Stand: Juli 2015

RESEARCH FOR RARE
Forschung für seltene Erkrankungen

Papers of the Month
BMBF-geförderte Forschungsverbünde für seltene Erkrankungen

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http://www.research4rare.de/aktuelles/veroeffentlichungen-der-forschungsverbuende/

2015

1. Juli 2015/Epub Juni (Forschungsverbund HOPE)
Mutations in the unfolded protein response regulator ATF6 cause the cone dysfunction disorder achromatopsia.
Kohl S, Zobor D, Chiang WC, Weisschuh N, Staller J, Menendez IG, Chang S, Beck SC, Garcia Garrido M, Sothilingam V, Seeliger MW, Stanzial F, Benedicenti F, Inzana F, Héon E, Vincent A, Beis J, Strom TM, Rudolph G, Roosing S, Hollander AI, Cremers FP, Lopez I, Ren H, Moore AT, Webster AR, Michaelides M, Koenekoop RK, Zrenner E, Kaufman RJ, Tsang SH, Wissinger B, Lin JH.
Nat Genet. 2015 Jul;47(7):757-65. Epub 2015 Jun 1

26. Mai 2015 (Forschungsverbund AID-Net)
Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases.
Holzinger D, Fassl SK, de Jager W, Lohse P, Röhrig UF, Gattorno M, Omenetti A, Chiesa S, Schena F, Austermann J, Vogl T, Kuhns DB, Holland SM, Rodríguez-Gallego C, López-Almaraz R, Arostegui JI, Colino E, Roldan R, Fessatou S, Isidor B, Poignant S, Ito K, Epple HJ, Bernstein JA, Jeng M, Frankovich J, Lionetti G, Church JA, Ong PY, LaPlant M, Abinun M, Skinner R, Bigley V, Sachs UJ, Hinze C, Hoppenreijs E, Ehrchen J, Foell D, Chae JJ, Ombrello A, Aksentijevich I, Sunderkoetter C, Roth J
J Allergy Clin Immunol. 2015 May 26. [Epub ahead of print].

7. Mai 2015/Epub April (Forschungsverbund FACE)
Acrofacial Dysostosis, Cincinnati Type, a Mandibulofacial Dysostosis Syndrome with Limb Anomalies, Is Caused by POLR1A Dysfunction
Weaver KN, Watt KE, Hufnagel RB, Navajas Acedo J, Linscott LL, Sund KL, Bender PL, König R, Lourenco CM, Hehr U, Hopkin RJ, Lohmann DR, Trainor PA, Wieczorek D, Saal HM.
Am J Hum Genet. 2015 May 7;96(5):765-74. Epub 2015 Apr 23.

GEFÖRDERT VOM
Bundesministerium für Bildung und Forschung

24. März 2015 (Forschungsverbund MND-net)

Haploinsufficiency of TBK1 causes familial ALS and fronto-temporal dementia

Freischmidt A, Wieland T, Richter B, Ruf W, Schaeffer V, Müller K, Marroquin N, Nordin F, Hübers A, Weydt P, Pinto S, Press R, Millecamps S, Molko N, Bernard E, Desnuelle C, Soriani MH, Dorst J, Graf E, Nordström U, Feiler MS, Putz S, Boeckers TM, Meyer T, Winkler AS, Winkelman J, de Carvalho M, Thal DR, Otto M, Brännström T, Volk AE, Kursula P, Danzer KM, Lichtner P, Dikic I, Meitinger T, Ludolph AC, Strom TM, Andersen PM, Weishaupt JH.

Nat Neurosci. 2015 May;18(5):631-6. Epub 2015 Mar 24.

9. März 2015 (Forschungsverbund IonNeurONet)

De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy

Syrbe S, Hedrich UB, Riesch E, Djémié T, Müller S, Møller RS, Maher B, Hernandez-Hernandez L, Synofzik M, Caglayan HS, Arslan M, Serratosa JM, Nothnagel M, May P, Krause R, Löffler H, Detert K, Dorn T, Vogt H, Krämer G, Schöls L, Mullis PE, Linnankivi T, Lehesjoki AE, Sterbova K, Craiu DC, Hoffman-Zacharska D, Korff CM, Weber YG, Steinlin M, Gallati S, Bertsche A, Bernhard MK, Merkenschlager A, Kiess W; EuroEPINOMICS RES, Gonzalez M, Züchner S, Palotie A, Suls A, De Jonghe P, Helbig I, Biskup S, Wolff M, Maljevic S, Schüle R, Sisodiya SM, Weckhuysen S, Lerche H, Lemke JR.

Nat Genet. 2015 Apr;47(4):393-9. Epub 2015 Mar 9

5. Januar 2015 (Forschungsverbund FACE)

CRIM1 haploinsufficiency causes defects in eye development in human and mouse

Beleggia F, Li Y, Fan J, Elcioğlu NH, Toker E, Wieland T, Maumenee IH, Akarsu NA, Meitinger T, Strom TM, Lang R, Wollnik B.

