Prof. Regina C. Betz, MD

Institute of Human Genetics

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Institute of Human Genetics

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Research Expertise

The aim of our research is the identification and functional characterization of genes for monogenic and genetically complex hair loss disorders with a major focus on the autoimmune disorder alopecia areata (AA). We have the largest sample of AA patients available worldwide, which includes a current total of more than 2.200 individuals of middle European origin. We have been able to demonstrate the contribution of the HLA-complex and the genes PTPN22, TRAF1/C5 , CTLA4, IL13 and KIAA0350 to the disease risk using candidate gene studies. By the use of genome-wide association studies, meta-analyses, immunochips and functional studies, we just recently identified HLA-DR as a key etiologic driver for AA as well as two loci outside the HLA-region: ACOXL/BCL2L11 and GARP. Future analyses and functional studies will contribute to a comprehensive understanding of AA.

Education / Training

University of Bonn, Germany, Human Genetics, Habilitation, 2009
University of Bonn, Germany, Human Genetics, Medical Specialist, 2007
Karolinska Institute, Stockholm, Sweden University of Saarland Clinical Medicine, Medical license, 1999
University of Saarland, Germany, Medicine, MD thesis, 1998

Appointments / Positions Held

2015 - present W2 Professorship, Institute of Human Genetics, University of Bonn, Germany
2010 - 2015 Heisenberg Professorship, Institute of Human Genetics, University of Bonn, Germany
2009 - 2010 Research Scientist, Institute of Human Genetics, University of Bonn, Germany
2004 - 2009 Independent Head of a Junior Research Group, Institute of Human Genetics, University of Bonn, Germany
2002 - 2004 Postdoctoral fellow, Department of Medical Genetics, University of Antwerp, Belgium
2000 - 2002 Postdoctoral fellow, Institute of Human Genetics, University of Bonn, Germany
1999 - 2000 Research Scientist, Institute of Human Genetics, University of Bonn, Germany

Honors / Awards

2010 - 2015 Heisenberg-Professorship from the DFG
2004 - 2009 Emmy Noether Independent Junior Research Group (DFG)
2008 PRO-SCIENTIA-Sponsorship Award of the Eckhart-Buddecke-Foundation for the advancement of basic medical research
2008 EP-Patent application 07 01 8871.9: “Maintenance of hair growth and treatment of hair loss.” (together with Prof. Nöthen, S. Pasternack Dipl.-biol., and Dr. Al Aboud)
2008 Lecture Prize at the Annual Meeting of the European Hair Research Society in Genoa, Italy
2006 Gottron-Just-Scientific Prize of the University and City of Ulm, Germany
2002 - 2004 Flemish Research Council Postdoctoral Fellowship
2000 - 2002 DFG Postdoctoral Fellowship

