

Supplementary Table 1. List of publications from participating JMF CEE network Centers

Austria

Schatorjé E, van der Flier M, Seppänen M, Browning M, Morsheimer M, Henriët S, Neves JF, Vinh DC, Alsina L, Grumach A, Soler-Palacin P, Boyce T, Celmeli F, Goudouris E, Hayman G, Herriot R, Förster-Waldl E, Seidel M, Simons A, de Vries E. [Primary immunodeficiency associated with chromosomal aberration - an ESID survey](#). Orphanet J Rare Dis. 2016 Aug 2;11(1):110.

Szilagyi K, Gazendam RP, van Hamme JL, Tool AT, van Houdt M, Vos WA, Verkuijlen P, Janssen H, Belot A, Juillard L, Förster-Waldl E, Boztug K, Kraal G, de Winther MP, Kuijpers TW, van den Berg TK. [Impaired microbial killing by neutrophils from patients with protein kinase C delta deficiency](#). J Allergy Clin Immunol. 2015 Nov;136(5):1404-7.e1-10

Woutsas S, Aytekin C, Salzer E, Conde CD, Apaydin S, Pichler H, Memaran-Dadgar N, Hosnut FO, Förster-Waldl E, Matthes S, Huber WD, Lion T, Holter W, Bilic I, Boztug K. [Hypomorphic mutation in TTC7A causes combined immunodeficiency with mild structural intestinal defects](#). Blood. 2015 Mar 5;125(10):1674-6.

Seidel MG, Duerr C, Woutsas S, Schwerin-Nagel A, Sadeghi K, Neesen J, Uhrig S, Santos-Valente E, Pickl WF, Schwinger W, Urban C, Boztug K, Förster-Waldl E. [A novel immunodeficiency syndrome associated with partial trisomy 19p13](#). J Med Genet. 2014 Apr;51(4):254-63.

Azerbaijan

Jannik S Glasmacher et al.. **Wiskott-Aldrich Syndrome: A Retrospective Study on 575 Patients Analyzing the Impact of Splenectomy, Stem Cell Transplantation, or No Definitive Treatment on Frequency of Disease-Related Complications and Physician-Perceived Quality of Life**. Blood. 2016 128:366

Belarus

Sharapova SO, A. Migas, I. Guryanova, S. Aleshkevich, S. Kletski, A. Durandy, M. Belevtsev. **Late-onset combined immune deficiency associated to skin granuloma due to heterozygous compound mutations in RAG1 gene in a 14 years old male**. Human Immunology. 2013 Jan;74(1):18-22

Sharapova SO, Guryanova IE, Pashchenko OE, et al...Belevtsev M.V., Aleinikova OV. **Molecular Characteristics, Clinical and Immunologic Manifestations of 11 Children with Omenn Syndrome in East Slavs** (Russia, Belarus, Ukraine). Journal Clinical Immunology. 2016 Jan, 36(1):46-55

Sharapova SO, Pashchenko OE, Guryanova IE, Migas AA, Kondratenko IV, Aleinikova OV. **Recent thymic emigrants, T regulatory cells, and BAFF level in children with X-linked agammaglobulinaemia in association with chronic respiratory disease.**Allergol Immunopathol (Madr). 2018 Jan - Feb;46(1):58-66

Sharapova SO, Haapaniemi E, Sakovich IS, Rojas J, Gámez-Díaz L, Mareika YE, Guryanova IE, Migas AA, Mikhaleuskaya TM, Grimbacher B, Aleinikova OV. **Novel LRBA Mutation and Possible Germinal Mosaicism in a Slavic Family.**J Clin Immunol. 2018 May;38(4):471-474

Sharapova SO, Fedorova AS, Pashchenko OE, Vahliarskaya SS, Guryanova IE, Migas AA, Kondratenko IV, Aleinikova OV. **Novel Mutations in SH2D1A Gene in X-linked Lymphoproliferative Syndrome, Diagnosed After B-Cell Non-Hodgkin Lymphoma.** J Pediatr Hematol Oncol. 2017 May;39(4):e203-e206

Bulgaria

Baleva, M.P., S. Mihaylova, P. Yankova, I. Atanasova, M. Nikolova-Vlahova, E.Naumova. **A rare case of Addison's disease, hepatitis, thyreoiditis, positive IgG anti-tissue transglutaminase antibodies and partial IgA deficiency.** Central Eur J of Immunol, 41(2), 2016

Baleva M, Lesichkova S, Gesheva N, Mihailova S, Gerova V, Vladimirov B, Pemchev P, Nikolova – Vlahova M, Naumova E. **Common variable immune deficiency, central diabetes insipidus and anemia.** Accepted for publication in Central European Journal of Immunology, 2019