Hum Mol Genet. 2015 Jan 5. [Epub ahead of print]

2014

4. Dezember 2014 (Forschungsverbund MitoNet)

Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy

Kopajtich R, Nicholls TJ, Rorbach J, Metodiev MD, Freisinger P, Mandel H, Vanlander A, Ghezzi D, Carrozzo R, Taylor RW, Marquard K, Murayama K, Wieland T, Schwarzmayr T, Mayr JA, Pearce SF, Powell CA, Saada A, Ohtake A, Invernizzi F, Lamantea E, Sommerville EW, Pyle A, Chinnery PF, Crushell E, Okazaki Y, Kohda M, Kishita Y, Tokuzawa Y, Assouline Z, Rio M, Feillet F, Mousson de Camaret B, Chretien D, Munnich A, Menten B, Sante T, Smet J, Régal L, Lorber A, Khoury A, Zeviani M, Strom TM, Meitinger T, Bertini ES, Van Coster R, Klopstock T, Rötig A, Haack TB, Minczuk M, Prokisch H. Am J Hum Genet. 2014 Dec 4;95(6):708-20 Proteinsynthese

2. November 2014 (Forschungsverbund IonNeurONet)

Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes

Schubert J, Siekierska A, Langlois M, May P, Huneau C, Becker F, Muhle H, Suls A, Lemke JR, de Kovel CG, Thiele H, Konrad K, Kawalia A, Toliat MR, Sander T, Rüschendorf F, Caliebe A, Nagel I, Kohl B, Kecskés A, Jacmin M, Hardies K, Weckhuysen S, Riesch E, Dorn T, Brilstra EH, Baulac S, Møller RS, Hjalgrim H, Koeleman BP; EuroEPINOMICS RES Consortium, Jurkat-Rott K, Lehman-Horn F, Roach JC, Glusman G, Hood L, Galas DJ, Martin B, de Witte PA, Biskup S, De Jonghe P, Helbig I, Balling R, Nürnberg P, Crawford AD, Esguerra CV, Weber YG, Lerche H.

Nat Genet. 2014 Dec;46(12):1327-32. doi: 10.1038/ng.3130. Epub 2014 Nov 2.

21. Oktober 2014 (Forschungsverbund Imprinting)
Epigenetic germline mosaicism in infertile men
Laurentino S, Beygo J, Nordhoff V, Kliesch S, Wistuba J, Borgmann J, Buiting K, Horsthemke B, Gromoll J.
Hum Mol Genet. 2014 Oct 21. pii: ddu540. [Epub ahead of print]

5. September 2014 (Forschungsverbund MND-net)
Serum microRNAs in patients with genetic amyotrophic lateral sclerosis and pre-manifest mutation carriers
Freischmidt A, Müller K, Zondler L, Weydt P, Volk AE, Božič AL, Walter M, Bonin M, Mayer B, von Arnim CA, Otto M, Dieterich C, Holzmann K, Andersen PM, Ludolph AC, Danzer KM, Weishaupt JH.
Brain. 2014 Nov;137(Pt 11):2938-50. Epub 2014 Sep 5.

12. August 2014 (Forschungsverbund Geramy)
A staging system for renal outcome and early markers of renal response to chemotherapy in AL amyloidosis
Palladini G, Hegenbart U, Milani P, Kimmich C, Foli A, Ho AD, Rosin MV, Albertini R, Moratti R, Merlini G, Schönland S.
Blood. 2014 Oct 9;124(15):2325-32. Epub 2014 Aug 12.

17. Juli 2014 (Forschungsverbund HOPE)
Mutation of POC1B in a Severe Syndromic Retinal Ciliopathy.
Beck BB, Phillips JB, Bartram MP, Wegner J, Thoenes M, Pannes A, Sampson J, Heller R, Göbel H, Koerber F, Neugebauer A, Hedergott A, Nürnberg G, Nürnberg P, Thiele H, Altmüller J, Toliat MR, Staubach S, Boycott KM, Valente EM, Janecke AR, Eisenberger T, Bergmann C, Tebbe L, Wang Y, Wu Y, Fry AM, Westerfield M, Wolfrum U, Bolz HJ.
Hum Mutat. 2014 Jul 17. doi: 10.1002/humu.22618. [Epub ahead of print]

30. Juni 2014 (Forschungsverbund TranSaRNet)
Cyclophosphamide Compared With Ifosfamide in Consolidation Treatment of Standard-Risk Ewing Sarcoma: Results of the Randomized Noninferiority Euro-EWING99-R1 Trial
Le Deley MC, Paulussen M, Lewis I, Brennan B, Ranft A, Whelan J, Le Teuff G, Michon J, Ladenstein R, Marec-Bérard P, van den Berg H, Hjorth L, Wheatley K, Judson I, Juergens H, Craft A, Oberlin O, Dirksen U.
J Clin Oncol. 2014 Aug 10;32(23):2440-8. Epub 2014 Jun 30

1. Mai 2014 (Forschungsverbund PID-net)
Human procaspase-1 variants with decreased enzymatic activity are associated with febrile episodes and may contribute to inflammation via RIP2 and NF-κB signalling
Heymann MC, Winkler S, Luksch H, Flecks S, Franke M, Ruß S, Ozen S, Yilmaz E, Klein C, Kallinich T, Lindemann D, Brenner S, Ganser G, Roesler J, Rösen-Wolff A, Hofmann SR.
J Immunol. 2014 May 1;192(9):4379-85

1. April 2014 (Forschungsverbund MitoNet)
MutationTaster2: mutation prediction for the deep-sequencing age
Nat Methods. 2014;11(4):361-2.
Schwarz JM, Cooper DN, Schuelke M, Seelow D.

