List of Publications

Prof. Bodo Grimbacher

Vice Director of the Institute of Immunodeficiency, Freiburg, Germany
Alumni of Marie-Curie-Researchers and Emmy-Nöther Fellows

PEER-REVIEWED ARTICLES (TOTAL 274)

TOTAL IMPACT FACTOR: 2,807.885

LAST-AUTHOR PUBLICATIONS:

87. Andreani V, Forde AJ, Fliegauf M, Bressan G, Noé V, Ott N, Saghafi S, Vornholz L, Isay SE, Ruland J, Henneke P, **Grimbacher B.**
STAT3 haploinsufficiency is associated with autosomal dominant hyper-IgE syndrome.
Sci Adv. 2025 Aug 29;11(35):eadw2464. **Impact Factor: 12.060**

86. Chandrasekaran P, Krausz M, Han Y, Mitsuiki N, Gabrysch A, Nöltner C, Proietti M, Heller T, Grou C, Calderon V, Subramanian P, Jones DR, Siu Y, Deming C, Conlan S, Holland SM, Segre JA, Uzel G, **Grimbacher B***, Falcone EL.* (*contributed equally)
The intestinal microbiome and metabolome discern disease severity in cytotoxic T-lymphocyte-associated protein 4 deficiency.
Microbiome. 2025 Feb 11;13(1):51. **Impact Factor: 13.800**

85. Posadas-Cantera S, Mitsuiki N, Emmerich F, Patiño V, Lorenz HM, Neth O, Dybedal I, Taskén K, Schäffer AA, **Grimbacher B***, Gámez-Díaz L*. (*contributed equally)
The effect of HLA genotype on disease onset and severity in CTLA-4 insufficiency.
Front Immunol. 2025 Jan 6;15:1447995. **Impact Factor: 5.700**

84. Yang L, Zerbato B, Pessina A, Brambilla L, Andreani V, Frey-Jakobs S, Fliegauf M, Barbouche MR, Zhang Q, Chiaradonna F, Proietti M, Du X, **Grimbacher B**.
PGM3 insufficiency: a glycosylation disorder causing a notable T cell defect.
Front Immunol. 2024 Dec 24;15:1500381. **Impact Factor: 5.700**

83. Ramirez NJ, Schulze JJ, Walter S, Werner J, Mrovecova P, Olek S, Sachsenmaier C, **Grimbacher B***, Salzer U*. (*contributed equally)
Epigenetic immune cell quantification for diagnostic evaluation and monitoring of patients with inborn errors of immunity and secondary immune deficiencies.
Clin Immunol. 2024 Mar;260:109920. **Impact Factor: 9.100**

82. Rolles B, Caballero-Oteyza A, Proietti M, Goldacker S, Warnatz K, Camacho-Ordonez N, Prader S, Schmid JP, Vieri M, Isfort S, Meyer R, Kirschner M, Brümmendorf TH, Beier F, **Grimbacher B.**
Telomere biology disorders may manifest as common variable immunodeficiency (CVID).
Clin Immunol. 2023 Dec;257:109837. **Impact Factor: 8.600**

81. Rojas-Restrepo J, Sindram E, Zenke S, Haberstroh H, Mitsuiki N, Gabrysch A, Huebscher K, Posadas-Cantera S, Krausz M, Kobbe R, Rohr JC, **Grimbacher B**, Gámez-Díaz L.
Functional Relevance of CTLA4 Variants: an Upgraded Approach to Assess CTLA4-Dependent Transendocytosis by Flow Cytometry.
J Clin Immunol. 2023 Nov;43(8):2076-2089. **Impact Factor: 8.137**

80. Vanselow S, Hanitsch L, Hauck F, Körholz J, Maccari ME, Meinhardt A, Sogkas G, Schuetz C, **Grimbacher B**.
Future Directions in the Diagnosis and Treatment of APDS and IEI: a Survey of German IEI Centers.
Front Immunol. 2023 Oct 5;14:1279652. **Impact Factor: 7.300**

79. Peng XP, Al-Ddafari MS, Caballero-Oteyza A, El Mezouar C, Mrovecova P, Dib SE, Massen Z, Smahi MC, Faiza A, Hassaïne RT, Lefranc G, Aribi M, **Grimbacher B.**
Next generation sequencing (NGS)-based approach to diagnosing Algerian patients with suspected inborn errors of immunity (IEIs).
Clin Immunol. 2023 Sep 9;256:109758. Online ahead of print. **Impact Factor: 8.600**

