drummond I, Janssen S, Allen SJ, Natarajan S, O'Toole JF, Attanasio M, Saunier S, Antignac C, Koenekoop RK, Ren H, Lopez I, Nayir A, Stoetzel C, Dollfus H, Massoudi R, Gleeson JG, Andreoli SP, Doherty DG, Lindstrad A, Golzio C, Katsanis N, Pape L, Abboud EB, Al-Rajhi AA, Lewis RA, **Omran H**, Lee EY, Wang S, Sekiguchi JM, Saunders R, Johnson CA, Garner E, Vanselow K, Andersen JS, Shlomai J, Nurnberg G, Nurnberg P, Levy S, Smogorzewska A, Otto EA, Hildebrandt F. Exome capture reveals ZNF423 and CEP164 mutations, linking renal ciliopathies to DNA damage response signaling. *Cell*. 150:533-48. 2012 (IF 31,9)
84. Bukowy-Bieryłło Z, Ziętkiewicz E, Loges NT, Wittmer M, Geremek M, Olbrich H, Fliegauf M, Voelkel K, Rutkiewicz E, Rutland J, Morgan L, Pogorzelski A, Martin J, Haan E, Berger W, **Omran H**, Witt M. RPGR mutations might cause reduced orientation of respiratory cilia. *Pediatr Pulmonol*. 48:352-363. 2013 (IF 2,4)
85. Schaumburg F, Schmalstieg C, Fiedler B, Brentrup A, **Omran H**, Becker K. A bumpy road to the diagnosis of a Kytococcus schroeteri shunt infection. *J Med Microbiol*. 62:165-168, 2013 (IF 2,29)
86. Olbrich H, Schmidts M, Werner C, Onoufriadis A, Loges NT, Raidt J, Banki NF, Shoemark A,Burgoyne T, Al Turki S, Hurles ME; UK10K Consortium, Köhler G, Schroeder J, Nürnberg G, Nürnberg P, Chung EM, Reinhardt R, Marthin JK, Nielsen KG, Mitchison HM, **Omran H**. Recessive HYDIN Mutations Cause Primary Ciliary Dyskinesia without Randomization of Left-Right Body Asymmetry. *Am J Hum Genet*. 91(4):672-84. 2012 (IF 11,2)
87. Halbritter J, Diaz K, Chaki M, Porath JD, Tarrier B, Fu C, Innis JL, Allen SJ, Lyons RH, StefanidisCJ, **Omran H**, Soliman NA, Otto EA. High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. *J Med Genet.* 49(12):756-767. 2012 (IF 5,7)
88. Antony D, Becker-Heck A, Zariwala MA, Schmidts M, Onoufriadis A, Forouhan M, Wilson R,Taylor-Cox T, Dewar A, Jackson C, Goggin P, Loges NT, Olbrich H, Jaspers M, Jorissen M, Leigh MW, Wolf WE, Daniels ML, Noone PG, Ferkol TW, Sagel SD, Rosenfeld M, Rutman A, Dixit A, O'Callaghan C, Lucas JS, Hogg C, Scambler PJ, Emes RD, Uk10k, Chung EM, Shoemark A, Knowles MR, **Omran H**, Mitchison HM. Mutations in CCDC39 and CCDC40 are the major cause of primary ciliary dyskinesia with axonemal disorganisation and absent inner dyneinarms. *Hum Mutat*. 34:462-472. 2013 (IF 5,2)
89. Wirschell M, Olbrich H, Werner C, Tritschler D, Bower R, Sale WS, Loges NT, Pennekamp P, Lindberg S, Stenram U, Carlén B, Horak E, Köhler G, Nürnberg P, Nürnberg G, Porter ME, **Omran H**. The nexin-dynein regulatory complex subunit DRC1 is essential for motile cilia function in algae and humans. *Nat Genet*. 45:262-268. 2013 (IF 35,2)

90. Tarkar A, Loges NT, Slagle CE, Francis R, Dougherty GW, Tamayo JV, Shook B, Cantino M,Schwartz D, Jahnke C, Olbrich H, Werner C, Raidt J, Pennekamp P, Abouhamed M, Hjeij R,Köhler G, Griese M, Li Y, Lemke K, Klena N, Liu X, Gabriel G, Tobita K, Jaspers M, Morgan LC,Shapiro AJ, Letteboer SJ, Mans DA, Carson JL, Leigh MW, Wolf WE, Chen S, Lucas JS, Onoufriadis A, Plagnol V, Schmidts M, Boldt K; UK10K, Roepman R, Zariwala MA, Lo CW, Mitchison HM, Knowles MR, Burdine RD, Loturco JJ, **Omran H**. DYX1C1 is required for axonemal dynein assembly and ciliary motility. *Nat Genet*. 45:995-1003. 2013. (IF 35,2)
