

Published

(PIGIT 2017-2022)

1. Sacco K, Castagnoli R, Vakkilainen S, Liu C, Delmonte OM, Oguz C, Kaplan IM, Alehashemi S, Burbelo PD, Bhuyan F, de Jesus AA, Dobbs K, Rosen LB, Cheng A, Shaw E, Vakkilainen MS, Pala F, Lack J, Zhang Y, Fink DL, Oikonomou V, Snow AL, Dalgard CL, Chen J, Sellers BA, Montealegre Sanchez GA, Barron K, Rey-Jurado E, Vial C, **Poli MC**, Licari A, Montagna D, Marseglia GL, Licciardi F, Ramenghi U, Discepolo V, Lo Vecchio A, Guarino A, Eisenstein EM, Imberti L, Sottini A, Biondi A, Mató S, Gerstbacher D, Truong M, Stack MA, Magliocco M, Bosticardo M, Kawai T, Danielson JJ, Hulett T, Askenazi M, Hu S; NIAID Immune Response to COVID Group; Chile MIS-C Group; Pavia Pediatric COVID-19 Group, Cohen JI, Su HC, Kuhns DB, Lionakis MS, Snyder TM, Holland SM, Goldbach-Mansky R, Tsang JS, Notarangelo LD. Immunopathological signatures in multisystem inflammatory syndrome in children and pediatric COVID-19. *Nat Med.* 2022 May;28(5):1050-1062. doi: 10.1038/s41591-022-01724-3. Epub 2022 Feb 17. PMID: 35177862; PMCID: PMC9119950.
2. Cárdenas GV, Iturriaga C, Hernández CD, Tejos-Bravo M, Pérez-Mateluna G, Cabalin C, Urzúa M, Venegas-Salas LF, Fraga JP, Rebolledo B, **Poli MC**,

Repetto GM, Casanello P, Castro-Rodríguez JA, Borzutzky A. Prevalence of filaggrin loss-of-function variants in Chilean population with and without atopic dermatitis. *Int J Dermatol*. 2022 Mar;61(3):310-315. doi: 10.1111/ijd.15887. Epub 2021 Sep 4. PMID: 34480753.

3. Cetin Gedik K, Lamot L, Romano M, Demirkaya E, Piskin D, Torreggiani S, Adang LA, Armangue T, Barchus K, Cordova DR, Crow YJ, Dale RC, Durrant KL, Eleftheriou D, Fazzi EM, Gattorno M, Gavazzi F, Hanson EP, Lee-Kirsch MA, Montealegre Sanchez GA, Neven B, Orcesi S, Ozen S, **Poli MC**, Schumacher E, Tonduti D, Uss K, Aletaha D, Feldman BM, Vanderver A, Brogan PA, Goldbach-Mansky R. The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology Points to Consider for Diagnosis and Management of Autoinflammatory Type I Interferonopathies: CANDLE/PRAAS, SAVI, and AGS. *Arthritis Rheumatol*. 2022 May;74(5):735-751. doi: 10.1002/art.42087. Epub 2022 Mar 21. PMID: 35315249.
4. Burbelo PD, Castagnoli R, Shimizu C, Delmonte OM, Dobbs K, Discepolo V, Lo Vecchio A, Guarino A, Licciardi F, Ramenghi U, Rey-Jurado E, Vial C, Marseglia GL, Licari A, Montagna D, Rossi C, Montealegre Sanchez GA, Barron K, Warner BM, Chiorini JA, Espinosa Y, Noguera L, Dropulic L, Truong M, Gerstbacher D, Mató S, Kanegaye J, Tremoulet AH; Pediatric Emergency Medicine Kawasaki Group, Eisenstein EM, Su HC, Imberti L, **Poli MC**, Burns JC, Notarangelo LD, Cohen JL. Autoantibodies Against Proteins Previously Associated With Autoimmunity in Adult and Pediatric Patients With COVID-

19 and Children With MIS-C. *Front Immunol.* 2022 Mar 11;13:841126. doi: 10.3389/fimmu.2022.841126. PMID: 35360001; PMCID: PMC8962198.

