Publications 2014-2018 Anita Rauch

In 2014-2018 first or senior author of 14, and co-author of 43 peer-reviewed original publications, senior author of 4 reviews, and contributions to 2 book chapters

The complete list of publications (in total 202 peer-reviewed publications (66 as first/senior author), 15 reviews, 6 book chapters) is available at http://www.researcherid.com/rid/C-5568-2014 (last 20 years H-index: 50)

List of the top 5 cited original publications as first or senior author:

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2. **Rauch A,** Thiel CT, Schindler D, Wick U, Crow YJ, Ekici AB, van Essen AJ, Goecke TO, Al-Gazali L, Chrzanowska KH, Zweier C, Brunner HG, Becker K, Curry CJ, Dallapiccola B, Devriendt K, Dörfler A, Kinning E, Megarbane A, Meinecke P, Semple RK, Spranger S, Toutain A, Trembath RC, Voß E, Wilson L, Hennekam R, de Zegher F, Dörr HG, Reis A: Mutations in the PCNT gene cause primordial dwarfism. *Science* 2008, 319:816-819. (**times cited 227**)
3. **Rauch A**, Hoyer J, Guth S, Zweier C, Kraus C, Becker C, Zenker M, Huffmeier U, Thiel C, Ruschendorf F, Nurnberg P, Reis A, Trautmann U: Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. *Am J Med Genet* 2006 A140:2063-74. (Journal IF 2.3; **times cited 216**)
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C) Contributions to books 2014-2018

6. **Expert adviser** for the chapter “Intellectual disability” in Firth HV & Hurst JA (2017) Oxford Desk Reference Clinical Genetics and Genomics, 2nd edition, Oxford University Press

5. Imthurn B, Berger W, Macas E, Magyar I, Oneda B, **Rauch A**, Xie M (2015): “Chapter 8: Polar body diagnosis (PBD): an alternative and supplement to preimplantation diagnosis for single embryo transfer” in Scott Sills E (Ed.) Screening the Single Euploid Embryo, Molecular Genetics in Reproductive Medicine, Springer

D) Oral contributions to international conferences / workshops (2014-2018)

Date	Meeting/Symposium	Location	Title of presentation
10.11.2014	Manchester bianual Birth defects meeting	Manchester (UK)	2novel entities with borderline IQ caused by 5p deletions
06.-10.05.2014	3rd COURSE IN NEXT GENERATION SEQUENCING	Bertinoro di Romagna (Italy)	Exome diagnostics in intellectual disability
23.-25.01.2014	39th Annual Conference of Indian Society of Human Genetics and International conference on Human Genetics	Ahmedabad (India)	Non-syndromic sporadic intellectual disability: an exome sequencing study
25.-	35th Annual David W.	Wisconsin	The significance of small copy

30.07.2014	Smith Workshop on Malformations and Morphogenesis	(USA)	number variants in neuro-developmental disorders
14.-19.08.2015	36th Annual David W. Smith Workshop on Malformations and Morphogenesis	St. Michaels, Maryland (USA)	Clinical and experimental evidence establish a link between KIF7 and C5orf42-related ciliopathies
13.-16.05.2015	4th COURSE IN NEXT GENERATION SEQUENCING	Bertinoro (Italy)	Exome diagnostics in intellectual disability
19.09.2015	Opening Symposium of the Center of Human Safety & Health and Genome Diagnostic Analysis	Dubai	Genetic Testing in Personalized Medicine
15.-17.04.2015	Annual meeting oft he German Society of Human Genetics	Graz (Austria)	High-Resolution microarray testing in prenatal diagnosis significantly increases diagnostic yield
02.10.2015	Symposium Human Genetics in the Era of Translational Medicine	Erlangen (Germany)	Individual Genomes and Personalized Medicine
9.-11.09.2015	European Dysmorphology Meeting	Strasbourg (France)	KDM1A mutations in intellectual disability
06.-09.06.2015	Annual meeting of the European Society of Human Genetics	Glasgow (Scottland)	NGS in diagnostics: Challenging example cases
03.12.2016	Symposium in Honor of Prof. Dr. John M. Opitz	Salt Lake City (USA)	A Praise to Clinical Genetics
29.05.2017	Annual meeting of the European Society of Human Genetics	Copenhagen (Denmark)	Clinical Exomes
11-13.04.2018	The Translational Science of Rare Disease	Tutzing (Germany)	New insights into the etiology of epileptic encephalopathy
05.-07.09.2018	European Dysmorphology Meeting	Strasbourg (France)	SURPRISING TWIST IN A LONG-LASTING UNKNOWN CASE WITH SYNDROMIC PRIMARY MICROCEPHALY
15.11.2018	Annual meeting of the Portugues Society of Human Genetics	Porto (Portugal)	Keynote Lecture: The Genetic Landscape of ID