

Annex I: List of consortium/ groups and corresponding members and affiliations

Solve-RD SNV-indel working group

Enzo Cohen¹, Isabel Cuesta², Daniel Danis³, Anne-Sophie Denommé-Pichon^{4,5,6}, Yannis Duffourd^{4,6}, Christian Gilissen^{7,8}, Mridul Johari⁹, Steven Laurie¹⁰, Shuang Li¹¹, Leslie Matalonga¹⁰, Isabelle Nelson¹, Sophia Peters¹², Ida Paramonov¹⁰, Sivakumar Prasanth¹³, Peter Robinson³, Karolis Sablauskas^{7,8}, Marco Savarese⁹, Wouter Steyaert^{7,8}, Ana Töpf¹⁴, Joeri K. van der Velde¹¹, and Antonio Vitobello⁴

Solve-RD-DITF-ITHACA

Siddharth Banka^{15,16}, Elisa Benetti¹⁷, Giorgio Casari^{18,19}, Andrea Cioffi²⁰, Jill Clayton-Smith^{15,16}, Bruno Dallapiccola²⁰, Elke de Boer^{7,21}, Anne-Sophie Denommé-Pichon^{4,5,6}, Kornelia Ellwanger^{22,23}, Laurence Faivre^{4,24}, Christian Gilissen^{7,8}, Holm Graessner^{22,23}, Tobias B. Haack²², Anna Hammarsjö²⁵, Marketa Havlovicova²⁶, Alexander Hoischen^{7,8,27}, Anne Hugon²⁸, Adam Jackson¹⁶, Tjitske Kleefstra^{7,21}, Anna Lindstrand²⁵, Estrella López-Martín²⁹, Milan Macek Jr.²⁶, Leslie Matalonga¹⁰, Manuela Morleo¹⁹, Vincenzo Nigro¹⁹, Ann Nordgren²⁵, Maria Pettersson²⁵, Michele Pinelli¹⁹, Simone Pizzi²⁰, Manuel Posada²⁹, Francesca Clementina Radio³⁰, Alessandra Renieri^{17,31,32}, Caroline Rooryck³³, Lukas Ryba²⁶, Martin Schwarz²⁶, Marco Tartaglia²⁰, Christel Thauvin^{4,24}, Annalaura Torella^{18,19}, Aurélien Trimouille³⁴, Alain Verloes^{28,35}, Lisenka Vissers^{7,21}, Antonio Vitobello⁴, Pavel Votypka²⁶, Klea Vyshka^{28,35} and Birte Zurek^{22,23}

Affiliations

¹Sorbonne Université, INSERM UMRS_974, Center of Research in Myology, 75013 Paris, France.

²Instituto de Salud Carlos III, Madrid, Spain

³Jackson Laboratory for Genomic Medicine, Farmington, CT 06032, USA.

⁴Inserm - University of Burgundy-Franche Comté, UMR1231 GAD, Dijon, France.

⁵Dijon University Hospital, Genetics Department, Dijon, France

⁶Dijon University Hospital, FHU-TRANSLAD, Dijon, France.

⁷Department of Human Genetics, Radboud University Medical Center, Nijmegen, The Netherlands.

⁸Radboud Institute for Molecular Life Sciences, Nijmegen, the Netherlands.

⁹Folkhälsan Research Center, University of Helsinki, Finland

¹⁰CNAG-CRG, Centre for Genomic Regulation (CRG), The Barcelona Institute of Science and Technology, Baldiri Reixac 4, Barcelona 08028, Spain.

¹¹Department of Genetics, Genomics Coordination Center, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands.

¹²Institute of Human Genetics, University of Bonn, Bonn, Germany.

¹³MRC Centre for Neuromuscular Diseases and National Hospital for Neurology and Neurosurgery, UCL Queen Square Institute of Neurology, London, UK.

¹⁴John Walton Muscular Dystrophy Research Centre, Translational and Clinical Research Institute, Newcastle University and Newcastle Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK.

¹⁵Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University Hospitals NHS Foundation Trust, Health Innovation Manchester, Manchester M13 9WL, UK.

¹⁶Manchester Centre for Genomic Medicine, Division of Evolution and Genomic Sciences, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester, Manchester, UK.

¹⁷Med Biotech Hub and Competence Center, Department of Medical Biotechnologies, University of Siena, Italy.

¹⁸Dipartimento di Medicina di Precisione, Università degli Studi della Campania "Luigi Vanvitelli," Napoli, Italy.

¹⁹Telethon Institute of Genetics and Medicine, Pozzuoli, Italy.

²⁰Genetics and Rare Diseases Research Division, Ospedale Pediatrico Bambino Gesù, IRCCS, 00146 Rome, Italy.

²¹Donders Institute for Brain, Cognition and Behaviour, Radboud University Medical Center, Nijmegen, The Netherlands.

²²Institute of Medical Genetics and Applied Genomics, University of Tübingen, Tübingen, Germany.

²³Centre for Rare Diseases, University of Tübingen, Tübingen, Germany.

²⁴Dijon University Hospital, Genetics Department and Centres of Reference for Development disorders and intellectual disabilities, FHU TRANSLAD and GIMI InstituteDijon, France.

²⁵Karolinska Institutet, Solna, Sweden.

²⁶Department of Biology and Medical Genetics, Charles University Prague-2nd Faculty of Medicine and University Hospital Motol, Prague, Czech Republic.

²⁷Department of Internal Medicine and Radboud Center for Infectious Diseases (RCI), Radboud University Medical Center, Nijmegen, The Netherlands.

²⁸Dept of Genetics, Assistance Publique-Hôpitaux de Paris - Université de Paris, Robert DEBRE University Hospital, 48 bd SERURIER, Paris, France.

²⁹Institute of Rare Diseases Research, Spanish Undiagnosed Rare Diseases Cases Program (SpainUDP) & Undiagnosed Diseases Network International (UDNI), Instituto de Salud Carlos III, Madrid, Spain.

³⁰Ospedale Pediatrico Bambino Gesù, Rome, Italy.

³¹Medical Genetics, University of Siena, Italy.

³²Genetica Medica, Azienda Ospedaliero-Universitaria Senese, Italy.

³³University Bordeaux, MRGM INSERM U1211, CHU de Bordeaux, Service de Génétique Médicale , F-33000 Bordeaux, France.

³⁴Service de Génétique Médicale, Centre Hospitalier Universitaire de Bordeaux, Bordeaux, France; MRGM, Maladies Rares: Génétique et Métabolisme, INSERM U1211, Université de Bordeaux, Bordeaux, France.

³⁵INSERM UMR 1141 "NeuroDiderot", Hôpital R DEBRE, Paris, France.