91. Hjeij R, Lindstrand A, Francis R, Zariwala MA, Liu X, Li Y, Damerla R, Dougherty GW, Abouhamed M, Olbrich H, Loges NT, Pennekamp P, Davis EE, Carvalho CM, Pehlivan D, Werner C, Raidt J, Köhler G, Häffner K, Reyes-Mugica M, Lupski JR, Leigh MW, Rosenfeld M, Morgan LC, Knowles MR, Lo CW, Katsanis N, **Omran H**. ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. *Am J **Hum G**enet*. 2013,93(2):357-367. (IF 11,2)
92. Zariwala MA, Gee HY, Kurkowiak M, Al-Mutairi DA, Leigh MW, Hurd TW, Hjeij R, Dell SD, Chaki M, Dougherty GW, Adan M, Spear PC, Esteve-Rudd J, Loges NT, Rosenfeld M, Diaz KA, Olbrich H, Wolf WE, Sheridan E, Batten TF, Halbritter J, Porath JD, Kohl S, Lovric S, Hwang DY, Pittman JE, Burns KA, Ferkol TW, Sagel SD, Olivier KN, Morgan LC, Werner C, Raidt J, Pennekamp P, Sun Z, Zhou W, Airik R, Natarajan S, Allen SJ, Amirav I, Wieczorek D, Landwehr K, Nielsen K, Schwerk N, Sertic J, Köhler G, Washburn J, Levy S, Fan S, Koerner-Rettberg C, Amselem S, Williams DS, Mitchell BJ, Drummond IA, Otto EA, **Omran H**, Knowles MR, Hildebrandt F. ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. *Am J Hum Genet.* 2013, 93(2):336-345. (IF 11,2)
93. Knowles MR, Ostrowski LE, Loges NT, Hurd T, Leigh MW, Huang L, Wolf WE, Carson JL, Hazucha MJ, Yin W, Davis SD, Dell SD, Ferkol TW, Sagel SD, Olivier KN, Jahnke C, Olbrich H, Werner C, Raidt J, Wallmeier J, Pennekamp P, Dougherty GW, Hjeij R, Gee HY, Otto EA, Halbritter J, Chaki M, Diaz KA, Braun DA, Porath JD, Schueler M, Baktai G, Griese M, Turner EH, Lewis AP, Bamshad MJ, Nickerson DA, Hildebrandt F, Shendure J, **Omran H**, Zariwala MA. Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. *Am J Hum Genet*. 2013 93(4):711-720. (IF 11,2)
94. Austin-Tse C, Halbritter J, Zariwala MA, Gilberti RM, Gee HY, Hellman N, Pathak N, Liu Y,Panizzi JR, Patel-King RS, Tritschler D, Bower R, O'Toole E, Porath JD, Hurd TW, Chaki M, Diaz KA, Kohl S, Lovric S, Hwang DY, Braun DA, Schueler M, Airik R, Otto EA, Leigh MW, Noone PG, Carson JL, Davis SD, Pittman JE, Ferkol TW, Atkinson JJ, Olivier KN, Sagel SD, Dell SD, Rosenfeld M, Milla CE, Loges NT, **Omran H**, Porter ME, King SM, Knowles MR, Drummond IA, Hildebrandt F. Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. *Am J Hum Genet*. 2013; 93(4):672-686. (IF 11,2)
95. Wallmeier J, Al-Mutairi DA, Chen CT, Loges NT, Pennekamp P, Menchen T, Ma L, Shamseldin HE, Olbrich H, Dougherty GW, Werner C, Alsabah BH, Köhler G, Jaspers M, Boon M, Griese M, Schmitt-Grohé S, Zimmermann T, Koerner-Rettberg C, Horak E, Kintner C, Alkuraya FS, Omran H. Mutations in CCNO result in congenital mucociliary clearance disorder with reduced generation of multiple motile cilia. *Nat Genet*. 2014; 46(6):646-51.
96. Ma L, Quigley I, **Omran H**, Kintner C. Multicilin drives centriole biogenesis via E2f proteins. *Genes Dev*. 2014; 1;28(13):1461-71.
97. Boon M, Wallmeier J, Ma L, Loges NT, Jaspers M, Olbrich H, Dougherty GW, Raidt J, Werner C, Amirav I, Hevroni A, Abitbul R, Avital A, Soferman R, Wessels M, O'Callaghan C, Chung EM, Rutman A, Hirst RA, Moya E, Mitchison HM, Van Daele S, De Boeck K, Jorissen M, Kintner C, Cuppens H, Omran H. MCIDAS mutations result in a mucociliary clearance disorder with reduced generation of multiple motile cilia. *Nat Commun*. 2014; 5:4418
98. Raidt J, Wallmeier J, Hjeij R, Onnebrink JG, Pennekamp P, Loges NT, Olbrich H, Häffner K, Dougherty GW, **Omran H**, Werner C. Ciliary beat pattern and frequency in genetic variants of primary ciliary dyskinesia. *Eur Respir J*. 2014;44(6):1579-88.