Wu Kevin, Purswani Pooja , Ujhazi Boglarka, Mihailova Snezhina , Naumova Elissaveta, Stefanov Stefan , Savic Sinisa , Sargur Ravishankar , Milojevic Diana , Jolan Walter. **Clinical**

Challenges in Recombination Activating Gene Deficiency and Arthritis. Frontiers in Pediatrics, in press

Croatia

Grubic Z, Stingl Jankovic K, Kelecic J, Batinic D, Dubravcic K, Zunec R. [A case of maternal-foetal chimerism identified during routine histocompatibility testing for hematopoietic stem cell transplantation.](#) Int J Immunogenet. 2016 Feb;43(1):1-7.

Tomac G, Bojanić I, Mazić S, Vidović I, Raos M, Čepulić BG, Seiwerth RS, Kelečić J, Labar B. [Haemolysis, pure red cell aplasia and red cell antibody formation associated with major and bidirectional ABO incompatible haematopoietic stem cell transplantation.](#) Blood Transfus 2018 Jul;16(4):397-404.

Czech Republic

Kralickova P, Milota T, Haviger J, Litzman J, Malkusova I, Jilek D, Petanová J, Vydlakova J, Zimulova A, Fronkova E, Svaton M, Kanderova V, Kalina T, Bloomfield M, Parackova Z, Klocperk A, Sediva A. **VID-associated tumors: Czech nationwide study focused on epidemiology, immunology and genetic background in a cohort of patients with CVID.** Frontiers in Immunology, 2019 Jan 22;9:3135

Kanderova V, Grombirikova H, Zentsova I, Reblova K, Klocperk A, Fejtkova M, Bloomfield M, Ravcukova B, Kalina T, Freiburger T, Sediva A. [Lymphoproliferation, immunodeficiency and early-onset inflammatory bowel disease associated with a novel mutation in Caspase 8.](#) Haematologica. 2019 Jan;104(1):e32-e34

Klocperk A, Paračková Z, Bloomfield M, Rataj M, Pokorný J, Unger S, Warnatz K, Šedivá A. [Follicular Helper T Cells in DiGeorge Syndrome.](#) Front Immunol. 2018 Jul 23;9:1730

Bloomfield M, Kanderová V, Paračková Z, Vrabcová P, Svatoň M, Froňková E, Fejtková M, Zachová R, Rataj M, Zentsová I, Milota T, Klocperk A, Kalina T, Šedivá A. [Utility of Ruxolitinib in a Child with Chronic Mucocutaneous Candidiasis Caused by a Novel STAT1 Gain-of-Function Mutation.](#) J Clin Immunol. 2018 Jun 22

Klocperk A, Mejstříková E, Kayserová J, Kalina T, Šedivá A. [Low marginal zone-like B lymphocytes and natural antibodies characterize skewed B-lymphocyte subpopulations in del22q11 DiGeorge patients.](#) Clin Immunol. 2015 Dec;161(2):144-9

Hungary

Maródi L. [Inborn errors of T cell immunity underlying autoimmune diseases.](#) Expert Rev Clin Immunol. 2017 Feb;13(2):97-99.

Soltész B, Tóth B, Sarkadi AK, Erdős M, Maródi L. [The Evolving View of IL-17-Mediated Immunity in Defense Against Mucocutaneous Candidiasis in Humans.](#) Int Rev Immunol. 2015;34(4):348-63.

Tóth B, Soltész B, Gyimesi E, Csorba G, Veres Á, Lányi Á, Kovács G, Maródi L, Erdős M. [Severe XLP Phenotype Caused by a Novel Intronic Mutation in the SH2D1A Gene.](#) J Clin Immunol. 2015 Jan;35(1):26-31.

Soltész B, Tóth B, Shabashova N, Bondarenko A, Okada S, Cypowij S, Abhyankar A, Csorba G, Taskó S, Sarkadi AK, Méhes L, Rozsival P, Neumann D, Chernyshova L, Tulassay Z, Puel A, Casanova JL, Sediva A, Litzman J, Maródi L. [New and recurrent gain-of-function STAT1 mutations in patients with chronic mucocutaneous candidiasis from Eastern and Central Europe.](#) J Med Genet. 2013 Sep;50(9):567-78.

L Marodi and the J Project Study Group. **Fifteen years of the J Project.** J Clin Immunol, 2019, in press

Lithuania

Sjakste T, Paramonova N, Wu LS, Zemeckiene Z, Sitkauskiene B, Sakalauskas R, Wang JY, Sjakste N. **PSMA6 (rs2277460, rs1048990), PSMC6 (rs2295826, rs2295827) and PSMA3 (rs2348071) genetic diversity in Latvians, Lithuanians and Taiwanese.** *Meta Gene* 2014; 2: 283-98.