78. Nöltner C, Bulashevska A, Hübscher K, Haberstroh H, **Grimbacher B***, Proietti M*. (*contributed equally)
Fecal Immunoglobulin Levels as a Modifier of the Gut Microbiome in Patients with Common Variable Immunodeficiency.
J Clin Immunol. 2023 Aug;43(6):1208-1220. Epub 2023 Mar 24. **Impact Factor: 8.137**

77. Haberstroh H, Hirsch A, Goldacker S, Zessack N, Warnatz K, **Grimbacher B***, Salzer U*. (*contributed equally)
A Toolkit for Monitoring Immunoglobulin G Levels from Dried Blood Spots of Patients with Primary Immunodeficiencies.
J Clin Immunol. 2023 Aug;43(6):1185-1192. Epub 2023 Mar 21. **Impact Factor: 8.137**

76. Staus P, Rusch S, El-Helou S, Müller G, Krausz M, Geisen U, Caballero-Oteyza A, Krüger R, Bakhtiar S, Lee-Kirsch MA, Fasshauer M, Baumann U, Hoyer BF, Farela Neves J, Borte M, Carrabba M, Hauck F, Ehl S, Bader P, von Bernuth H, Atschekzei F, Seppänen MRJ, Warnatz K, Nieters A, Kindle G, **Grimbacher B.**
The GAIN Registry - a New Prospective Study for Patients with Multi-organ Autoimmunity and Autoinflammation.
J Clin Immunol. 2023 Aug;43(6):1289-1301. **Impact Factor: 8.137**

PEER-REVIEWED ARTICLES (2)

75. Lim YW, Ramirez NJ, Asensio MA, Chiang Y, Müller G, Mrovecova P, Mitsuiki N, Krausz M, Camacho-Ordonez N, Warnatz K, Adler AS, **Grimbacher B.**
Sequencing the B Cell Receptor Repertoires of Antibody-Deficient Individuals With and Without Infection Susceptibility.
J Clin Immunol. 2023 Jul;43(5):940-950. **Impact Factor: 8.137**

74. Krausz M, Mitsuiki N, Falcone V, Komp J, Posadas-Cantera S, Lorenz HM, Litzman J, Wolff D, Kanariou M, Heinkele A, Speckmann C, Häcker G, Hengel H, Gámez-Díaz L, **Grimbacher B**.
Do common infections trigger disease-onset or -severity in CTLA-4 insufficiency?
Front Immunol. 2022 Nov 2;13:1011646. **Impact Factor: 7.300**

73. Krausz M, Uhlmann A, Rump IC, Ihorst G, Goldacker S, Sogkas G, Posadas-Cantera S, Schmidt R, Feißt M, Alsina L, Dybedal I, Recher M, Warnatz K, **Grimbacher B**.
The ABACHAI clinical trial protocol: Safety and efficacy of abatacept (s.c.) in patients with CTLA-4 insufficiency or LRBA deficiency: A non controlled phase 2 clinical trial.
Contemp Clin Trials Commun. 2022 Sep 24;30:101008. **Impact Factor: 1.653**

72. Fliegauf M, Kinnunen M, Posadas-Cantera S, Camacho-Ordonez N, Abolhassani H, Alsina L, Atschekzei F, Bogaert DJ, Burns SO, Church JA, Dückers G, Freeman AF, Hammarström L, Hanitsch LG, Kerre T, Kobbe R, Sharapova SO, Siepermann K, Speckmann C, Steiner S, Verma N, Walter JE, Westermann-Clark E, Goldacker S, Warnatz K, Varjosalo M, **Grimbacher B**.
Detrimental NFKB1 missense variants affecting the Rel-homology domain of p105/p50.
Front Immunol. 2022 Aug 29;13:965326. **Impact Factor: 7.300**

71. Ramirez N, Posadas-Cantera S, Langer N, de Oteyza ACG, Proietti M, Keller B, Zhao F, Gernedl V, Pecoraro M, Eibel H, Warnatz K, Ballestar E, Geiger R, Bossen C, **Grimbacher B**.
Multi-omics analysis of naïve B cells of patients harboring the C104R mutation in TACI.
Front Immunol. 2022 Aug 16;13:938240. **Impact Factor: 7.300**