99. Hjeij R, Onoufriadis A, Watson CM, Slagle CE, Klena NT, Dougherty GW, Kurkowiak M, Loges NT, Diggle CP, Morante NF, Gabriel GC, Lemke KL, Li Y, Pennekamp P, Menchen T, Konert F, Marthin JK, Mans DA, Letteboer SJ, Werner C, Burgoyne T, Westermann C, Rutman A, Carr IM, O'Callaghan C, Moya E, Chung EM; UK10K Consortium, Sheridan E, Nielsen KG, Roepman R, Bartscherer K, Burdine RD, Lo

CW, **Omran H**, Mitchison HM. CCDC151 mutations cause primary ciliary dyskinesia by disruption of the outer dynein arm docking complex formation. *Am J Hum Genet*. 2014; 95(3):257-74.

100. Narasimhan V, Hjeij R, Vij S, Loges NT, Wallmeier J, Koerner-Rettberg C, Werner C, Thamilselvam SK, Boey A, Choksi S, Pennekamp P, Roy S, **Omran H.** Mutations in CCDC11, Which Encodes a Coiled-coil Containing Ciliary Protein, Causes situs inversus Due to Dysmotility of Monocilia in the Left-Right Organizer. *Hum Mutat*. 2014
101. Epting D, Slanchev K, Boehlke C, Hoff S, Loges NT, Yasunaga T, Indorf L, Nestel S, Lienkamp SS, **Omran H**, Kuehn EW, Ronneberger O, Walz G, Kramer-Zucker A. The Rac1 regulator ELMO controls basal body migration and docking in multiciliated cells through interaction with Ezrin. *Development.* 2015; 142:174-184.
102. Werner C, Onnebrink JG, **Omran H.** Diagnosis and management of primary ciliary dyskinesia. *Cilia.* 2015; 2. doi: 10.1186/s13630-014-0011-8. eCollection 2015.
103. Funk MC, Bera AN, Menchen T, Kuales G, Thriene K, Lienkamp SS, Dengjel J, **Omran H**, Frank M, Arnold SJ. Cyclin O (Ccno) functions during deuterosome-mediated centriole amplification of multiciliated cells. *EMBO J*. 2015 Feb 23. pii: e201490805.
104. Amirav I, Mussaffi H, Roth Y, Schmidts M, **Omran H**, Werner C; Israeli PCD Consortium Investigators. A reach-out system for video microscopy analysis of ciliary motions aiding PCD diagnosis. *BMC Res Notes*. 2015 Dec;8(1):999. doi: 10.1186/s13104-015-0999-x. Epub 2015 Mar 8.
105. Frommer A, Hjeij R, Loges NT, Edelbusch C, Jahnke C, Raidt J, Werner C, Wallmeier J, Große-Onnebrink J, Olbrich H, Cindrić S, Jaspers M, Boon M, Memari Y, Durbin R, Kolb-Kokocinski A, Sauer S, Marthin JK, Nielsen KG, Amirav I, Elias N, Eitan K, Shoseyov D, Haeffner K, **Omran H.** Immunofluorescence Analysis and Diagnosis of Primary Ciliary Dyskinesia with Radial Spoke Defects. *Am J Respir Cell Mol Biol*. 2015 Mar 19.
106. Wheway G, Schmidts M, Mans DA, Szymanska K, Nguyen TM, Racher H, Phelps IG, Toedt G, Kennedy J, Wunderlich KA, Sorusch N, Abdelhamed ZA, Natarajan S, Herridge W, van Reeuwijk J, Horn N, Boldt K, Parry DA, Letteboer SJ, Roosing S, Adams M, Bell SM, Bond J, Higgins J, Morrison EE, Tomlinson DC, Slaats GG, van Dam TJ, Huang L, Kessler K, Giessl A, Logan CV, Boyle EA, Shendure J, Anazi S, Aldahmesh M, Al Hazzaa S, Hegele RA, Ober C, Frosk P, Mhanni AA, Chodirker BN, Chudley AE, Lamont R, Bernier FP, Beaulieu CL, Gordon P, Pon RT, Donahue C, Barkovich AJ, Wolf L, Toomes C, Thiel CT, Boycott KM, McKibbin M, Inglehearn CF; UK10K Consortium; University of Washington Center for Mendelian Genomics, Stewart F, **Omran H**, Huynen MA, Sergouniotis PI, Alkuraya FS, Parboosingh JS, Innes AM, Willoughby CE, Giles RH, Webster AR, Ueffing M, Blacque O, Gleeson JG, Wolfrum U, Beales PL, Gibson T, Doherty D, Mitchison HM, Roepman R, Johnson CA. An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. *Nat Cell Biol.* 2015 17(8):1074-87.
