The 2021 EULAR and ACR points to consider for diagnosis and management of autoinflammatory type I interferonopathies: CANDLE/PRAAS, SAVI and AGS

Kader Cetin Gedik,¹ Lovro Lamot,² Micol Romano,³ Erkan Demirkaya,³ David Piskin,^{4,5} Sofia Torreggiani,^{6,7} Laura A Adang,⁸ Thais Armangue,⁹ Kathe Barchus,¹⁰ Devon R Cordova,¹¹ Yanick J Crow,^{12,13} Russell C Dale,¹⁴ Karen L Durrant,^{10,15} Despina Eleftheriou,¹⁶ Elisa M Fazzi,¹⁷ Marco Gattorno ,¹⁸ Francesco Gavazzi,^{8,19} Eric P Hanson,²⁰ Min Ae Lee-Kirsch,²¹ Gina A Montealegre Sanchez,²² Bénédicte Neven,²³ Simona Orcesi,^{24,25} Seza Ozen ,²⁶ M Cecilia Poli,²⁷ Elliot Schumacher,¹⁰ Davide Tonduti,²⁸ Katsiaryna Uss,¹ Daniel Aletaha ,²⁹ Brian M Feldman ,^{30,31} Adeline Vanderver,^{8,32} Paul A Brogan ,¹⁶ Raphaëla Goldbach-Mansky

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For numbered affiliations see end of article.

Correspondence to

Dr Raphaëla Goldbach-Mansky, Translational Autoinflammatory Diseases Section, Laboratory of Clinical Immunology and Microbiology, National Institute of Health, Bethesda, Maryland, USA; goldbacr@mail.nih.gov

CKG, LL and MR contributed equally.

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ABSTRACT

Objective Autoinflammatory type I interferonopathies, chronic atypical neutrophilic dermatosis with lipodystrophy and elevated temperature/proteasome-associated autoinflammatory syndrome (CANDLE/PRAAS), stimulator of interferon genes (STING)-associated vasculopathy with onset in infancy (SAVI) and Aicardi-Goutières syndrome (AGS) are rare and clinically complex immunodysregulatory diseases. With emerging knowledge of genetic causes and targeted treatments, a Task Force was charged with the development of 'points to consider' to improve diagnosis, treatment and long-term monitoring of patients with these rare diseases.

Methods Members of a Task Force consisting of rheumatologists, neurologists, an immunologist, geneticists, patient advocates and an allied healthcare professional formulated research questions for a systematic literature review. Then, based on literature, Delphi questionnaires and consensus methodology, 'points to consider' to guide patient management were developed.

Results The Task Force devised consensus and evidence-based guidance of 4 overarching principles and 17 points to consider regarding the diagnosis, treatment and long-term monitoring of patients with the autoinflammatory interferonopathies, CANDLE/PRAAS, SAVI and AGS.

Conclusion These points to consider represent state-of-the-art knowledge to guide diagnostic evaluation, treatment and management of patients with CANDLE/PRAAS, SAVI and AGS and aim to standardise and improve care, quality of life and disease outcomes.

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Author affiliations

¹Translational Autoinflammatory Diseases Section, Laboratory of Clinical Immunology and Microbiology, National Institute of Health and Infectious Diseases, National Institutes of Health, Bethesda, Maryland, USA

²Department of Pediatrics, University Hospital Centre Zagreb, University of Zagreb School of Medicine, Zagreb, Croatia

³Division of Paediatric Rheumatology, Department of Paediatrics, Schulich School of Medicine and Dentistry, University of Western Ontario, London, Ontario, Canada

⁴Department of Epidemiology and Biostatistics, Schulich School of Medicine & Dentistry, University of Western Ontario, London, Ontario, Canada

⁵London Health Sciences Center, Lawson Health Research Institute, London, Ontario, Canada

⁶Translational Autoinflammatory Diseases Section, Laboratory of Clinical Immunology and Microbiology, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Maryland, USA

⁷UOC Pediatria a Media Intensità di Cura, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milano, Lombardia, Italy

⁸Division of Neurology, The Children's Hospital of Philadelphia, Philadelphia, Pennsylvania, USA