Zemeckienė Z, Vitkauskienė A, Sjakste T, Sitkauskienė B, Sakalauskas R. **Proteasomes and proteasomal gene polymorphism in association with inflammation and various diseases.** *Medicina* 2013; 49(5): 207-13.

Baş M, Greve J, Hoffmann TK, Reshef A, Aberer W, Maurer M, Kivity S, Farkas H, Floccard B, Arcoletto F, Martin L, Sitkauskiene B, Bouillet L, Schmid-Grendelmeier P, Li H, Zanichelli

A. Repeat treatment with icatibant for multiple hereditary angioedema attacks: FAST-2 open-label study. Allergy 2013; 68(11): 1452-9

Romania

Gulacsy V, Soltesz B, Petrescu C, Bataneant M, Gyimesi E, Serban M, Marodi L, Toth B. **A novel large deletion and single nucleotide insertion in the Wiskott-Aldrich syndrome protein gene.** Eur J Haematol, 2015 Jul;95(1):93-8

A.Heinze, M.C.Elze, S.Kloess, O.Ciocarlie, C.Konigs, S.Betz, M.Bremm, R.Esser, T.Klingebl, M.Serban, J.L.Hutton, U.Koehl. **Age-matched dendritic cell subpopulations reference values in childhood.** Scandinavian Journal of Immunology, 2013, 77, 213-2209

Slovakia

Geier CB, Piller A, Eibl MM, Ciznar P, Ilencikova D, Wolf HM: **Terminal 14Q32.33 deletion as a novel cause of agammaglobulinemia.** Clin Immunol. 2017 Oct;183:41-45.

B, Tulinska J, Palkovicova Murinova L, Buocikova V, Liskova A, Rausova K, Kuricova Smolkova M, Patayova H, Sustrova M, Neubauerova Svorcova E, Ilavska S, Szabova M, Nemessanyi T, Jahnova E, Dusinska M, Ciznar P, Fuortes L. **Impact of interleukin 13 (IL13) genetic polymorphism Arg130Gln on total serum immunoglobulin (IgE) levels and interferon (IFN)- γ gene expression.** Clin Exp Immunol. 2017 Apr;188(1):45-52.

Lobotková D, Dická E, Rolný V, Stankovič I, Čižnár P. **Systemic Listeria monocytogenes infection in a 2-year-old immunocompetent child.** Infection. 2014 Dec; 42(6); 1055-9.

Common variable immune deficiency, central diabetes insipidus and anemia. Accepted for publication in Central European Journal of Immunology, 2018.

Wu K, Purswani P, Ujhazi B, Mihailova S, Naumova E, Stefanov S, Savic S, Sargur R, Milojevic D, Jolan W. **Clinical Challenges in Recombination Activating Gene Deficiency and Arthritis.** Frontiers, in press

Slovenia

Blazina Š, Debeljak M, Košnik M, Simčič S, Stopinšek S, Markelj G, Toplak N, Kopač P, Zakotnik B, Pokorn M, Avčin T. **Functional Complement Analysis Can Predict Genetic Testing Results and Long-Term Outcome in Patients With Complement Deficiencies.** Front Immunol. 2018 Mar 21;9:500.

Blazina Š, Markelj G, Jeverica AK, Toplak N, Bratanič N, Jazbec J, Kopač P, Debeljak M, Ihan A, Avčin T.

Autoimmune and Inflammatory Manifestations in 247 Patients with Primary Immunodeficiency - a Report from the Slovenian National Registry. J Clin Immunol. 2016; 36: 764-773.

Holcar M, Goropevšek A, Ihan A, Avčin T.

Age-Related Differences in Percentages of Regulatory and Effector T Lymphocytes and Their Subsets in Healthy Individuals and Characteristic STAT1/STAT5 Signalling Response in Helper T Lymphocytes. J Immunol Res. 2015:352934.

Debeljak M, Toplak N, Abazi N, Szabados B, Mulaosmanović V, Radović J, Perko D, Vojnović J, Constantin T, Kuzmanovska D, Avčin T.

The carrier rate and spectrum of MEFV gene mutations in central and southeastern European populations. Clin Exp Rheumatol. 2015; 33 (6 Suppl 94): S19-23.

Perko D, Debeljak M, Toplak N, Avčin T.

Clinical features and genetic background of the periodic Fever syndrome with aphthous stomatitis, pharyngitis, and adenitis: a single center longitudinal study of 81 patients. Mediators Inflamm. 2015: 293417.