70. Rojas-Restrepo J, Caballero-Oteyza A, Huebscher K, Haberstroh H, Fliegauf M, Keller B, Kobbe R, Warnatz K, Ehl S, Proietti M, **Grimbacher B.**
Establishing the Molecular Diagnoses in a Cohort of 291 Patients With Predominantly Antibody Deficiency by Targeted Next-Generation Sequencing: Experience From a Monocentric Study.
Front Immunol. 2021 Dec 17;12:786516. **Impact Factor: 6.429**

69. van Schewick CM, Lowe DM, Burns SO, Workman S, Symes A, Guzman D, Moreira F, Watkins J, Clark I, **Grimbacher B.**
Bowel Histology of CVID Patients Reveals Distinct Patterns of Mucosal Inflammation.
J Clin Immunol. 2021 Oct 1. Online ahead of print. **Impact Factor: 6.780**

68. Frede N, Rojas-Restrepo J, Caballero Garcia de Oteyza A, Buchta M, Hübscher K, Gámez-Díaz L, Proietti M, Saghafi S, Chavoshzadeh Z, Soler-Palacin P, Galal N, Adeli M, Aldave-Becerra JC, Al-Ddafari MS, Ardenyz Ö, Atkinson TP, Kut FB, Çelmeli F, Rees H, Kilic SS, Kirovski I, Klein C, Kobbe R, Korganow AS, Lilic D, Lunt P, Makwana N, Metin A, Özgür TT, Karakas AA, Seneviratne S, Sherkat R, Sousa AB, Unal E, Patiroglu T, Wahn V, von Bernuth H, Whiteford M, Doffinger R, Jouhadi Z, **Grimbacher B**.
Genetic Analysis of a Cohort of 275 Patients with Hyper-IgE Syndromes and/or Chronic Mucocutaneous Candidiasis.
J Clin Immunol. 2021 Nov;41(8):1804-1838. **Impact Factor: 6.780**

67. Egg D, Rump IC, Mitsuiki N, Rojas-Restrepo J, Maccari ME, Schwab C, Gabrysch A, Warnatz K, Goldacker S, Patiño V, Wolff D, Okada S, Hayakawa S, Shikama Y, Kanda K, Imai K, Sotomatsu M, Kuwashima M, Kamiya T, Morio T, Matsumoto K, Mori T, Yoshimoto Y, Dybedal I, Kanariou M, Kucuk ZY, Chapdelaine H, Petruzelkova L, Lorenz HM, Sullivan KE, Heimall J, Moutschen M, Litzman J, Recher M, Albert MH, Hauck F, Seneviratne S, Pachlopnik Schmid J, Kolios A, Unglik G, Klemann C, Snapper S, Giulino-Roth L, Svaton M, Platt CD, Hambleton S, Neth O, Gosse G, Reinsch S, Holzinger D, Kim YJ, Bakhtiar S, Atschekzei F, Schmidt R, Sogkas G, Chandrakasan S, Rae W, Derfalvi B, Marquart HV, Ozen A, Kiykim A, Karakoc-Aydiner E, Králíčková P, de Bree G, Kiritsi D, Seidel MG, Kobbe R, Dantzer J, Alsina L, Armangue T, Lougaris V, Agyeman P, Nyström S, Buchbinder D, Arkwright PD, **Grimbacher B.**
Therapeutic options for CTLA-4 insufficiency.
J Allergy Clin Immunol. 2021 Jun 7:S0091-6749(21)00891-5. **Impact Factor: 10.793**

66. Fliegauf M, Krüger R, Steiner S, Hanitsch LG, Büchel S, Wahn V, von Bernuth H, **Grimbacher B**.
A Pathogenic Missense Variant in NFKB1 Causes Common Variable Immunodeficiency Due to Detrimental Protein Damage.
Front Immunol. 2021 Apr 27;12:621503. doi: 10.3389/fimmu.2021.621503. **Impact Factor: 6.429**

65. van Schewick CM, Nöltner C, Abel S, Burns SO, Workman S, Symes A, Guzman D, Proietti M, Bulashevska A, Moreira F, Soetedjo V, Lowe DM, **Grimbacher B.**
Altered Microbiota, Impaired Quality of Life, Malabsorption, Infection, and Inflammation in CVID Patients With Diarrhoea
Front Immunol. 2020 Jul 31;11:1654. **Impact Factor: 6.429**