107. Raidt J, Werner C, Menchen T, Dougherty GW, Olbrich H, Loges NT, Schmitz R, Pennekamp P, **Omran H.** Ciliary function and motor protein composition of human fallopian tubes. *Hum Reprod*. 2015, 30:2871-2880.
108. Olbrich H, Cremers C, Loges NT, Werner C, Nielsen KG, Marthin JK, Philipsen M, Wallmeier J, Pennekamp P, Menchen T, Edelbusch C, Dougherty GW, Schwartz O, Thiele H, Altmüller J, Rommelmann F, **Omran H**. Loss-of-Function GAS8 Mutations Cause Primary Ciliary Dyskinesia and Disrupt the Nexin-Dynein Regulatory Complex. *Am J Hum Genet.* 2015, 97:546-554.
109. Werner, C. Titieni, A. **Omran, H**. Hochfrequenzvideo-mikroskopie zur Diagnostik der primären ziliären Dyskinesie. *Pädiatrische Praxis* 2015, 84(4):563-573 Park JH, Hogrebe M, Grüneberg M, DuChesne I, von der Heiden AL, Reunert J, Schlingmann KP, Boycott KM, Beaulieu CL, Mhanni AA, Innes AM, Hörtnagel K, Biskup S, Gleixner EM, Kurlemann G, Fiedler B, Omran H, Rutsch F, Wada Y, Tsiakas K, Santer R, Nebert DW, Rust S, Marquardt T. SLC39A8 Deficiency: A Disorder of Manganese Transport and Glycosylation. Am J Hum Genet. 2015 Dec 3;97(6):894-903. doi: 10.1016/j.ajhg.2015.11.003.
110. Werner C, Lablans M, Ataian M, Raidt J, Wallmeier J, Große-Onnebrink J, Kuehni CE, Haarman EG, Leigh MW, Quittner AL, Lucas JS, Hogg C, Witt M, Priftis KN, Yiallouros P, Nielsen KG, Santamaria F, Ückert F, **Omran H**. An international registry for primary ciliary dyskinesia. Eur Respir J. 2016 Mar;47(3):849-59. doi: 10.1183/13993003.00776-2015. Epub 2015 Dec 8.

111. Grosse-Onnebrink J, Werner C, Loges NT, Hörmann K, Blum A, Schmidt R, Olbrich H, **Omran H.** Effect of TH2 cytokines and interferon gamma on beat frequency of human respiratory cilia. Pediatr Res. 2016 May;79(5):731-5
112. Amirav I, Wallmeier J, Loges NT, Menchen T, Pennekamp P, Mussaffi H, Abitbul R, Avital A, Bentur L, Dougherty GW, Nael E, Lavie M, Olbrich H, Werner C, Kintner C, **Omran H;** Israeli PCD Consortium Investigators#. Systematic Analysis of CCNO Variants in a Defined Population: Implications for Clinical Phenotype and Differential Diagnosis. Hum Mutat. 2016 Apr; 37(4):396-405. doi: 10.1002/humu.22957. Epub 2016 Feb 4
113. Dougherty GW, Loges NT, Klinkenbusch JA, Olbrich H, Pennekamp P, Menchen T, Raidt J, Wallmeier J, Werner C, Westermann C, Ruckert C, Mirra V, Hjeij R, Memari Y, Durbin R, Kolb-Kokocinski A, Praveen K, Kashef MA, Kashef S, Eghtedari F, Häffner K, Valmari P, Baktai G, Aviram M, Bentur L, Amirav I, Davis EE, Katsanis N, Brueckner M, Shaposhnykov A, Pigino G, Dworniczak B, **Omran H**. DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Dynein Arm Complexes. Am J Respir Cell Mol Biol. 2016 Aug;55(2):213-24.
114. Boldt K, van Reeuwijk J, Lu Q, Koutroumpas K, Nguyen TM, Texier Y, van Beersum SE, Horn N, Willer JR, Mans DA, Dougherty G, Lamers IJ, Coene KL, Arts HH, Betts MJ, Beyer T, Bolat E, Gloeckner CJ, Haidari K, Hetterschijt L, Iaconis D, Jenkins D, Klose F, Knapp B, Latour B, Letteboer SJ, Marcelis CL, Mitic D, Morleo M, Oud MM, Riemersma M, Rix S, Terhal PA, Toedt G, van Dam TJ, de Vrieze E, Wissinger Y, Wu KM, Apic G, Beales PL, Blacque OE, Gibson TJ, Huynen MA, Katsanis N, Kremer H, **Omran H**, van Wijk E, Wolfrum U, Kepes F, Davis EE, Franco B, Giles RH, Ueffing M, Russell RB, Roepman R; UK10K Rare Diseases Group. An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nat Commun. 2016 May 13;7:11491. doi: 10.1038/ncomms11491.