5. Forbes LR, Eckstein OS, Gulati N, Peckham-Gregory EC, Ozuah NW, Lubega J, El-Mallawany NK, Agrusa JE, **Poli MC**, Vogel TP, Chaimowitz NS, Rider NL, Mace EM, Orange JS, Caldwell JW, Aldave-Becerra JC, Jolles S, Saettini F, Chong HJ, Stray-Pedersen A, Heslop HE, Kamdar KY, Rouce RH, Muzny DM, Jhangiani SN, Gibbs RA, Coban-Akdemir ZH, Lupski JR, McClain KL, Allen CE, Chinn IK. Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. *J Allergy Clin Immunol.* 2022 Feb;149(2):758-766. doi: 10.1016/j.jaci.2021.07.015. Epub 2021 Jul 28. PMID: 34329649; PMCID: PMC8795244.
6. Davidson S, Yu CH, Steiner A, Ebstein F, Baker PJ, Jarur-Chamy V, Hrovat Schaale K, Laohamonthonkul P, Kong K, Calleja DJ, Harapas CR, Balka KR, Mitchell J, Jackson JT, Geoghegan ND, Moghaddas F, Rogers KL, Mayer-Barber KD, De Jesus AA, De Nardo D, Kile BT, Sadler AJ, **Poli MC**, Krüger E, Goldbach Mansky R, Masters SL. Protein kinase R is an innate immune sensor of proteotoxic stress via accumulation of cytoplasmic IL-24. *Sci Immunol.* 2022 Feb 11;7(68):eabi6763. doi: 10.1126/sciimmunol.abi6763. Epub 2022 Feb 11. PMID: 35148201.
7. Cetin Gedik K, Lamot L, Romano M, Demirkaya E, Piskin D, Torreggiani S, Adang LA, Armangue T, Barchus K, Cordova DR, Crow YJ, Dale RC, Durrant KL, Eleftheriou D, Fazzi EM, Gattorno M, Gavazzi F, Hanson EP, Lee-Kirsch MA, Montealegre Sanchez GA, Neven B, Orcesi S, Ozen S, **Poli MC**, Schumacher E, Tonduti D, Uss K, Aletaha D, Feldman BM, Vanderver A,

Brogan PA, Goldbach-Mansky R. The 2021 European Alliance of Associations for Rheumatology/American College of Rheumatology points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS. *Ann Rheum Dis*. 2022 May;81(5):601-613. doi: 10.1136/annrheumdis-2021-221814. Epub 2022 Jan 27. PMID: 35086813; PMCID: PMC9036471.

8. Carmona-Rivera C, Zhang Y, Dobbs K, Markowitz TE, Dalgard CL, Oler AJ, Claybaugh DR, Draper D, Truong M, Delmonte OM, Licciardi F, Ramenghi U, Crescenzo N, Imberti L, Sottini A, Quaresima V, Fiorini C, Discepolo V, Lo Vecchio A, Guarino A, Pierri L, Catzola A, Biondi A, Bonfanti P, **Poli Harlowe MC**, Espinosa Y, Astudillo C, **Rey-Jurado E**, Vial C, **de la Cruz J**, Gonzalez R, Pinera C, Mays JW, Ng A, Platt A; NIH COVID Autopsy Consortium; COVID STORM Clinicians, Drolet B, Moon J, Cowen EW, Kenney H, Weber SE, Castagnoli R, Magliocco M, Stack MA, Montealegre G, Barron K, Hewitt SM, Arkin LM, Chertow DS, Su HC, Notarangelo LD, Kaplan MJ. Multicenter analysis of neutrophil extracellular trap dysregulation in adult and pediatric COVID-19. *medRxiv [Preprint]*. 2022 Mar 3:2022.02.24.22271475. doi: 10.1101/2022.02.24.22271475. Update in: *JCI Insight*. 2022 Jul 19;; PMID: 35262093; PMCID: PMC8902885.
9. Martinez CA, Ebstein F, Nicholas SK, De Guzman M, Forbes LR, Delmonte OM, Bosticardo M, Castagnoli R, Krance R, Notarangelo LD, Krüger E, Orange JS, **Poli MC**. HSCT corrects primary immunodeficiency and immune dysregulation in patients with POMP-related auto-inflammatory disease. *Blood*. 2021 May 21: blood.2021011005. doi: 10.1182/blood.2021011005.