64. Lorenzini T, Fliegauf M, Klammer N, Frede N, Proietti M, Bulashevska A, Camacho-Ordonez N, Varjosalo M, Kinnunen M, de Vries E, van der Meer JWM, Ameratunga R, Roifman CM, Schejter YD, Kobbe R, Hautala T, Atschekzei F, Schmidt RE, Schröder C, Stepensky P, Shadur B, Pedroza LA, van der Flier M, Martínez-Gallo M, Gonzalez-Granado LI, Allende LM, Shcherbina A, Kuzmenko N, Zakharova V, Neves JF, Svec P, Fischer U, Ip W, Bartsch O, Barış S, Klein C, Geha R, Chou J, Alosaimi M, Weintraub L, Boztug K, Hirschmugl T, Dos Santos Vilela MM, Holzinger D, Seidl M, Lougaris V, Plebani A, Alsina L, Piquer-Gibert M, Deyà-Martínez A, Slade CA,

PEER-REVIEWED ARTICLES (3)

Aghamohammadi A, Abolhassani H, Hammarström L, Kuismin O, Helminen M, Allen HL, Thaventhiran JE, Freeman AF, Cook M, Bakhtiar S, Christiansen M, Cunningham-Rundles C, Patel NC, Rae W, Niehues T, Brauer N, Syrjänen J, Seppänen MRJ, Burns SO, Tuijnenburg P, Kuijpers TW; NIHR-BioResource – Rare Diseases Consortium, Warnatz K, **Grimbacher B**.
Characterization of the clinical and immunological phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations.
J Allergy Clin Immunol. 2020 Apr 9. **Impact Factor: 10.793**

63.El-Helou SM, Biegner AK, Bode S, Ehl SR, Heeg M, Maccari ME, Ritterbusch H, Speckmann C, Rusch S, Scheible R, Warnatz K, Atschekzei F, Beider R, Ernst D, Gerschmann S, Jablonka A, Mielke G, Schmidt RE, Schürmann G, Sogkas G, Baumann UH, Klemann C, Viemann D, von Bernuth H, Krüger R, Hanitsch LG, Scheibenbogen CM, Wittke K, Albert MH, Eichinger A, Hauck F, Klein C, Rack-Hoch A, Sollinger FM, Avila A, Borte M, Borte S, Fasshauer M, Hauenherm A, Kellner N, Müller AH, Ülzen A, Bader P, Bakhtiar S, Lee JY, Heß U, Schubert R, Wölke S, Zielen S, Ghosh S, Laws HJ, Neubert J, Oommen PT, Hönig M, Schulz A, Steinmann S, Schwarz K, Dückers G, Lamers B, Langemeyer V, Niehues T, Shai S, Graf D, Müglich C, Schmalzing MT, Schwaneck EC, Tony HP, Dirks J, Haase G, Liese JG, Morbach H, Foell D, Hellige A, Wittkowski H, Masjosthusmann K, Mohr M, Geberzahn L, Hedrich CM, Müller C, Rösen-Wolff A, Roesler J, Zimmermann A, Behrends U, Rieber N, Schauer U, Handgretinger R, Holzer U, Henes J, Kanz L, Boesecke C, Rockstroh JK, Schwarze-Zander C, Wasmuth JC, Dilloo D, Hülsmann B, Schönberger S, Schreiber S, Zeuner R, Ankermann T, von Bismarck P, Huppertz HI, Kaiser-Labusch P, Greil J, Jakoby D, Kulozik AE, Metzler M, Naumann-Bartsch N, Sobik B, Graf N, Heine S, Kobbe R, Lehmberg K, Müller I, Herrmann F, Horneff G, Klein A, Peitz J, Schmidt N, Bielack S, Groß-Wieltsch U, Classen CF, Klasen J, Deutz P, Kamitz D, Lassay L, Tenbrock K, Wagner N, Bernbeck B, Brummel B, Lara-Villacanas E, Münstermann E, Schneider DT, Tietsch N, Westkemper M, Weiß M, Kramm C, Kühnle I, Kullmann S, Girschick H, Specker C, Vinnemeier-Laubenthal E, Haenicke H, Schulz C, Schweigerer L, Müller TG, Stiefel M, Belohradsky BH, Soetedjo V, Kindle G, **Grimbacher B**.
The German National Registry of Primary Immunodeficiencies (2012-2017).
Front Immunol. 2019 Jul 19 **Impact factor: 5.511**

