

## Scientific Output 2016-2021 Anita Rauch

The complete list of publications in peer-reviewed Journals is available at <http://www.researcherid.com/rid/C-5568-2014> (H-index 56 in 01/2022);

### **1) Peer-reviewed publications in international scientific journals 2016-2021**

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**2021**

240. Laugwitz L, Seibt A, Herebian D, Peralta S, Kienzle I, Buchert R, Falb R, Gauck D, Müller A, Grimmel M, Beck-Woedel S, Kern J, Daliri K, Katibeh P, Danhauser K, Leiz S, Alesi V, Baertling F, Vasco G, Steinfeld R, Wagner M, Caglayan AO, Gumus H, Burmeister M, Mayatepek E, Martinelli D, Tamhankar PM, Tamhankar V, Joset P, Steindl K, **Rauch A**, Bonnen PE, Froukh T, Groeschel S, Krägeloh-Mann I, Haack TB, Distelmaier F. Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. *J Med Genet.* 2021 Oct 16:jmedgenet-2021-107729. doi: 10.1136/jmedgenet-2021-107729. Online ahead of print. PMID: 34656997
239. Krumm L, Pozner T, Kaindl J, Regensburger M, Günther C, Turan S, Asadollahi R, **Rauch A**, Winner B (2021) Generation and characterization of an endogenously tagged SPG11-human iPSC line by CRISPR/Cas9 mediated knock-in. *Stem Cell Res* 56:102520, DOI: 10.1016/j.scr.2021.102520, PMID: 34479069
238. VanSickle EA, Michael J, Bachmann AS, Rajasekaran S, Prokop JW, Kuzniecky R, Hofstede FC, Steindl K, **Rauch A**, Lipson MH, Bupp CP (2021) Expanding the phenotype: Four new cases and hope for treatment in Bachmann-Bupp syndrome. *Am J Med Genet A* doi: 10.1002/ajmg.a.62473. Online ahead of print. PMID: 34477286
237. Collier JJ, Guissart C, Oláhová M, Sasorith S, Piron-Prunier F, Suomi F, Zhang D, Martinez-Lopez N, Leboucq N, Bahr A, Azzarello-Burri S, Reich S, Schöls L, Polvikoski TM, Meyer P, Larrieu L, Schaefer AM, Alsaif HS, Alyamani S, Zuchner S, Barbosa IA, Deshpande C, Pyle A, **Rauch A**, Synofzik M, Alkuraya FS, Rivier F, Ryten M, McFarland R, Delahodde A, McWilliams TG, Koenig M, Taylor RW (2021) Developmental Consequences of Defective ATG7-Mediated Autophagy in Humans. *N Engl J Med* 2021 Jun 24;384(25):2406-2417. doi: 10.1056/NEJMoa1915722. PMID: 34161705.
236. Škorić-Milosavljević D, Lahrouchi N, Bosada FM, Dombrowsky G, Williams SG, Lesurf R, Tjong FVY, Walsh R, El Bouchikhi I, Breckpot J, Audain E, Ilgun A, Beekman L, Ratbi I, Strong A, Muenke M, Heide S, Muir AM, Hababa M, Cross L, Zhou D, Pastinen T; German Competence Network for Congenital Heart Defects, Zackai E, Atmani S, Ouldil K, Adadi N, Steindl K, **Rauch A**, Brook D, Wilsdon A, Kuipers I, Blom NA, Mulder BJ, Mefford HC, Keren B, Joset P, Kruszka P, Thiffault I, Sheppard SE, Roberts A, Lodder EM, Keavney BD, Clur SB, Mital S, Hitz MP, Christoffels VM, Postma AV, Bezzina CR (2021) Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. *Genet Med* 2021 Jun 10. doi: 10.1038/s41436-021-01212-y. Online ahead of print. PMID: 34113005
235. Gogoll L, Steindl K, Joset P, Zweier M, Baumer A, Gerth-Kahlert C, Tutschek B, **Rauch A (2021)** Confirmation of Ogden syndrome as an X-linked recessive fatal disorder due to a recurrent NAA10 variant and review of the literature. *Am J Med Genet A* 2021 Jun 1. doi: 10.1002/ajmg.a.62351. Online ahead of print. PMID: 34075687
234. Ernst ME, Baugh EH, Thomas A, Bier L, Lippa N, Stong N, Mulhern MS, Kushary S, Akman CI, Heinzen EL, Yeh R, Bi W, Hanchard NA, Burrage LC, Leduc MS, Chong JSC, Bend R, Lyons MJ, Lee JA, Suwannarat P, Brilstra E, Simon M, Koopmans M, van Binsbergen E, Groeppe D, Fleischer J, Nava C, Keren B, Mignot C, Mathieu S, Mancini GMS, Madan-Khetarpal S, Infante EM, Bluvstein J, Seeley A, Bachman K, Klee EW, Schultz-Rogers LE, Hasadsri L, Barnett S, Ellingson MS, Ferber MJ, Narayanan V, Ramsey K, **Rauch A**, Joset P, Steindl K, Sheehan T, Poduri A, Vasquez A, Ruivenkamp C, White SM, Pais L, Monaghan KG, Goldstein DB, Sands TT, Aggarwal V (2021)

- CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia 2021 May 26. doi: 10.1111/epi.16931. Online ahead of print. PMID: 34041744
233. Parenti I, Lehalle D, Nava C, Torti E, Leitão E, Person R, Mizuguchi T, Matsumoto N, Kato M, Nakamura K, de Man SA, Cope H, Shashi V; Undiagnosed Diseases Network, Friedman J, Joset P, Steindl K, **Rauch A**, Muffels I, van Hasselt PM, Petit F, Smol T, Le Guyader G, Bilan F, Sorlin A, Vitobello A, Philippe C, van de Laar IMBH, van Slegtenhorst MA, Campeau PM, Au PYB, Nakashima M, Saitsu H, Yamamoto T, Nomura Y, Louie RJ, Lyons MJ, Dobson A, Plomp AS, Motazacker MM, Kaiser FJ, Timberlake AT, Fuchs SA, Depienne C, Mignot C (2021) Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Hum Genet 2021 Jul;140(7):1109-1120. doi: 10.1007/s00439-021-02283-2. Epub 2021 May 4. PMID: 33944996
232. Zanoni P, Steindl K, Sengupta D, Joset P, Bahr A, Sticht H, Lang-Muritano M, van Ravenswaaij-Arts CMA, Shinawi M, Andrews M, Attie-Bitach T, Maystadt I, Belnap N, Benoit V, Delplancq G, de Vries BBA, Grotto S, Lacombe D, Larson A, Mourmans J, Öunap K, Petrilli G, Pfundt R, Ramsey K, Snijders Blok L, Tsatsaris V, Vitobello A, Faivre L, Wheeler PG, Wevers MR, Wojcik M, Zweier M, Gozani O, **Rauch A (2021)** Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genet Med 2021 Aug;23(8):1474-1483. doi: 10.1038/s41436-021-01158-1. Epub 2021 May 3.
231. Harris HK, Nakayama T, Lai J, Zhao B, Argyrou N, Gubbels CS, Soucy A, Genetti CA, Suslovitch V, Rodan LH, Tiller GE, Lesca G, Gripp KW, Asadollahi R, Hamosh A, Applegate CD, Turnpenny PD, Simon MEH, Volker-Touw CML, Gassen KLIV, Binsbergen EV, Pfundt R, Gardeitchik T, Vries BBA, Immken LL, Buchanan C, Willing M, Toler TL, Fassi E, Baker L, Vansenne F, Wang X, Ambrus JL Jr, Fannemel M, Posey JE, Agolini E, Novelli A, **Rauch A**, Boonsawat P, Fagerberg CR, Larsen MJ, Kibaek M, Labalme A, Poisson A, Payne KK, Walsh LE, Aldinger KA, Balciuniene J, Skraban C, Gray C, Murrell J, Bupp CP, Pascolini G, Grammatico P, Broly M, Küry S, Nizon M, Rasool IG, Zahoor MY, Kraus C, Reis A, Iqbal M, Uguen K, Audebert-Bellanger S, Ferec C, Redon S, Baker J, Wu Y, Zampino G, Syrbe S, Brosse I, Jamra RA, Dobyns WB, Cohen LL, Blomhoff A, Mignot C, Keren B, Courtin T, Agrawal PB, Beggs AH, Yu TW (2021) Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genet Med. 2021 Mar 3. doi: 10.1038/s41436-021-01114-z. Online ahead of print. PMID: 33658631
230. Rodríguez-Palmero A, Boerrigter MM, Gómez-Andrés D, Aldinger KA, Marcos-Alcalde Í, Popp B, Everman DB, Lovgren AK, Arpin S, Bahrambeigi V, Beunders G, Bisgaard AM, Bjerregaard VA, Bruel AL, Challman TD, Cogné B, Coubes C, de Man SA, Denommé-Pichon AS, Dye TJ, Elmslie F, Feuk L, García-Miñaur S, Gertler T, Giorgio E, Gruchy N, Haack TB, Haldeman-Englert CR, Haukanes BI, Hoyer J, Hurst ACE, Isidor B, Soller MJ, Kushary S, Kvarnung M, Landau YE, Leppig KA, Lindstrand A, Kleinendorst L, MacKenzie A, Mandrile G, Mendelsohn BA, Moghadasi S, Morton JE, Moutton S, Müller AJ, O'Leary M, Pacio-Míguez M, Palomares-Bralo M, Parikh S, Pfundt R, Pode-Shakked B, **Rauch A**, Repnikova E, Revah-Politi A, Ross MJ, Ruivenkamp CAL, Sarrazin E, Savatt JM, Schlüter A, Schönewolf-Greulich B, Shad Z, Shaw-Smith C, Shieh JT, Shohat M, Spranger S, Thiese H, Mau-Them FT, van Bon B, van de Burgt I, van de Laar IMBH, van Drie E, van Haelst MM, van Ravenswaaij-Arts CM, Verdura E, Vitobello A, Waldmüller S, Whiting S, Zweier C, Prada CE, de Vries BBA, Dobyns WB, Reiter SF, Gómez-Puertas P, Pujol A, Tümer Z (2021) DLG4-related synaptopathy: a new rare brain disorder. Genet Med. 2021 Feb 17. doi: 10.1038/s41436-020-01075-9. Online ahead of print. PMID: 33597769
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228. Meng L, Isohanni P, Shao Y, Graham BH, Hickey SE, Brooks S, Suomalainen A, Joset P, Steindl K, **Rauch A**, Hackenberg A, High FA, Armstrong-Javors A, Mencacci NE, González-Latapi P, Kamel WA, Al-Hashel JY, Bustos BI, Hernandez AV, Krainc D, Lubbe SJ, Esch HV, De Luca C, Ballon K, Ravelli C, Burglen L, Qebibo L, Calame DG, Mitani T, Marafi D, Pehlivan D, Saadi NW, Sahin Y, Maroofian R, Efthymiou S, Houlden H, Maqbool S, Rahman F, Gu S, Posey JE, Lupski JR, Hunter JV, Wangler MF, Carroll CJ, Yang Y (2021) MED27 Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Ann Neurol. 2021 Jan 14. doi: 10.1002/ana.26019. Epub ahead of print. PMID: 33443317.
227. Lenaerts L, Reynhout S, Verbinnen I, Laumonnier F, Toutain A, Bonnet-Brilhault F, Hoorne Y, Joss S, Chassevent AK, Smith-Hicks C, Loeys B, Joset P, Steindl K, **Rauch A**, Mehta SG, Chung WK, Devriendt K, Holder SE, Jewett T, Baldwin LM, Wilson WG, Towner S, Srivastava S, Johnson HF, Daumer-Haas C, Baethmann M, Ruiz A, Gabau E, Jain V, Varghese V, Al-Beshri A, Fulton S, Wechsberg O, Orenstein N, Prescott K, Childs AM, Faivre L, Moutton S, Sullivan JA, Shashi V, Koudijs SM, Heijligers M, Kivuva E, McTague A, Male A, van Ierland Y, Plecko B, Maystadt I, Hamid R, Hannig VL, Houge G, Janssens V (2021) The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genet Med. 2021 Feb;23(2):352-362. doi: 10.1038/s41436-020-00981-2. Epub 2020 Oct 27. PMID: 33106617.
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223. Asadollahi R, Britschgi C, Joset P, Oneda B, Schindler D, Meier UR, **Rauch-A (2020)** Severe reaction to radiotherapy provoked by hypo-morphic germline mutations in ATM