115. Dau C, Fliegauf M, **Omran H**, Schlensog M, Dahl E, van Roeyen CR, Kriz W, Moeller MJ, Braun GS. The atypical cadherin Dachsous1 localizes to the base of the ciliary apparatus in airway epithelia. Biochem Biophys Res Commun. 2016 May 13;473(4):1177-84. doi: 10.1016/j.bbrc.2016.04.036. Epub 2016 Apr 10.
116. Wheway G, Schmidts M, Mans DA, Szymanska K, Nguyen TM, Racher H, Phelps IG, Toedt G, Kennedy J, Wunderlich KA, Sorusch N, Abdelhamed ZA, Natarajan S, Herridge W, van Reeuwijk J, Horn N, Boldt K, Parry DA, Letteboer SJ, Roosing S, Adams M, Bell SM, Bond J, Higgins J, Morrison EE, Tomlinson DC, Slaats GG, van Dam TJ, Huang L, Kessler K, Giessl A, Logan CV, Boyle EA, Shendure J, Anazi S, Aldahmesh M, Al Hazzaa S, Hegele RA, Ober C, Frosk P, Mhanni AA, Chodirker BN, Chudley AE, Lamont R, Bernier FP, Beaulieu CL, Gordon P, Pon RT, Donahue C, Barkovich AJ, Wolf L, Toomes C, Thiel CT, Boycott KM, McKibbin M, Inglehearn CF; UK10K Consortium; University of Washington Center for Mendelian Genomics, Stewart F, **Omran H**, Huynen MA, Sergouniotis PI, Alkuraya FS, Parboosingh JS, Innes AM, Willoughby CE, Giles RH, Webster AR, Ueffing M, Blacque O, Gleeson JG, Wolfrum U, Beales PL, Gibson T, Doherty D, Mitchison HM, Roepman R, Johnson CA. An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. Nat Cell Biol. 2015 Aug;17(8):1074-87.
117. Kobbernagel HE, Buchvald FF, Haarman EG, Casaulta C, Collins SA, Hogg C, Kuehni CE, Lucas JS, **Omran H**, Quittner AL, Werner C, Nielsen KG. Study protocol, rationale and recruitment in a European multi-centre randomized controlled trial to determine the efficacy and safety of azithromycin maintenance therapy for 6 months in primary ciliary dyskinesia. BMC Pulm Med. 2016 16:104.
118. Wallmeier J, Shiratori H, Dougherty GW, Edelbusch C, Hjeij R, Loges NT, Menchen T, Olbrich H, Pennekamp P, Raidt J, Werner C, Minegishi K, Shinohara K, Asai Y, Takaoka K, Lee C, Griese M, Memari Y, Durbin R, Kolb-Kokocinski A, Sauer S, Wallingford JB, Hamada H, **Omran H.** TTC25 Deficiency Results in Defects of the Outer Dynein Arm Docking Machinery and Primary Ciliary Dyskinesia with Left-Right Body Asymmetry Randomization. Am J Hum Genet. 2016, 99:460-9.
119. Abitbul R, Amirav I, Blau H, Alkrinawi S, Aviram M, Shoseyov D, Bentur L, Avital A, Springer C, Lavie M, Prais D, Dabbah H, Elias N, Elizur A, Goldberg S, Hevroni A, Kerem E, Luder A, Roth Y, Cohen-Cymberknoh M, Ben Ami M, Mandelberg A, Livnat G, Picard E, Rivlin J, Rotschild M, Soferman R, Loges NT, Olbrich H, Werner C, Wolter A, Herting M, Wallmeier J, Raidt J, **Omran H,** Mussaffi H. Primary ciliary dyskinesia in Israel: Prevalence, clinical features, current diagnosis and management practices. Respir Med. 2016, 119:41-47.
120. Amirav I, Roduta Roberts M, Mussaffi H, Mandelberg A, Roth Y, Abitbul R, Luder A, Blau H, Alkrinawi S, Aviram M, Ben-Ami M, Rotschild M, Bentur L, Shoseyov D, Cohen-Cymberknoh M, Kerem E, Avital

A, Springer C, Hevroni A, Dabbah H, Elizur A, Picard E, Goldberg S, Rivlin J, Livnat G, Lavie M, Alias N, Soferman R, Olbrich H, Raidt J, Wallmeier J, Werner C, Loges NT, **Omran H**. Collecting clinical data in primary ciliary dyskinesia- challenges and opportunities. F1000Res. 2016, 5:2031.

121. Fuchs S, Kaiser-Labusch P, Bank J, Ammann S, Kolb-Kokocinski A, Edelbusch C, **Omran H**, Ehl S. Tyrosine kinase 2 is not limiting human antiviral type III interferon responses. Eur J Immunol. 2016, 46:2639-2649.