62. Eskandarian Z, Fliegauf M, Bulashevska A, Proietti M, Hague R, Smulski CR, Schubert D, Warnatz K, **Grimbacher B**.
Assessing the Functional Relevance of Variants in the IKAROS Family Zinc Finger Protein 1(IKZF1) in a Cohort of Patients With Primary Immunodeficiency.
Front Immunol. 2019 Apr 16 **Impact factor: 5.511**

61. Klemann C, Camacho-Ordonez N, Yang L, Eskandarian Z, Rojas-Restrepo JL, Frede N, Bulashevska A, Heeg M, Al-Ddafari MS, Premm J, Seidl M, Ammann S, Sherkat R, Radhakrishnan N, Warnatz K, Unger S, Kobbe R, Hüfner A, Leahy TR, Ip W, Burns SO, Fliegauf M, **Grimbacher B**.
Clinical and Immunological Phenotype of Patients With Primary Immunodeficiency Due to Damaging Mutations in *NFKB2*.
Front Immunol. 2019 Mar 19 **Impact factor: 5.511**

60. Egg D, Schwab C, Gabrysch A, Arkwright PD, Cheesman E, Giulino-Roth L, Neth O, Snapper S, Okada S, Moutschen M, Delvenne P, Pecher AC, Wolff D, Kim YJ, Seneviratne S, Kim KM, Kang JM, Ojaimi S, McLean C, Warnatz K, Seidl M, **Grimbacher B**.
Increased Risk for Malignancies in 131 Affected CTLA4 Mutation Carriers.
Front Immunol. 2018 Sep 10 **Impact factor: 5.511**

59. Scheuerlein P, Pietsch L, Camacho-Ordonez N, Reiser V, Patel S, Burns SO, Warnatz K, **Grimbacher B**.
Is It Safe to Switch From Intravenous Immunoglobulin to Subcutaneous Immunoglobulin in Patients With Common Variable Immunodeficiency and Autoimmune Thrombocytopenia?
Front Immunol. 2018 Jul 19;9:1656 . **Impact factor: 5.511**

58. Frey-Jakobs S, Hartberger JM, Fliegauf M, Bossen C, Wehmeyer ML, Neubauer JC, Bulashevska A, Proietti M, Fröbel P, Nöltner C, Yang L, Rojas-Restrepo J, Langer N, Winzer S, Engelhardt KR, Glocker C, Pfeifer D, Klein A, Schäffer AA, Lagovsky I, Lachover-Roth I, Béziat V, Puel A, Casanova JL, Fleckenstein B, Weidinger S, Kilic SS, Garty BZ, Etzioni A, **Grimbacher B**.
ZNF341 controls STAT3 expression and thereby immunocompetence.
Sci Immunol. 2018 Jun 15;3(24). **Impact factor: 10.551**

57. Gámez-Díaz L, Sigmund EC, Reiser V, Vach W, Jung S, **Grimbacher B**.
Rapid Flow Cytometry-Based Test for the Diagnosis of Lipopolysaccharide Responsive Beige-Like Anchor (LRBA) Deficiency.
Front Immunol. 2018 Apr 23;9:720. **Impact factor: 6.429**

56. Schwab C, Gabrysch A, Olbrich P, Patiño V, Warnatz K, Wolff D, Hoshino A, Kobayashi M, Imai K, Takagi M, Dybedal I, Haddock JA, Sansom DM, Lucena JM, Seidl M, Schmitt-Graeff A, Reiser V, Emmerich F, Frede N, Bulashevska A, Salzer U, Schubert D, Hayakawa S, Okada S, Kanariou M, Kucuk ZY, Chapdelaine H, Petruzelkova L, Sumnik Z, Sediva A, Slatter M, Arkwright PD, Cant A, Lorenz HM, Giese T, Lougaris V, Plebani A, Price C, Sullivan KE, Moutschen M, Litzman J, Freiberger T, van de Veerdonk FL, Recher M, Albert MH, Hauck F, Seneviratne S, Pachlopnik Schmid J, Kolios A, Unglik G, Klemann C, Speckmann C, Ehl S, Leichtner A, Blumberg R, Franke A, Snapper S, Zeissig S, Cunningham-Rundles C, Giulino-Roth L, Elemento O, Dückers G, Niehues T, Fronkova E, Kanderová V, Platt CD, Chou J, Chatila TA, Geha R, McDermott E, Bunn S, Kurzai M, Schulz A, Alsina L, Casals F, Deyà-Martinez A, Hambleton S, Kanegane H, Taskén K, Neth O, **Grimbacher B**.
Phenotype, penetrance, and treatment of 133 cytotoxic T-lymphocyte antigen 4-insufficient subjects.
J Allergy Clin Immunol. 2018 May 4. **Impact factor: 13.081**