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220. Kummeling J, Stremmelaar DE, Raun N, Reijnders MRF, Willemsen MH, Ruiterkamp-Versteeg M, Schepens M, Man CCO, Gilissen C, Cho MT, McWalter K, Sinnema M, Wheless JW, Simon MEH, Genetti CA, Casey AM, Terhal PA, van der Smagt JJ, van Gassen KLI, Joset P, Bahr A, Steindl K, **Rauch A**, Keller E, Raas-Rothschild A, Koolen DA, Agrawal PB, Hoffman TL, Powell-Hamilton NN, Thiffault I, Engleman K, Zhou D, Bodamer O, Hoefele J, Riedhammer KM, Schwaibold EMC, Tasic V, Schubert D, Top D, Pfundt R, Higgs MR, Kramer JM, Kleefstra T (2020) Characterization of SETD1A haploinsufficiency in humans and *Drosophila* defines a novel neurodevelopmental syndrome. Mol Psychiatry. 2020 Apr 28. doi: 10.1038/s41380-020-0725-5. Online ahead of print. PMID: 32346159
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217. Werling AM, Grünblatt E, Oneda B, Bobrowski E, Gundelfinger R, Taurines R, Romanos M, **Rauch A**, Walitza S (2020) High-resolution chromosomal microarray analysis for copy-number variations in high-functioning autism reveals large aberration typical for intellectual disability. J Neural Transm (Vienna) 127:81-94. doi: 10.1007/s00702-019-02114-9. Epub 2019 Dec 14. PMID: 31838600
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#### **4) Contributions to books 2016-2021**