10 Most Relevant Publications for Prof. Regina Betz

1. **Betz RC**, Petukhova L, Ripke S, [..] Clynes D, de Bakker PIW, Nöthen MM, Daly MJ, Christiano AM: Meta-analysis of genome-wide association studies in alopecia areata resolves HLA associations and reveals two new susceptibility loci. Nat Commun, Jan 2015 22, 6:5966
2. Basmanav FB, Oprisoreanu AM, Pasternack SM, Thiele H, Fritz G, Wenzel J, Größer L, Wehner M, Wolf S, Fagerberg C, Bygum A, Altmüller J, Rütten A, Parmentier L, El Shabrawi-Caelen L, Hafner C, Nürnberg P, Kruse R, Schoch S, Hanneken S, **Betz RC**. 2014. Mutations in POGLUT1, encoding protein O-glucosyltransferase 1, cause autosomal dominant Dowling-Degos disease. Am J Hum Genet 94:135-143.
3. Pasternack SM, Refke M, Paknia E, Hennies HC, Franz T, Schäfer N, Fryer A, van Steensel M, Sweeney E, Just M, Grimm C, Kruse R, Ferrándiz C, Nöthen MM, Fischer U, **Betz RC**. 2013. Mutations in SNRPE, encoding a core protein of the spliceosome, cause autosomal-dominant hypotrichosis simplex. Am J Hum Genet 92:81-87.
4. Jagielska D, Redler S, Brockschmidt FF, Herold C, Garcia Bartels N, Hanneken S, Eigelshoven S, Refke M, Barth S, Giehl KA, Kruse R, Lutz G, Wolff H, Blaumeiser B, Böhm M, Blume-Peytavi U, Becker T, Nöthen MM, **Betz RC**. 2012. A follow-up study of a genome-wide association scan in alopecia areata: replication of previously identified loci and identification of IL13 and KIAA0350 as new susceptibility loci supported with genome-wide significance. J Invest Dermatol 132:2192-2197.
5. Wen Y, Liu Y, Xu Y, Zhao Y, Hua R, Wang K, Sun M, Li Y, Yang S, Zhang XJ, Kruse R, Cichon S, **Betz RC**, Nothen MM, van Steensel MA, van Geel M, Steijlen PM, Hohl D, Huber M, Dunnill GS, Kennedy C, Messenger A, Munro CS, Terrinoni A, Hovnanian A, Bodemer C, de Prost Y, Paller AS, Irvine AD, Sinclair R, Green J, Shang D, Liu Q, Luo Y, Jiang L, Chen HD, Lo WH, McLean WH, He CD, Zhang X. 2009. Loss-of-function mutations of an inhibitory upstream ORF in the human hairless transcript cause Marie Unna hereditary hypotrichosis. Nat Genet 41: 228-33.
6. Pasternack SM, von Kugelgen I, Aboud KA, Lee YA, Ruschendorf F, Voss K, Hillmer AM, Molderings GJ, Franz T, Ramirez A, Nurnberg P, Nothen MM, **Betz RC**. 2008. G protein-coupled receptor P2Y5 and its ligand LPA are involved in maintenance of human hair growth. Nat Genet 40: 329-34.
7. Hillmer AM, Brockschmidt FF, Hanneken S, Eigelshoven S, Steffens M, Flaquer A, Herms S, Becker T, Kortum AK, Nyholt DR, Zhao ZZ, Montgomery GW, Martin NG, Muhleisen TW, Alblas MA, Moebus S, Jockel KH, Brocker-Preuss M, Erbel R, Reinartz R, **Betz RC**, Cichon S, Propping P, Baur MP, Wienker TF, Kruse R, Nothen MM. 2008. Susceptibility variants for male-pattern baldness on chromosome 20p11. Nat Genet 40: 1279-81.
8. **Betz RC**, Planko L, Eigelshoven S, Hanneken S, Pasternack SM, Bussow H, Van Den Bogaert K, Wenzel J, Braun-Falco M, Rutten A, Rogers MA, Ruzicka T, Nothen MM, Magin TM, Kruse R. 2006. Loss-of-function mutations in the keratin 5 gene lead to Dowling-Degos disease. Am J Hum Genet 78: 510-9.
9. Levy-Nissenbaum E, **Betz RC**, Frydman M, Simon M, Lahat H, Bakhan T, Goldman B, Bygum A, Pierick M, Hillmer AM, Jonca N, Toribio J, Kruse R, Dewald G, Cichon S, Kubisch C, Guerrin M, Serre G, Nothen MM, Pras E. 2003. Hypotrichosis simplex of the scalp is associated with nonsense mutations in CDSN encoding corneodesmosin. Nat Genet 34: 151-3.
10. **Betz RC**, Schoser BG, Kasper D, Ricker K, Ramirez A, Stein V, Torbergsen T, Lee YA, Nöthen MM, Wienker TF, Malin JP, Propping P, Reis A, Mortier W, Jentsch TJ, Vorgerd M, Kubisch C. 2001. Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. Nat Genet 28: 218-9.