Curriculum Vitae

Personal Data

Title	Dr. med.
First name	Miriam
Name	Schmidts
Current position	Clinical Fellow and Group Leader
Current institution(s)/site(s), country	Center for Pediatrics and Adolescent Medicine, Medical Center - University of Freiburg, Germany
Identifiers/ORCID	0000-0002-1714-6749

Qualifications and Career

Stages	**Periods and Details**
Degree programme	Medical studies, 1998-2005, University of Freiburg, Germany
Doctorate	2003-2005, Doctoral thesis (Dr. med.) on Hereditary Nephrotic Syndromes, Nephrology Division, University Medical Hospital Freiburg (*Magna cum Laude*)
Stages of academic/ professional career	2018-present: Clinical Fellow, Center for Pediatrics and Adolescent Medicine, University Hospital Freiburg; ERC starting grant holder 2015-present: Assistant Professor Genome Research Division, Human Genetics department, Radboud University Hospital *Nijmegen*, *The Netherlands* 2011-2014: Clinical Research Fellow; Fellowship Action Medical Research (AMR), ICH (UCL), *London, UK*; 2010: MRCP (UK) equivalence 2009-2011: Academic Research Fellow (DFG (German Research Foundation) Research Fellowship), Molecular Medicine Unit, Institute of Child Health (ICH), University College London (UCL), *London, UK* 2005-2008: Clinical Fellow, Center for Pediatrics and Adolescent Medicine, University 2005: Final German Medical Exam “Ärztliche Prüfung”

Supplementary Career Information

1 child (* 2015), maternity leave 2015, career delay due to childbirth and subsequent childcare

Activities in the Research System

Since 2023	SFB SmallData PI + IRTG graduate school board member
2021-present	Freiburg Center for Rare Diseases (FZSE) board member
2021-present	CIBSS (Center for Integrated Biological Signalling Studies) Excellence Cluster associated investigator
Since 2020	SFB1453 Nephgen (PI, board member)
2020-2022	Scientific Advisor EMBO Cilia Conference 2022 (Cologne)
2019-present	Scientific advisory board of the Horizon2020 project LYSOCIL
2018-present	Spemann Graduate School of Biology and Medicine (SGBM) member
2016-2018	Organising committee for EMBO Cilia 2017 conference in Copenhagen
2014-2016	Local organising committee for EMBO Cilia 2016 conference in Amsterdam
2013	Co-Founder of Jeune Syndrome Foundation UK (http://www.jeunes.org.uk)
2012-present	Scientific Advisor for the Ciliopathy Alliance UK
2011-present	Member of the UK10K consortium (http://www.uk10k.org)

Invited lectures at international conferences and meetings

2022	*FASEB Cilia* Tucson, AZ, USA
2022	GfE (German developmental Biology society) conference, Stuttgart, Germany
2018	EMBO Cilia conference, Copenhagen, Denmark
2017	*Gordon Research Conference*, Cilia, Mucus and Mucociliary Interactions, TX, USA
2014	*Turkish Genetic Conference* (Istanbul);Turkey
2014	*British Society for Human Genetics* (BSHG) annual conference;UK
2014	*5th Genetic Conference Kuweit*, Kuwait City
2013	Plenary talk at the German Pediatric Nephrology Association (GPN) Munster) Plenary talk; at the UCL Rare Disease Conference Ciliopathies, London, UK; Lecture at the Ciliopathy Alliance Family Conference, Northampton, UK;
2013	German Pediatric Nephrology (GPN), Hamburg, Germany
2012	German Pediatric Nephrology (GPN) Genetics Group Annual meeting