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

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#### **6) Oral contributions to international conferences 2016-2021**

- 2016 Oral presentation on “Further Delineation of a novel 2q11.1q11.2 Microduplication syndrome” at the 27th European Meeting on Dysmorphology, Strasbourg, France
- 2016 Invited oral presentation on NIPT at the Cartagenia inc. workshop at the Annual meeting of the European Society of Human Genetics in Barcelona, Spain
- 2017 Oral presentation on “Two novel European cases of SPATA5 mutations causing epileptic encephalopathy” at the 28th European Meeting on Dysmorphology, Strasbourg, France
- 2017 Invited lecture “Clinical exomes” at the Annual meeting of the European Society of Human Genetics in Copenhagen, Denmark
- 2017 Invited lecture on “New Molecular Tools in Genetics – Rise of the Next Generation” at the Joint Meeting of Swiss, Austrian and German Pediatric Pathologists in Zurich, Switzerland
- 2018 Oral presentation on “Surprising twist in a long-lasting unknown case with syndromic primary microcephaly” at the 29th European Meeting on Dysmorphology, Strasbourg, France
- 2018 Invited lecture “New insights into the etiology of epileptic encephalopathy” at the Translational Science of Rare Disease Meeting in Tutzing, Germany
- 2018 Invited key-note lecture “The Genetic Landscape of ID” at the 22nd Anual Meeting of the Portuguese Society of Human Genetics, Porto, Portugal
- 2019 Oral presentation on “Phenotypic surprises from prenatal rasopathy testing” at the 30th European Meeting on Dysmorphology, Strasbourg, France
- 2019 Invited lecture “Genetic basis of personalized medicine” at the 7<sup>th</sup> International IZKF-Symposium, Bad Staffelstein, Germany
- 2020 Invited lecture on “Genomics of Dysmorphology” at the Pan Arab Human Genetics Conference in Dubai, UAE

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#### **7. Outreach activities 2016-2021**

- 2016 Invited lecture on “*Neue diagnostische Wege*” at the 6<sup>th</sup> rare disease day Switzerland in Zurich, Switzerland
- 2016 Invited lecture at the Swiss MedLab conference on “Hochdurchsatzsequenzierung in der Diagnostik monogener Erkrankungen”, Bern, Switzerland
- 2016 Invited lecture at the annual conference of the Swiss Society of Pediatrics “Exomsequencing und Paneldiagnostik – Angebote in der Schweiz”, St. Gallen,

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Switzerland

- 2016 Lecture on “*Metabolische und genetische Erforschung frühkindlicher epileptischer Enzephalopathien*” at the *radiz-rare disease initiative Zurich Symposium*
- 2016 Lecture “Indikation und Beratung zu genetischen Untersuchungen und Interpretation erhaltenener Resultate” at the *Pädiatrie update refresher course*, Zürich, Switzerland
- 2016 Invited lecture at the special scientific symposium honoring the legay of Professor John M. Opitz, *Advances in the knowledge of Human Genetics*, Salt Lake City, Utah, USA
- 2016 Invited lecture on *New diagnostic approaches* and panel discussion on “Pränatale Diagnostik- schaffe ich das oder nicht?” at the 46<sup>th</sup> conference of the *Schweizerischer Verband Medizinischer PraxisAssistentinnen*, Davos, Switzerland
- 2017 Invited lecture “Lesen im Genom- Was wird aus mir?” Akademie Berlingen, Switzerland
- 2018 Invited lecture “Ebenen der genetischen Information und Zusatzbefunde» at the ethical board of the canton of Zurich
- 2018 Invited lecture “Vorgeburtliche Beratung am Beispiel Trisomie 21» at the *Kompetenzzentrum Medizin-Ethik-Recht Helvetiae*, Zürich, Switzerland
- 2019 Invited lecture “Genanalyse – Risikominimierung” and panel discussion “Seltene Krankheit – Genetik, Diagnostik und der Wunsch nach weiteren Kindern” at the children with rare disease support group meeting in Volketswil, Switzerland
- 2019 Invited lecture “Erbgut editieren – Klinische Anwendung und Dilemmata» Colegium generale HS 2019, University of Bern, Switzerland
- 2019 Invited lecture “Gentechnik – CRISPR Wunderwaffe oder Teufelswerkzeug?” at the Alumni-NDS MiG lecture series, Zurich, Switzerland
- 2019 Invited lecture “Im Genom lesen” at the *Trendtage Gesundheit*, Luzern, Switzerland