PEER-REVIEWED ARTICLES (4)

55. Petersen BS, August D, Abt R, Alddafari M, Atarod L, Baris S, Bhavsar H, Brinkert F, Buchta M, Bulashevska A, Chee R, Cordeiro AI, Dara N, Dückers G, Elmarsafy A, Frede N, Galal N, Gerner P, Glocker EO, Goldacker S, Hammermann J, Hasselblatt P, Havlicekova Z, Hübscher K, Jesenak M, Karaca NE, Karakoc-Aydiner E, Kharaghani MM, Kilic SS, Kiykim A, Klein C, Klemann C, Kobbe R, Kotlarz D, Laass MW, Leahy TR, Mesdaghi M, Mitton S, Neves JF, Öztürk B, Pereira LF, Rohr J, Restrepo JLR, Ruzaike G, Saleh N, Seneviratne S, Senol E, Speckmann C, Tegtmeyer D, Thankam P, van der Werff Ten Bosch J, von Bernuth H, Zeissig S, Zeissig Y, Franke A, **Grimbacher B**.
Targeted Gene Panel Sequencing for Early-onset Inflammatory Bowel Disease and Chronic Diarrhea.
Inflamm Bowel Dis. 2017 Dec;23(12):2109-2120. **Impact factor: 4.525**

54. Schepp J, Chou J, Skrabl-Baumgartner A, Arkwright PD, Engelhardt KR, Hambleton S, Morio T, Röther E, Warnatz K, Geha R, **Grimbacher B**.
14 Years after Discovery: Clinical Follow-up on 15 Patients with Inducible Co-Stimulator Deficiency.
Front Immunol. 2017 Aug 16;8:964. **Impact factor: 6.429**

53. Schubert D, Klein MC, Hassdenteufel S, Caballero-Oteyza A, Yang L, Proietti M, Bulashevska A, Kemming J, Kühn J, Winzer S, Rusch S, Fliegauf M, Schäffer AA, Pfeffer S, Geiger R, Cavalié A, Cao H, Yang F, Li Y, Rizzi M, Eibel H, Kobbe R, Marks AL, Peppers BP, Hostoffer RW, Puck JM, Zimmermann R, **Grimbacher B**.
Plasma cell deficiency in human subjects with heterozygous mutations in Sec61 translocon alpha 1 subunit (SEC61A1).
J Allergy Clin Immunol. 2017 Aug 4 **Impact factor: 13.081**

52. Pott MC, Frede N, Wanders J, Hammarström L, Glocker EO, Glocker C, Tahami F, **Grimbacher B**.
Autoantibodies against BAFF, APRIL or IL21 - an alternative pathogenesis for antibody-deficiencies?
BMC Immunol. 2017 Jun 26;18(1):34. **Impact factor: 2.485**

51. Schepp J, Proietti M, Frede N, Buchta M, Hübscher K, Rojas Restrepo J, Goldacker S, Warnatz K, Pachlopnik Schmid J, Duppenthaler A, Lougaris V, Uriarte I, Kelly S, Hershfield M, **Grimbacher B**.
Screening of 181 Patients With Antibody Deficiency for Deficiency of Adenosine Deaminase 2 Sheds New Light on the Disease in Adulthood.
Arthritis Rheumatol. 2017 Aug;69(8):1689-1700. Epub 2017 Jul 5. **Impact factor: 6.918**

50. Schepp J, Bulashevska A, Mannhardt-Laakmann W, Cao H, Yang F, Seidl M, Kelly S, Hershfield M, **Grimbacher B**.
Deficiency of Adenosine Deaminase 2 Causes Antibody Deficiency.
J Clin Immunol. 2016 Apr;36(3):179-86. Epub 2016 Feb 27. **Impact factor: 3.184**