122. Lucas JS, Barbato A, Collins SA, Goutaki M, Behan L, Caudri D, Dell S, Eber E, Escudier E, Hirst RA, Hogg C, Jorissen M, Latzin P, Legendre M, Leigh MW, Midulla F, Nielsen KG, **Omran H**, Papon JF, Pohunek P, Redfern B, Rigau D, Rindlisbacher B, Santamaria F, Shoemark A, Snijders D, Tonia T, Titieni A, Walker WT, Werner C, Bush A, Kuehni CE. European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. Eur Respir J. 2017, 4; 49.
123. Paff T, Loges NT, Aprea I, Wu K, Bakey Z, Haarman EG, Daniels JM, Sistermans EA, Bogunovic N, Dougherty GW, Höben IM, Große-Onnebrink J, Matter A, Olbrich H, Werner C, Pals G, Schmidts M, **Omran H**, Micha D. Mutations in PIH1D3 Cause X-Linked Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. Am J Hum Genet. 2017, 100:160-168.
124. Goutaki M, Maurer E, Halbeisen FS, Amirav I, Barbato A, Behan L, Boon M, Casaulta C, Clement A, Crowley S, Haarman E, Hogg C, Karadag B, Koerner-Rettberg C, Leigh MW, Loebinger MR, Mazurek H, Morgan L, Nielsen KG, **Omran H**, Schwerk N, Scigliano S, Werner C, Yiallouros P, Zivkovic Z, Lucas JS, Kuehni CE; PCD Italian Consortium.; Swiss PCD Group.; French Reference Centre for Rare Lung Diseases.; Genetic Disorders of Mucociliary Clearance Consortium. The international primary ciliary dyskinesia cohort (iPCD Cohort): methods and first results. Eur Respir J. 2017, 49
125. Kyriacou K, Yiallouros PK, Kouis P, Papatheodorou SI, **Omran H**. Better experimental screening and treatment for primary ciliary dyskinesia: The FP7 BESTCILIA project. Ultrastruct Pathol. 2017 41:110-112.
126. Emiralioglu N, Wallmeier J, Olbrich H, **Omran H**, Ozcelik U. DYNC2H1 Mutation Causes Jeune Syndrome and Recurrent Lung Infections Associated with Ciliopathy. Clin Respir J. 2017, 12:1017-1020.
127. Edelbusch C, Cindrić S, Dougherty GW, Loges NT, Olbrich H, Rivlin J, Wallmeier J, Pennekamp P, Amirav I, **Omran H.** Mutation of serine/threonine protein kinase 36 (STK36) causes primary ciliary dyskinesia with a central pair defect. Hum Mutat. 2017, 38:964-969.
128. Sigg MA, Menchen T, Lee C, Johnson J, Jungnickel MK, Choksi SP, Garcia G 3rd, Busengdal H, Dougherty GW, Pennekamp P, Werner C, Rentzsch F, Florman HM, Krogan N, Wallingford JB, **Omran H,** Reiter JF. Evolutionary Proteomics Uncovers Ancient Associations of Cilia with Signaling Pathways. Dev Cell. 2017, 43:744-762.
129. König J, Kranz B, König S, Schlingmann KP, Titieni A, Tönshoff B, Habbig S, Pape L, Häffner K, Hansen M, Büscher A, Bald M, Billing H, Schild R, Walden U, Hampel T, Staude H, Riedl M, Gretz N, Lablans M, Bergmann C, Hildebrandt F, **Omran H**, Konrad M; Gesellschaft für Pädiatrische Nephrologie (GPN). Phenotypic Spectrum of Children with Nephronophthisis and Related Ciliopathies. Clin J Am Soc Nephrol. 2017, 12:1974-1983.
130. Lucas JS, Evans HJ, Haarman EG, Hirst RA, Hogg C, Jackson CL, Nielsen KG, **Omran H,** Papon JF, Robinson P, Shoemark A, Walker WT. Exploring the Art of Ciliary Beating: The Benefits of High-Speed Video Analysis. Chest. 2017, 152:1348-1349.
131. Pol A, Renkema GH, Tangerman A, Winkel EG, Engelke UF, de Brouwer APM, Lloyd KC, Araiza RS, van den Heuvel L, **Omran H**, Olbrich H, Oude Elberink M, Gilissen C, Rodenburg RJ, Sass JO, Schwab KO, Schäfer H, Venselaar H, Sequeira JS, Op den Camp HJM, Wevers RA. Mutations in SELENBP1, encoding a novel human methanethiol oxidase, cause extraoral halitosis. Nat Genet. 2018, 50:120-129.
132. Potratz J, Ahlmann M, Rössig C, **Omran H**, Masjosthusmann K. Successful Extracorporeal Life Support in a Pediatric Hematopoietic Stem Cell Transplant Recipient With Periengraftment Respiratory Failure. J Pediatr Hematol Oncol. 2018, 40:e256-e259.
133. Schaefer C, Mallela N, Seggewiß J, Lechtape B, **Omran H**, Dirksen U, Korsching E, Potratz J. Target discovery screens using pooled shRNA libraries and next-generation sequencing: A model workflow and analytical algorithm. PLoS One. 2018, 13(1):e0191570. doi: 10.1371/journal.pone.0191570. eCollection 2018.