⁹Pediatric Neuroimmunology Unit, Neurology Service, Sant Joan de Deu Children's Hospital, and IDIBAPS-Hospital Clinic, University of Barcelona, Barcelona, Spain

¹⁰Autoinflammatory Alliance, San Francisco, California, USA

¹¹Aicardi-Goutières Syndrome Americas Association, Manhattan Beach, California, USA

¹²Centre for Genomic and Experimental Medicine, MRC Institute of Genetics and Molecular Medicine, University of Edinburgh, Edinburgh, UK

¹³Laboratory of Neurogenetics and Neuroinflammation, Institut Imagine, Université de Paris, Paris, Île-de-France, France

¹⁴Kids Neuroscience Centre, Faculty of Medicine and Health, The University of Sydney, Sydney, New South Wales, Australia

¹⁵Kaiser San Francisco Hospital, San Francisco, California, USA

¹⁶Great Ormond Street Institute of Child Health, University College London, London, UK

¹⁷Child Neurology and Psychiatry Unit, Department of Clinical and Experimental Sciences ASST Civil Hospital, University of Brescia, Brescia, Italy

¹⁸Center for Autoinflammatory diseases and Immunodeficiencies, IRCCS Istituto Giannina Gaslini, Genoa, Italy

¹⁹Department of Molecular and Translational Medicine, University of Brescia, Brescia, Italy

²⁰Riley Hospital for Children, Indiana University School of Medicine, Indianapolis, Indiana, USA

²¹Department of Pediatrics, Medical Faculty Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany

²²Intramural Clinical Management and Operations Branch (ICMOB), Division of Clinical Research, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Maryland, USA

²³Necker Children's Hospital, Assistance Publique-Hôpitaux de Paris, Université de Paris, Institut Imagine Institut des Maladies Génétiques, Paris, Île-de-France, France

Recommendation

- ²⁴Child Neurology and Psychiatry Unit, IRCCS Mondino Foundation, Pavia, Italy
²⁵Department of Brain and Behavioral Sciences, University of Pavia, Pavia, Lombardia, Italy
²⁶Pediatric Rheumatology, Hacettepe University, Ankara, Turkey
²⁷Department of Pediatrics, Facultad de Medicina Clínica Alemana Universidad del Desarrollo, Santiago, Chile
²⁸Child Neurology Unit, COALA (Center for Diagnosis and Treatment of Leukodystrophies), V. Buzzi Children's Hospital, Milano, Italy
²⁹Department of Rheumatology, Medical University of Vienna, Vienna, Austria
³⁰Division of Rheumatology, Hospital for Sick Children, Toronto, Ontario, Canada
³¹30Department of Pediatrics, Faculty of Medicine, University of Toronto Institute of Health Policy Management and Evaluation, Toronto, Ontario, Canada
³²Department of Neurology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, Pennsylvania, USA

Twitter Seza Ozen @drsezaozen

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Contributors All authors contributed to the formulation of the points to consider. In details, the steering committee of the Task Force (RG-M, PB, AV, BF, ED) defined the research questions for the SLR. A systematic literature review was conducted by KCG, MR, LL with support from a librarian and epidemiologist (DH and DP) under supervision of a senior methodologist (ED). KCG, LL, MR extracted the data. RGM, PB and AV synthesised the results form SLR and Delphi questionnaires and

generated draft statements. The manuscript was drafted by KCG, LL and MR and revised by RG-M, PB, AV, ED and BF. DA oversaw the proceedings and provided advice of this points to consider project as EULAR methodologist. All other authors participated in the Task Force meetings, in two pre-meeting Delphi questionnaires, suggested and agreed upon the research questions, read the final statements prior to the manuscript, discussed results and made contributions to the text. All authors approved the final version of the manuscript.

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ORCID iDs

Marco Gattorno <http://orcid.org/0000-0003-0704-1916>
 Seza Ozen <http://orcid.org/0000-0003-2883-7868>
 Daniel Aletaha <http://orcid.org/0000-0003-2108-0030>
 Brian M Feldman <http://orcid.org/0000-0002-7813-9665>
 Paul A Brogan <http://orcid.org/0000-0001-6178-6893>
 Raphaela Goldbach-Mansky <http://orcid.org/0000-0001-7865-5769>