Scientific Results

Category A

1. Riedhammer KM*, Nguyen TT*, Koşukcu C, Calzada-Wack J, Li Y, Saygılı S, Wimmers V, Kim GJ, Chrysanthou M, Bakey Z, Kraiger M, Sanz-Moreno A, Amarie OV, Rathkolb B, Klein-Rodewald T, Garrett L, Hölter SM, Seisenberger C, Haug S, Schlosser P, Marschall S, Wurst W, Fuchs H, Gailus-Durner V, Wuttke M, Hrabe de Angelis M, Ćomić J, Akgün Doğan Ö, Özlük Y, Taşdemir M, Ağbaş A, Canpolat N, Çalışkan S, Weber R, Bergmann C, Jeanpierre C, Saunier S, Lim TY, Hildebrandt F, Alhaddad B, Wu K, Antony D, Matschkal J, Schaaf C, Renders L, Schmaderer C, Meitinger T, Heemann U, Köttgen A, Arnold SJ, Ozaltin F, **Schmidts M*,** Hoefele J*. (2024) Implication of *FOXD2* dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT). ***Kidney International*** 105(4):844-864. [open access]
2. Bakey Z*, Cabrera OA*, Hoefele J, Antony D, Wu K, Stuck MW, Micha D, Eguether T, Smith AO, van der Wel NN, Wagner M, Strittmatter L, Beales PL, Jonassen JA, Thiffault I, Cadieux-Dion M, Boyes L, Sharif S, Tüysüz B, Dunstheimer D, Niessen HWM, Devine W, Lo CW, Mitchison HM, **Schmidts M*,** Pazour GJ*. (2023) IFT74 variants cause skeletal ciliopathy and motile cilia defects in mice and humans. ***PLoS Genet***. 14;19(6):e1010796. [open access]
3. Sanderson LE*, Lanko K*, Alsagob M*, Almass R*, Al-Ahmadi N*, Najafi M, Al-Muhaizea MA, Alzaidan H, AlDhalaan H, Perenthaler E, van der Linde HC, Nikoncuk A, Kühn NA, Antony D, Owaidah TM, Raskin S, Vieira LGDR, Mombach R, Ahangari N, Silveira TRD, Ameziane N, Rolfs A, Alharbi A, Sabbagh RM, AlAhmadi K, Alawam B, Ghebeh H, AlHargan A, Albader AA, Binhumaid FS, Goljan E, Monies D, Mustafa OM, Aldosary M, AlBakheet A, Alyounes B, Almutairi F, Al-Odaib A, Aksoy DB, Basak AN, Palvadeau R, Trabzuni D, Rosenfeld JA, Karimiani EG, Meyer BF, Karakas B, Al-Mohanna F, Arold ST, Colak D, Maroofian R, Houlden H, Bertoli-Avella AM, **Schmidts M***, Barakat TS*, van Ham TJ*, Kaya N*. (2021) Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. ***Brain***. 144(3):769-780. doi: 10.1093/brain/awaa459. [open access]
4. 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***. (2018) Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. ***Am J Hum Genet.*** 103(6):995-1008. doi: 10.1016/j.ajhg.2018.10.020 [open access]
5. Paff T*, Loges NT*, Aprea I, Wu K, Bakey Z, Haarman EG, Daniels JMA, 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. (2017) Mutations in PIH1D3 Cause X-Linked

Primary Ciliary Dyskinesia with Outer and Inner Dynein Arm Defects. ***Am J Hum Genet.*** 100(1):160-168. doi: 10.1016/j.ajhg.2016.11.019. [open access]

6. **Schmidts M***, Hou Y*, Cortés CR, Mans DA, Huber C, Boldt K, Patel M, van Reeuwijk J, Plaza JM, van Beersum SEC, Yap ZM, Letteboer SJF, Taylor SP, Herridge W, Johnson CA, Scambler PJ, Ueffing M, Kayserili H, Krakow D, King SM, UK10K, Beales PL, Al-Gazali L, Wicking C, Cormier-Daire V, Roepman R,, Mitchison HM*, Witman GB*. (2015) TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. ***Nat Commun.*** 6:7074. doi: 10.1038/ncomms8074. [open access]
7. Wheway G*, **Schmidts M***, Mans DA*, Szymanska K*, Nguyen TMT*, 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 SJF, Roosing S, Adams M, Bell SM, Bond J, Higgins J, Morrison EE, Tomlinson DC, Slaats GG, van Dam TJP, 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*. (2015) An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. ***Nat Cell Biol.*** 17(8):1074-1087. doi: 10.1038/ncb3201. [open access]
8. **Schmidts M**, Vodopiutz J, Christou-Savina S, Cortés CR, McInerney-Leo AM, Emes RD, Arts HH, Tüysüz B, D'Silva J, Leo PJ, Giles TC, Oud MM, Harris JA, Koopmans M, Marshall M, Elçioglu N, Kuechler A, Bockenhauer D, Moore AT, Wilson LC, Janecke AR, Hurles ME, Emmet W, Gardiner B, Streubel B, Dopita B, Zankl A, Kayserili H, Scambler PJ, Brown MA, Beales PL, Wicking C, UK10K, Duncan EL, Mitchison HM. (2013) Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. ***Am J Hum Genet.*** 93(5):932-944. doi: 10.1016/j.ajhg.2013.10.003. [open access]
9. **Schmidts M,** Frank V, Eisenberger T, al Turki S, Bizet AA, Antony D, Rix S, Decker C, Bachmann N, Bald M, Vinke T, Toenshoff B, Di Donato N, Neuhann T, Hartley JL, Maher ER, Bogdanović R, Peco-Antić A, Mache C, Hurles ME, Joksić I, Guć-Šćekić M, Dobricic J, Brankovic-Magic M, UK10K, Bolz HJ, Pazour GJ, Beales PL, Scambler PJ, Saunier S, Mitchison HM, Bergmann C. (2013) Combined NGS approaches identify mutations in the intraflagellar transport gene *IFT140* in skeletal ciliopathies with early progressive kidney disease. ***Human Mutation.*** 34(5):714-724. doi: 10.1002/humu.22294. [open access]
10. Mitchison HM, **Schmidts M**, Loges, N T, Freshour, J, Dritsoula, A, Hirst RA, O'Callaghan C, Blau H, Al Dabbagh M, Olbrich H, Beales PL, Yagi T, Mussaffi H, Chung EMK, Omran H*, Mitchell DR*. (2012). Mutations in axonemal dynein assembly factor DNAAF3 cause primary ciliary dyskinesia ***Nat Genet.*** 44(4):381-389. doi: 10.1038/ng.1106.

