**Co-investigators**

Monique M. Ryan, MBBS, FRACP,MMed (Royal Children’s Hospital, Parkville, Victoria); Andrew J Kornberg, MBBS, FRACP (Royal Children’s Hospital, Parkville, Victoria); Victoria RodriguezCasero, MD (Royal Children’s Hospital, Parkville, Victoria), Alison Wray, MBBS (Royal Children’s Hospital, Parkville, Victoria), Kristi J. Jones, MBBS, PhD (The Children’s Hospital at Westmead, Westmead), Kathryn North, MD (The Children’s Hospital at Westmead, Westmead); Nathalie Goemans, MD (University Hospitals Leuven, Leuven), Gunnar Marceo Buyse (University Hospitals Leuven, Leuven); Craig Campbell, MD (Children’s Hospital); Jean Mah, MD (Alberta Children’s Hospital), Harvey Sarnat (Alberta Children’s Hospital); Kathryn Selby (Children’s & Women’s Health Centre of British Columbia & University of British Columbia); Thomas Voit (Groupe Hospitalier Pitie-Salpetriere); Valerie Doppler (Groupe Hospitalier PitieSalpetriere); Denis De Castro (Groupe Hospitalier Pitie-Salpetriere); Brigitte Chabrol, MD (Unite de Medecine Infantile); Nicolas Levy (Unite de Medecine Infantile); Cecile Halbert (Unite de Medecine Infantile); Yann Pereon (Laboratoire d’Exploration), Armelle Magot (Laboratoire d’Exploration); Julie Perrier (Laboratoire d’Exploration); Jean-Yves Mahe (Laboratoire d’Exploration); Ulrike Schara, MD (University of Essen), Soren Lutz, MD (University of Essen); Melanie Busse, MD (University of Essen), Adela Della Marina, MD (University of Essen);

Janbernd Kirschner, MD (University Hospital, Freiburg), Angela Stanescu, MD (University

Hospital, Freiburg), Annette Pohl, MD (University Hospital, Freiburg), Cornelia RensingZimmerman, MD (University Hospital, Freiburg); Enrico Bertini, MD (U.O. Medicina Molecolare di Malattie, Rome), Adele D’Amico, MD (U.O. Medicina Molecolare di Malattie, Rome), Annamaria Kofler, MD (U.O. Medicina Molecolare di Malattie, Rome), Adelina Carlesi, MD (U.O. Medicina Molecolare di Malattie, Rome), Anna Maria Bonetti, MD (U.O. Medicina Molecolare di Malattie, Rome), Luigino Santecchia, MD (U.O. Medicina Molecolare di Malattie, Rome), Francesco Emma, MD (U.O. Medicina Molecolare di Malattie, Rome), Gianluca Bergami, MD (U.O. Medicina Molecolare di Malattie, Rome); Eugenio Maria Mercuri, MD (Dipartimento di

Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome), Gessica Vasco, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome), Flaviana Bianco, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome), Elena Stacy Mazzone, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e

Neuroscienze, Rome), Roberto De Sanctis, MD (Dipartimento di Scienze Pediatric MedicoChirurgiche e Neuroscienze, Rome), Paolo Alfieri, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome), Marika Pane, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome), Sonia Messina, MD (Dipartimento di Scienze Pediatric Medico-Chirurgiche e Neuroscienze, Rome); Giacomo Pietro Comi, MD (Unita Operative di Neurologia, Milan), Francesca Magri, MD (Unita Operative di Neurologia, Milan), Valeria Lucchini, MD (Unita Operative di Neurologia, Milan), Stefania Paola Corti, MD (Unita Operative di Neurologia, Milan), Maurizio Gualtiero Moggio, MD (Unita Operative di Neurologia, Milan), Monica Sciacco, MD (Unita Operative di Neurologia, Milan), Nereo Bresolin, MD (Unita Operative di Neurologia, Milan), Alessandro Cesare Prelle, MD (Unita Operative di Neurologia, Milan), Roberta Magri, MD (Unita Operative di Neurologia, Milan), Roberta Virgilio,

Md (Unita Operative di Neurologia, Milan), Costanza Lamperti, MD (Unita Operative di