134. Höben IM, Hjeij R, Olbrich H, Dougherty GW, Nöthe-Menchen T, Aprea I, Frank D, Pennekamp P, Dworniczak B, Wallmeier J, Raidt J, Nielsen KG, Philipsen MC, Santamaria F, Venditto L, Amirav I,

Mussaffi H, Prenzel F, Wu K, Bakey Z, Schmidts M, Loges NT**, Omran H.** Mutations in C11orf70 Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry Due to Defects of Outer and Inner Dynein Arms. Am J Hum Genet. 2018, 102:973-984.

135. Halbeisen FS, Goutaki M, Spycher BD, Amirav I, Behan L, Boon M, Hogg C, Casaulta C, Crowley S, Haarman EG, Karadag B, Koerner-Rettberg C, Loebinger MR, Mazurek H, Morgan L, Nielsen KG, **Omran H**, Santamaria F, Schwerk N, Thouvenin G, Yiallouros P, Lucas JS, Latzin P, Kuehni CE. Lung function in patients with primary ciliary dyskinesia: an iPCD Cohort study. Eur Respir J. 2018 Aug 23;52(2). pii: 1801040. doi: 10.1183/13993003.01040-2018. Print 2018 Aug.
136. Ta-Shma A, Hjeij R, Perles Z, Dougherty GW, Abu Zahira I, Letteboer SJF, Antony D, Darwish A, Mans DA, Spittler S, Edelbusch C, Cindrić S, Nöthe-Menchen T, Olbrich H, Stuhlmann F, Aprea I, Pennekamp P, Loges NT, Breuer O, Shaag A, Rein AJJT, Gulec EY, Gezdirici A, Abitbul R, Elias N, Amirav I, Schmidts M, Roepman R, Elpeleg O, **Omran H.** Homozygous loss-of-function mutations in MNS1 cause laterality defects and likely male infertility. PLoS Genet. 2018, 14(8):e1007602.
137. Radine A, Werner C, Raidt J, Dougherty GW, Kerschke L, **Omran H,** Grosse-Onnebrink J. Comparison of Nocturnal Cough Analysis in Healthy Subjects and in Patients with Cystic Fibrosis and Primary Ciliary Dyskinesia: A Prospective Observational Study. Respiration. 2018 Nov 8:1-10. doi: 10.1159/000493323. [Epub ahead of print]
138. Loges NT, Antony D, Maver A, Deardorff MA, Güleç EY, Gezdirici A, Nöthe-Menchen T, Höben IM, Jelten L, Frank D, Werner C, Tebbe J, Wu K, Goldmuntz E, Čuturilo G, Krock B, Ritter A, Hjeij R, Bakey Z, Pennekamp P, Dworniczak B, Brunner H, Peterlin B, Tanidir C, Olbrich H, **Omran H,** Schmidts M. Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. Am J Hum Genet. 2018 pii: S0002-9297(18)30372-0. doi: 10.1016/j.ajhg.2018.10.020. [Epub ahead of print

Übersichtsarbeiten

1. Hildebrandt F., Otto E., **Omran H.** Nephronophthise und verwandte Erkrankungen. *Med Gen*1 2:225-231, 2000
2. Hildebrandt F. und **Omran H.** New insights: Nephronophthisis / medullary cystic kidney disease. *Ped Nephrol* 16:168-176, 2001 (IF 2,0)
3. **Omran H.** und Hildebrandt F. Nephronophthise und „medullary cystic kidney disease". *Nieren u Hochdruckk* 30:304-309, 2001
4. Ibañez-Tallon I., Heintz N., **Omran H**. To beat or not to beat, roles of cilia in development and disease. *Hum Mol Genet*, 12:R27-35, 2003 (IF 7,7)
5. El Zein L., **Omran H.**, Bouvagnet P. Lateralization defects and ciliary dyskinesia: lessons from algae. *Trends Genet.* 19:162-7, 2003 (IF 9,3)
6. **Omran H**. Genetische Defekte bei Primärer Ziliärer Dyskinesie. *Monatsschr Kinderheilkd* 153:246-254, 2005
7. **Omran H**. Genetische Grundlagen und Diagnostik hereditärer Erkrankungen. *Neuropädiatrie in Klinik und Praxis*, 2:1-9, 2005
8. Storm van's Gravesande K. und **Omran H**. Primary Ciliary Dyskinesia: clinical presentation, diagnosis and genetics. *Ann Med* 37:439-449, 2005 (IF 5,4)
9. Fliegauf M. und **Omran H**. Novel tools to unravel molecular mechanisms in cilia-related disorders. *Trends Genet*. 22:241-5, 2006 (IF 9,3)
10. **H. Omran** und N. Hornef. Genetische Defekte bei primärer ziliärer Dyskinesie. *Atemw.-Lungenkrkh*. 32:194–200, 2006