**contributed equally*

Category B

1. Riedhammer KM, Nguyen TT, Koşukcu C, Calzada-Wack J, Li Y, Saygılı S, Wimmers V, Kim GJ, Chrysanthou M, Bakey Z, Kraiger M, Sanz-Moreno A, Amarie OV, Rathkolb B, Klein-Rodewald T, Garrett L, Hölter SM, Seisenberger C, Haug S, Schlosser P, Marschall S, Wurst W, Fuchs H, Gailus-Durner V, Wuttke M, Hrabe de Angelis M, Ćomić J, Akgün Doğan Ö, Özlük Y, Taşdemir M, Ağbaş A, Canpolat N, Çalışkan S, Weber R, Bergmann C, Jeanpierre C, Saunier S, Lim TY, Hildebrandt F, Alhaddad B, Wu K, Antony D, Matschkal J, Schaaf C, Renders L, Schmaderer C, Meitinger T, Heemann U, Köttgen A, Arnold SJ, Ozaltin F, **Schmidts M,** Hoefele J. (2023) Implication of *FOXD2* dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT). ***medRxiv***, doi: 10.1101/2023.03.21.23287206. [accepted for publication in ***Kidney International***]

2. Antony D, Yýlmaz Güleç E, Bakey Z, Schüle I, Kim GJ, Skatulla I, Brunner HG, Arnold SJ, **Schmidts M**. Base editing derived models of human *WDR34* and *WDR60* disease alleles replicate. ***bioRxiv***. doi.org/10.1101/2022.03.14.483768 retrograde IFT and hedgehog signaling defects and suggest disturbed Golgi protein transport. ***bioRxiv*** 2022. doi.org/10.1101/2022.03.14.483768
3. **Schmidts M***, Beales PL*. Ciliopathies: Their Role in Pediatric Renal Disease. *In*: Pediatric Nephrology 2nd edition. Edited by Denis Geary and Franz Schaefer. Springer (in press, *2023*). *Book chapter*
4. **Schmidts M**, Liebau MC. Genetic Kidney Diseases of Childhood. Front Pediatr. 2018 Dec 19;6:409. *Editorial**.***
5. **Schmidts M, Mitchison H.** Dynein related skeletal abnormalities *In***:** Dyneins: Structure, Biology and Disease. Edited by Steve King. Elsevier, (*2016*). *Book chapter*.
6. **Schmidts M.** Role of cilia in development and disease in the skeleton. *In:* Cilia: development and disease, CRC press, the Taylor and Francis group. (*2016*). *Book chapter*.
7. **Schmidts M***, Beales PL*. Ciliopathies: Their Role in Pediatric Renal Disease. *In*: Pediatric Nephrology 2nd edition. Springer Business and Media Publishing (*2016*). *Book Chapter*.

Academic Distinctions

2018	Friedrich Linneweh Award for Pediatric Research
2016	ERC starting grant (10% funding rate)
2014	Radboudumc Hypatia Tenure Track Fellowship, Nijmegen, Netherlands
2014	Radboud University Excellence Initative Fellowship, Nijmegen, Netherlands
2014	Robin Winter Prize (British Clinical Genetics Society, CGS, UK)
2013	Young Investigator Award of the International Skeletal Dysplasia Society (ISDS)
2013	Junior Investigator award at the Joubert Syndrome Biennial Conference, Boston, USA
2013	BSHG (British Society for Human Genetics) National Travel fellowship for ESHG 2013
2011	Action Medical Research Clinical Training Fellowship, UK (< 10% funding rate)
2011	Else-Kröner-Fresenius Award for Pediatric Nephrology, Germany
2009	Postdoctoral Fellowship (German Research Foundation, DFG, Germany) for 24 months

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I may **revoke** my consent in whole or in part at any time – with effect for the future, freely and without giving reasons – vis-à-vis the DFG (postmaster@dfg.de). The lawfulness of the processing carried out up to that point remains unaffected. Insofar as I transmit "special categories of personal data" relating to third parties, I confirm that the necessary legitimation under data protection law exists (e.g. based on consent).

I have taken note of the DFG's Data Protection Notice relating to research funding, which I can access at www.dfg.de/privacy_policy and I will forward it to such persons whose data the DFG processes as a result of being mentioned in this CV.