10. Vogel TP, Top KA, Karatzios C, Hilmers DC, Tapia LI, Moceri P, Giovannini-Chami L, Wood N, Chandler RE, Klein NP, Schlaudecker EP, **Poli MC**, Muscal E, Munoz FM. Multisystem inflammatory syndrome in children and adults (MIS-C/A): Case definition & guidelines for data collection, analysis, and presentation of immunization safety data. *Vaccine*. 2021 May 21;39(22):3037-3049. doi: 10.1016/j.vaccine.2021.01.054.
11. Niño-Taravilla C, Espinosa-Vielma YP, Otaola-Arca H, **Poli-Harlowe C**, Tapia LI, Ortiz-Fritz P. Pediatric Inflammatory Multisystem Syndrome Temporally Associated with SARS-CoV-2 Treated with Tocilizumab. *Pediatr Rep*. 2020 Dec 4;12(3):142-148. doi: 10.3390/pediatric12030029
12. Verdugo P, Alvarez P, **Poli C**. Et al. Parámetros Hematológicos y Biomarcadores predictores de gravedad en Síndrome Inflamatorio Pediátrico Multisistémico asociado a SARS-CoV-2. *Accepted Revista Médica de Chile 2020*.
13. Cofré F. **Poli MC** et al. Espectro clínico de la infección en niños por SARS-co-V-2 en un Centro de Referencia pediátrico en plena Pandemia. Reporte del Comité Clínico COVID, Hospital de Niños Roberto del Rio, Santiago, Chile. *Aceptado Revista Chilena de Infectología Aug. 2020*
14. Contreras JL, Ladino MA, Aránguiz K, Mendez GP, Coban-Akdemir Z, Yuan B, Gibbs RA, Burrage LC, Lupski JR, Chinn IK, Vogel TP, Orange JS, **Poli MC**. Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the

Literature. Front Pediatr. 2021 May 20;9:673957. doi: 10.3389/fped.2021.673957.

15. Omar Valenzuela, Sebastián Ibáñez, **M. Cecilia Poli**, Patricia Roessler, Mabel Aylwin, Gigia Roizen, Mirentxu Iruretagoyena, Vivianne Agar, Javiera Donoso, Margarita Fierro, José Montes. First Report of Tocilizumab Use in a Cohort of Latin American Patients Hospitalized for Severe COVID-19 Pneumonia. Front. Med. 7:596916. doi: 10.3389/fmed.2020.596916
16. Saettini F, **Poli C**, Vengoechea J, Bonanomi S, Orellana JC, Fazio G, Rodriguez FH, Noguera LP, Booth CA, Jarur-Chamy V, Shams M, Iascone M, Vukic M, Gasperini S, Quadri M, Barroeta Seijas AB, Rivers E, Mauri M, Badolato R, Cazzaniga G, Bugarin C, Gaipa G, Kroes W, Moratto D, van Oostaijen-Ten Dam MM, Baas F, van der Maarel S, Piazza R, Coban-Akdemir Z, Lupski JR, Yuan B, Chinn IK, Daxinger L, Biondi A. Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in Folliculin Interacting Protein 1 deficiency. Blood. 2020 Sep 9: blood.2020006441. doi: 10.1182/blood.2020006441. Epub ahead of print. PMID: 32905580.
17. Cook, Sarah A., Comrie, William A., **Poli, M. Cecilia**; Similuk, Morgan; Oler, Andrew J.; Faruqi, Aiman J.; Kuhns, Douglas B.; Yang, Sheng; Vargas-Hernandez, Alexander; Carisey, Alexandre F.; Fournier, Benjamin; Anderson, D. Eric; Price, Susan; Smelkinson, Margery; Abou Chahla, Wadih; et. al. HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. SCIENCE (0036-8075) Year: 2020 Volume: 369 Issue: 6500 Start Page: 202.