11. Zariwala MA, Knowles MR, **Omran H.** Genetic Defects in Ciliary Structure and Function. *Annu Rev Physiol.* 69:423-450, 2006 (IF 20,8)
12. von Schnakenburg C, Fliegauf M, **Omran H**. Nephrocystin and ciliary defects not only in the kidney? Pediatr Nephrol. 22:765-9, 2007 (IF 2,9)

13. Fliegauf M, Benzing T, **Omran H**. When cilia go bad: cilia defects and ciliopathies. *Nat Rev Mol Cell Biol*. 8:880-93, 2007. Erratum in: *Nat Rev Mol Cell Biol*. 2008 Jan;9(1):88. (IF 37,1)
14. **H. Omran**. Diagnostik der primären ziliären Dyskinesie. *Der Pneumologe*. 4:267-275, 2007
15. Barbato A, Frischer T, Kuehni CE, Snijders D, Azevedo I, Baktai G, Bartoloni L, Eber E,Escribano A, Haarman E, Hesselmar B, Hogg C, Jorissen M, Lucas J, Nielsen KG, O'CallaghanC, **Omran H**, Pohunek P, Strippoli MP, Bush A. Primary ciliary dyskinesia: a consensus statement on diagnostic and treatment approaches in children. *Eur Respir J.* 34:1264-76, 2009 (IF 6,3)
16. **Omran H**. NPHP proteins: gatekeepers of the ciliary compartment. *J Cell Biol.* 190:715-7, 2010 (IF 10,8)

Buchkapitel

1. **Omran, H**., Ketelsen U.P., Rudnik-Schöneborn S., Wirth B., Zerres K., und Korithenberg R.Axonale Neuropathie: Keine Seltenheit bei infantiler spinaler Muskelatriophie (SMA I) mit homozygoten SMN-Deletionen. In: Bentele K.H.P. und Kohlschütter A. Aktuelle Neuropädiatrie1998, Novartis Pharma Verlag, Nürnberg, p 317-318, 1998
2. Hildebrandt F. und **Omran H**. Positional cloning and linkage analysis. In: Hildebrandt F. und Igarashi, P. (eds) Techniques in Molecular Medicine. Springer, Berlin – Heidelberg – New York, p352-363, 1999
3. **Omran H.**, Häffner K., Sasmaz G., Völz A., Wienker T.F., Korinthenberg R., Brandis M., Hildebrandt F. Homozygotiekartierung des ersten Genortes für das Senior-Løken Syndrom (SLS1) auf Chromosom 3. Aktuelle Neuropädiatrie 2001, Novartis Pharma Verlag, Nürnberg, p161-164, 2002
4. **Omran H**. Basic knowledge on genetics and inheritance. In: Panteliadis C.P. und Korinthenberg R. (eds) Paediatric Neurology. Thieme Verlag, Heidelberg, 184-187, 2005 (IF 8,2)
5. **Omran H**. & Panteliadis C.P. Neural tube defects. In: Panteliadis C.P. und Korinthenberg R.(eds) Paediatric Neurology. Thieme Verlag, Heidelberg, 252- 266, 2005 (IF 8,2)
6. **Omran H**. Genetics of Neurocutaneous syndromes. In Panteliadis C.P., Benjamin R., Cremer H.-J., Hagel C., Omran H. (eds) Neurocutaneous Disorders Hemangiomas a clinical and diagnostic approach. Pantel Publikation, Thessaloniki 15-40, 2007
7. **Omran H**. & Ermisch-Omran B. Nephronophthisis and Medullary cystic kidney disease. In Geary DF and Schaefer F (eds) Comprehensive Pediatric Nephrology. Mosby Elsevier, Philadelphia143-154, 2008
8. **Omran H**. Genetische Grundlagen und Diagnostik hereditärer Erkrankungen. In Aksu F (ed) Neuropädiatrie. Uni-Med Bremen, London 117-126, 2008
9. Becker-Heck A., Loges, N.T., **Omran, H**. Dynein Dysfunction as a Cause of Primary Ciliary Dyskinesia and Other Ciliopathies, in: S.M. King (Ed.), Dyneins. Academic Press is an imprint of Elsevier., London, Waltham, San Diego, pp. 602-628.1
10. **Heymut Omran** and Niki T. Loges. Immunofluorescence Staining of Ciliated RespiratoryEpithelial Cells. In: Stephen M. King and Gregory J. Pazour, editors, Methods in Cell Biology(Volume 91). Academic Press, 2009, p. 123-134.
11. König J., Ermisch-Omran, B. and **Omran, H**. Nephronophthisis and Autosomal Dominant Interstitial Kidney Disease (ADIKD). Pediatric Kidney Disease. 2017. p 369-388