7. März 2014 (Forschungsverbund EB-net)
Targeting epidermal lipids for treatment of Mendelian disorders of cornification
Kiritsi D, Schauer F, Wölfle U, Valari M, Bruckner-Tuderman L, Has C, Happle R.
Orphanet J Rare Dis. 2014 Mar 7;9:33

28. Februar 2014 (Forschungsverbund BMFS)
Cooperativity of RUNX1 and CSF3R mutations in severe congenital neutropenia: a unique pathway in myeloid leukemogenesis
Blood. 2014 Apr 3;123(14):2229-37
Skokowa J, Steinemann D, Katsman-Kuipers JE, Zeidler C, Klimenkova O, Klimiankou M, Unalan M, Kandabarau S, Makaryan V, Beekman R, Behrens K, Stocking C, Obenauer J, Schnittger S, Kohlmann A, Valkhof MG, Hoogenboezem R, Göhring G, Reinhardt D, Schlegelberger B, Stanulla M, Vandenberghe P, Donadieu J, Zwaan CM, Touw IP, van den Heuvel-Eibrink MM, Dale DC, Welte K.

17. Januar 2014 (Forschungsverbund FACE)
Birth prevalence and initial treatment of Robin sequence in Germany: a prospective epidemiologic study
Orphanet Journal of Rare Diseases 2014, 9:9
Vatlach S, Maas C, Poets CF

2013

26. Dezember 2013: (Forschungsverbund PID-NET)
Deficiency of innate and acquired immunity caused by an IKBKB mutation.
Pannicke U, Baumann B, Fuchs S, Henneke P, Rensing-Ehl A, Rizzi M, Janda A, Hese K, Schlesier M, Holzmann K, Borte S, Laux C, Rump EM, Rosenberg A, Zelinski T, Schrezenmeier H, Wirth T, Ehl S, Schroeder ML, Schwarz K. N Engl J Med. 2013 Dec 26;369(26):2504-14.

15. November 2013: (Forschungsverbund IonNeurONet)
Transient compartment-like syndrome and normokalaemic periodic paralysis due to a Cav1.1 mutation.
Fan C, Lehmann-Horn F, Weber MA, Bednarz M, Groome JR, Jonsson MK, Jurkat-Rott K.
Brain. 2013 Nov 15. [Epub ahead of print]

9. Oktober 2013: (Forschungsverbund Imprinting)
Deep bisulfite sequencing of aberrantly methylated Loci in a patient with multiple methylation defects.
Beygo J, Ammerpohl O, Gritzan D, Heitmann M, Rademacher K, Richter J, Caliebe A, Siebert R, Horsthemke B, Buiting K.
PLoS One. 2013 Oct 9;8(10):e76953. doi: 10.1371/journal.pone.0076953.

17. September 2013: (Forschungsverbund NIRK)
Topical Enzyme-Replacement Therapy Restores Transglutaminase 1 Activity and Corrects Architecture of Transglutaminase-1-Deficient Skin Grafts.
Aufenvenne K, Larcher F, Hausser I, Duarte B, Oji V, Nikolenko H, Del Rio M, Dathe M, Traupe H.
Am J Hum Genet. 2013 Sep 17

30. August 2013: (Forschungsverbund MND-Net)
Quality of life in fatal disease: the flawed judgement of the social environment.
Lulé D, Ehlich B, Lang D, Sorg S, Heimrath J, Kübler A, Birbaumer N, Ludolph AC
J Neurol. 2013 Aug 30. [Epub ahead of print]

9. Juli 2013: (Forschungsverbund mitoNet)
ELAC2 Mutations Cause a Mitochondrial RNA Processing Defect Associated with Hypertrophic Cardiomyopathy.
Haack TB, Kopajtich R, Freisinger P, Wieland T, Rorbach J, Nicholls TJ, Baruffini E, Walther A, Danhauser K, Zimmermann FA, Husain RA, Schum J, Mundy H, Ferrero I, Strom TM, Meitinger T, Taylor RW, Minczuk M, Mayr JA, Prokisch H.
Am J Hum Genet. 2013 Jul 9

19. Juni 2013: (Forschungsverbund MD-Net)
Dystrophin-deficient pigs provide new insights into the hierarchy of physiological derangements of dystrophic muscle.
Klymiuk N, Blutke A, Graf A, Krause S, Burkhardt K, Wuensch A, Krebs S, Kessler B, Zakhartchenko V, Kurome M, Kemter E, Nagashima H, Schoser B, Herbach N, Blum H, Wanke R, Aartsma-Rus A, Thirion C, Lochmüller H, Walter MC, Wolf E.
Hum Mol Genet. 2013 Jun 19.