18. High percentage of smudge cells in a patient with Covid-19: rediscovering their utility. Joaquin Jerez and **Daniel M. Ernst**. eJHaem;2020:1-2.
19. Hypophosphatemia induced by carboxymaltose iron and imatinib. Report of two cases. Danisa Ivanovic-Zuvic, Alexandra Fischman, Macarena Jimenez, Alejandra Martínez, **Daniel Ernst**, Luis Toro, Francisco J Guarda, Pablo Florenzano. Rev Med Chile 2020;148:404-408.
20. **Rey-Jurado E**, Bohmwald K, Gálvez NMS, Becerra D, Porcelli SA, Carreño LJ, Kalergis AM. Contribution of NKT cells to the immune response and pathogenesis triggered by respiratory viruses. Virulence. 2020 Dec;11(1):580-593. doi: 10.1080/21505594.2020.1770492. PMID: 32463330; PMCID: PMC7549913.
21. **Rey-Jurado E**, Bohmwald K, Correa HG, Kalergis AM. TCR Repertoire Characterization for T Cells Expanded in Response to hRSV Infection in Mice Immunized with a Recombinant BCG Vaccine. Viruses. 2020 Feb 20;12(2):233. doi: 10.3390/v12020233. PMID: 32093256; PMCID: PMC7077260.
22. Abarca K, **Rey-Jurado E**, Muñoz-Durango N, Vázquez Y, Soto JA, Gálvez NMS, Valdés-Ferrada J, Iturriaga C, Urzúa M, Borzutzky A, Cerda J, Villarroel L, Madrid V, González PA, González-Aramundiz JV, Bueno SM, Kalergis AM. Safety and immunogenicity evaluation of recombinant BCG vaccine against respiratory syncytial virus in a randomized, double-blind, placebo-controlled phase I clinical trial. EClinicalMedicine. 2020 Oct 6;27:100517. doi: 10.1016/j.eclinm.2020.100517. PMID: 33073219; PMCID: PMC7548429.

23. Cytarabine and hematopoietic cell transplantation for mantle cell lymphoma. Analysis of 20 patients. James Campbell, Sebastian Hurtado, Consuelo Kutz, Katherine Soto, **Daniel Ernst**. Rev Med Chile 2019; 147:9-17. Corresponding author.
24. Boone, ATS; Chinn, IK; Alaez-Verson, C; Yamazaki-Nakashimada, MA; Carrillo-Sanchez, K; Garcia-Cruz, MDH; **Poli, MC**; Serrano, MEG; Torres, EAM; Zermeno, DM; Forbes, LR; Espinosa-Rosales, FJ; Espinosa-Padilla, SE; Orange, JS; Reyes. Failing to Make Ends Meet: The Broad Clinical Spectrum of DNA Ligase IV Deficiency. Case Series and Review of the Literature Frontiers in Pediatrics (2296-2360), Jan 2019 Volume: 6
25. Alejandra Aird, Macarena Lagos, Alexander Vargas-Hernández, Jennifer E Posey, Zeynep Coban-Akdemir, Shalini Jhangiani, Emily M Mace, Anaïd Reyes, Alejandra King, Felipe Cavagnaro, Lisa R Forbes, Ivan K Chinn, James R Lupski, Jordan S Orange, **María Cecilia Poli**. Novel heterozygous mutation in NFKB2 1 is associated with early onset CVID and a functional defect in NK cells complicated by disseminated CMV infection and severe nephrotic syndrome Frontiers in Pediatrics. 2019; 7: 303. Published online 2019 Jul 30. doi: 10.3389/fped.2019.00303
26. Bustamante Ogando, Juan Carlos; Partida Gaytan, Armando; Aldave Becerra, Juan Carlos; Alvarez Cardona, Aristoteles; Bezrodnik, Liliana; Borzutzky, Arturo; **Poli, Cecilia**, Galicia, Lizbeth Blancas; Cabanillas, Diana; Condino-Neto, Antonio; Ranero, Agustin De Colsa; Padilla, Sara Espinosa; Fernandes, Juliana Folloni; Garcia Campos, Jorge Alberto; Tello, Hector Gomez; Gonzalez Serrano, Maria Edith; et. al. Latin American consensus on