49. Gámez-Díaz L, August D, Stepensky P, Revel-Vilk S, Seidel MG, Noriko M, Morio T, Worth AJ, Blessing J, Van de Veerdonk F, Feuchtinger T, Kanariou M, Schmitt-Graeff A, Jung S, Seneviratne S, Burns S, Belohradsky BH, Rezaei N, Bakhtiar S, Speckmann C, Jordan M, **Grimbacher B**.
The extended phenotype of LPS-responsive beige-like anchor protein (LRBA) deficiency.
J Allergy Clin Immunol. 2016 Jan;137(1):223-230. **Impact factor: 11.476**

48. Celmeli F, Oztoprak N, Turkkahraman D, Seyman D, Mutlu E, Frede N, Köksoy S, **Grimbacher B**.
Successful Granulocyte Colony Stimulating Factor Treatment of Relapsing Candida albicans Meningoencephalitis Caused by CARD9 Deficiency.
Pediatr Infect Dis J. 2016 Apr;35(4):428-31. **Impact factor: 2.723**

47. Depner M, Fuchs S, Raabe J, Frede N, Glocker C, Doffinger R, Gkrania-Klotsas E, Kumararatne D, Atkinson TP, Schroeder HW Jr, Niehues T, Dückers G, Stray-Pedersen A, Baumann U, Schmidt R, Franco JL, Orrego J, Ben-Shoshan M, McCusker C, Jacob CM, Carneiro-Sampaio M, Devlin LA, Edgar JD, Henderson P, Russell RK, Skytte AB, Seneviratne SL, Wanders J, Stauss H, Meyts I, Moens L, Jesenak M, Kobbe R, Borte S, Borte M, Wright DA, Hagin D, Torgerson TR, **Grimbacher B**. The Extended Clinical Phenotype of 26 Patients with Chronic Mucocutaneous Candidiasis due to Gain-of-Function Mutations in STAT1.
J Clin Immunol. 2016 Jan;36(1):73-84. Epub 2015 Nov 25. **Impact factor: 2.654**

46. Volk T, Pannicke U, Reisli I, Bulashevska A, Ritter J, Björkman A, Schäffer AA, Fliegauf M, Sayar EH, Salzer U, Fisch P, Pfeifer D, Di Virgilio M, Cao H, Yang F, Zimmermann K, Keles S, Caliskaner Z, Güner Ş, Schindler D, Hammarström L, Rizzi M, Hummel M, Pan-Hammarström Q, Schwarz K, **Grimbacher B**.
DCLRE1C (ARTEMIS) mutations causing phenotypes ranging from atypical severe combined immunodeficiency to mere antibody deficiency.
Hum Mol Genet. 2015 Dec 20;24(25):7361-72. Epub 2015 Oct 16. **Impact factor: 6.677**

45. Elgizouli M, Lowe DM, Speckmann C, Schubert D, Hülsdünker J, Eskandarian Z, Dudek A, Schmitt-Graeff A, Wanders J, Jørgensen SF, Fevang B, Salzer U, Nieters A, Burns S, **Grimbacher B**.
Activating PI3Kδ mutations in a cohort of 669 patients with primary immunodeficiency.
Clin Exp Immunol. 2016 Feb;183(2):221-9. Epub 2015 Nov 9. **Impact factor: 3.278**

44. Fliegauf M, Bryant VL, Frede N, Slade C, Woon ST, Lehnert K, Winzer S, Bulashevska A, Scerri T, Leung E, Jordan A, Keller B, de Vries E, Cao H, Yang F, Schäffer AA, Warnatz K, Browett P, Douglass J, Ameratunga RV, van der Meer JW, **Grimbacher B**.
Haploinsufficiency of the NF-κB1 Subunit p50 in Common Variable Immunodeficiency.
Am J Hum Genet. 2015 Sep 3;97(3):389-403. Epub 2015 Aug 13. **Impact factor: 10.987**

43. Dziadzio M, Ammann S, Canning C, Boyle F, Hassan A, Cale C, Elawad M, Fiil BK, Gyrd-Hansen M, Salzer U, Speckmann C, **Grimbacher B**.
Symptomatic Males and Female Carriers in a Large Caucasian Kindred with XIAP Deficiency.
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