- the supportive management of patients with severe combined immunodeficiency JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY (0091-6749) Year: 2019 Volume: 144 Issue: 4 Start Page: 897 End Page: 905
27. Hoyos-Bachilloglu, Rodrigo; Sotomayor F, Cristian; **Poli H, Cecilia**. Severe combined immunodeficiency: the time for newborn screening has come. Revista Chilena de Pediatría (3704106) Year: 2019 Volume: 90 Issue: 6: 581-588
28. Goudouris E.S.; Segundo G.R.S.; **Poli C**. Repercussions of inborn errors of immunity on growth ISI SCOPUS JORNAL DE PEDIATRIA (0021-7557) Year: 2019 Volume: 95 Start Page: S49 End Page: S58
29. Ebstein F, **Poli Harlowe MC**, Studencka-Tursi M, Krüger E. Contribution of the Unfolded Protein Response (UPR) to the pathogenesis of Proteasome – Associates Autoinflammatory Syndromes (PRAAS). Front Immunol. 2019;10:2756. Published 2019 Nov 26. doi:10.3389/fimmu.2019.02756
30. **Poli MC**. New Autoinflammatory diseases. [Curr Opin Pediatr](#). 2018 Dec;30(6):837-847. doi: 10.1097/MOP.0000000000000696.
31. Dorjbal B, Stinson JR, Ma CA, Weinreich MA, Miraghazadeh B, Hartberger JM, Frey-Jakobs S, Weidinger S, Moebus L, Franke A, Schäffer AA, Bulashevskaya A, Fuchs S, Ehl S, Limaly S, Arkwright PD, Briggs TA, Langley C, Bethune C, Whyte AF, Alachkar H, Nejentsev S, DiMaggio T, Nelson CG, Stone KD, Nason M, Brittain EH, Oler AJ, Veltri DP, Leahy TR, Conlon N, **Poli MC**, Borzutzky A, Cohen JI, Davis J, Lambert MP, Romberg N, Sullivan KE, Paris K, Freeman AF, Lucas L, Chandrakasan S, Savic S, Hambleton S, Patel SY, Jordan MB, Theos A, Lebensburger J, Atkinson TP, Torgerson TR,

- Chinn IK, Milner JD, Grimbacher B, Cook MC, Snow AL. [Hypomorphic caspase activation and recruitment domain 11 \(CARD11\) mutations associated with diverse immunologic phenotypes with or without atopic disease.](#) J Allergy Clin Immunol. 2018 Aug 28. pii: S0091-6749(18)31201-6. doi: 10.1016/j.jaci.2018.08.013. [Epub ahead of print]. PMID: 30170123
32. Coban-Akdemir Z, White JJ, Song X, Jhangiani SN, Fatih JM, Gambin T, Bayram Y, Chinn IK, Karaca E, Punetha J, **Poli C**; Baylor-Hopkins Center for Mendelian Genomics, Boerwinkle E, Shaw CA, Orange JS, Gibbs RA, Lappalainen T, Lupski JR, Carvalho CMB. [Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles.](#) Am. J Hum Genet. 2018 Aug 2;103(2):171-187. doi: 10.1016/j.ajhg.2018.06.009. Epub 2018 Jul 19.
33. **Poli MC**, Ebstein F, Nicholas SK, de Guzman MM, Forbes LR, Chinn IK, Mace EM, Vogel TP, Carisey AF, Benavides F, Coban-Akdemir ZH, Gibbs RA, Jhangiani SN, Muzny DM, Carvalho CMB, Schady DA, Jain M, Rosenfeld JA, Emrick L, Lewis RA, Lee B; Undiagnosed Diseases Network members, Zieba BA, Küry S, Krüger E, Lupski JR, Bostwick BL, Orange JS. Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. Am. J Hum Genet. 2018 May 22. pii: S0002-9297(18)30142-3. doi: 10.1016/j.ajhg.2018.04.010.
34. Humanized anti-CD123 antibody facilitates NK cell antibody-dependent cell-mediated cytotoxicity (ADCC) of Hodgkin lymphoma targets via ARF-6/PLD-1. **Daniel Ernst**, Brent A. Williams, Xing-Hua Wang, Nara Yon, Kyung-Phil

Kim, Jodi Chiu, Zhi Juan Luo, Karin G. Hermans, Joerg Krueger, Armand Keating. Blood Cancer Journal. Accepted for publication Dec. 13th 2018. DOI: 10.1038/s41408-018-0168-2. First & Corresponding author.

35. Interpreting an atypical clinical case: the timing of events determines the diagnosis and impacts the prognosis of non-Hodgkin lymphomas. **Daniel Ernst** and Karen Escobar. Rev Med Chile 2018; 146: 266-270. First & Corresponding author.

36. Acquired Natural Killer Cell Dysfunction in the Tumor Microenvironment of Classic Hodgkin Lymphoma. Jodi Chiu, **Daniel Ernst**, Armand Keating. Front. Immunol. 9:267. doi: 10.3389/fimmu.2018.00267. Co-first & Corresponding author.

37. Chinn IK, Eckstein OS, Peckham-Gregory EC, Goldberg BR, Forbes LR, Nicholas SK, Mace EM, Vogel TP, Abhyankar HA, Diaz MI, Heslop HE, Krance RA, Martinez CA, Nguyen TC, Bashir DA, Goldman JR, Stray-Pedersen A, Pedroza LA, **Poli MC**, Aldave Becerra JC, McGhee SA, Al-Herz W, Chamdin A, Coban-Akdemir ZH, Jhangiani SN, Muzny DM, Cao TN, Hong DN, Gibbs RA, Lupski JR, Orange JS, McClain KL, Allen CE. Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood. 2018 Apr 9. pii: blood-2017-11-814244. doi: 10.1182/blood-2017-11-814244.

38. Resistance of leukemia cells to cytarabine chemotherapy is mediated by bone marrow stroma, involves cell-surface equilibrative nucleoside transporter-1 removal and correlates with patient outcome. Patricia Macanas-Pirard, Richard Broekhuizen, Alfonso González, Claudia Oyanadel, **Daniel Ernst**, Patricia García, Viviana P. Montecinos, Felipe Court, Mauricio Ocqueteau, Pablo Ramirez, Bruno Nervi. *Oncotarget*. 2017 Feb 1. doi: 10.18632/oncotarget.14981
39. Response to ABVD chemotherapeutic protocol in patients with early-stage Hodgkin`s lymphoma. Javier Díaz, Katherine Soto and **Daniel Ernst**. *Rev Med Chile* 2017; 145: 619-622. Senior & Corresponding author.
40. **Poli C**. Hoyos- Bachiloglu R. Borzutzky A. Primary immunodeficiency epidemiology in Chile evaluated through ICD-10 coded hospital admissions. *Alergologia e Inmunopathologia* 2017;45 (1):33-39.