Neurologia, Milan); Yoram Nevo, MD (Hadassah Medical Center, Jerusalem), Taia DorWollman, MD (Hadassah Medical Center, Jerusalem),; Juan Vilchez, PhD (Hospital Universitario La Fe, Valencia), Nuria Muelas, MD (Hospital Universitario La Fe, Valencia), Teresa Sevilla, PhD (Hospital Universitario La Fe, Valencia), Patricia Smeyers, PhD (Hospital Universitario La Fe, Valencia), Alberto de la Osa, MD (Hospital Universitario La Fe, Valencia); Jaume Colomer, PhD (Hopsital Sant Joan de Deu, Barcelona), Carlos Ignacio Ortez, PhD (Hopsital Sant Joan de Deu, Barcelona), Andres Nascimento, PhD (Hopsital Sant Joan de Deu, Barcelona), Ana Febrer, PhD (Hopsital Sant Joan de Deu, Barcelona), Julita Medina, PhD (Hopsital Sant Joan de Deu, Barcelona); Mar Tulinus, MD, PhD (Avdelningen for Pediatrik, Gotenburg), brynja Thorarinsdottir, MD (Avdelningen for Pediatrik, Gotenburg), Niklas Darin, MD (Avdelningen for Pediatrik, Gotenburg): Thomas Sejersen, MD, PhD (Neuropediatriken,

Stockholm), Mia Hovmoller, MD (Neuropediatriken, Stockholm); Katherine Bushby, MD (Clinical Research Center, Newcastle upon Tyne), Volker Straub, MD (Clinical Research Center, Newcastle upon Tyne), Michela Guglieri, MD (Clinical Research Center, Newcastle upon Tyne), Anna Sarkozy, MD (Clinical Research Center, Newcastle upon Tyne), Tracey Willis, MD (Clinical Research Center, Newcastle upon Tyne), Michelle Eagle, MD, Anna Mayhew, MD (Clinical Research Center, Newcastle upon Tyne); Francesco Muntoni, MD (Great Ormond

Street Hospital, London), Sebahattin Cirak, MD (Great Ormond Street Hospital, London), Adnan Yousaf Manzur, MD (Great Ormond Street Hospital, London), Stephanie Ann Robb, MD (Great Ormond Street Hospital, London), Maria Kinali, MD (Great Ormond Street Hospital, London); Rosaline Christina Mary Quinlivan, MD (Robert Jones & Agnes Hunt Orthopedic Hospital, Oswestry), Martin Richard Smith, MD (Robert Jones & Agnes Hunt Orthopedic Hospital, Oswestry), Rajesh Pandey, MD (Robert Jones & Agnes Hunt Orthopedic Hospital, Oswestry); Brenda Wong, MD (Cincinnati Children’s Hospital, Cincinnati), James Collins, MD (Cincinnati Children’s Hospital, Cincinnati); Richard Finkel, MD (The Children’s Hospital of Philadelphia, Philadelphia), Cartsen Bonnemann, MD (The Children’s Hospital of Philadelphia, Philadelphia), Michele Yang, MD (The Children’s Hospital of Philadelphia, Philadelphia), Aileen Reghan Foley, MD (The Children’s Hospital of Philadelphia, Philadelphia), Sabrina Yum, MD (The Children’s Hospital of Philadelphia, Philadelphia); Jacinda Sampson, MD, PhD (University of Utah Medical Center, Salt Lake City), Mark Bromberg, MD (University of Utah Medical Center, Salt Lake City), Kathryn Swoboda, MD (University of Utah Medical Center, Salt Lake City); John Day, MD, PhD (University of Minnesota, Minneapolis), Peter Karachunski, MD (University of Minnesota, Minneapolis); Katherine Mathews, MD (University of Iowa, Iowa), Daniel Bonthius, MD (University of Iowa, Iowa), Karla Sue Laubenthal, MD (University of Iowa, Iowa); Basil Darras, MD (Children’s Hospital, Boston), Peter Kang, MD (Children’s Hospital, Boston); Julie Parson, MD (The Children’s Hospital, Colorado); Richard Barohn, MD (University of Kansas, Kansas City), Majed Dasouki, MD (University of Kansas, Kansas City), Heather Anderson, MD (University of Kansas, Kansas City), Jeffrey Burns, MD (University of Kansas, Kansas City), Mazen Dimachkie, MD (University of Kansas, Kansas City), Mamatha Pasnoor, MD, Yunxia Wang, MD (University of Kansas, Kansas City); Emma Ciafaloni, MD (University of Rochester, Rochester), Chad Heatwole, MD (University of Rochester, Rochester); Anne Connolly, MD (Washington University, St. Louis), Alan Pestronk, MD (Washington University, St. Louis), Muhammad Al-Lozi, MD (Washington University, St. Louis), Glenn Lopate, MD (Washington University, St. Louis), Paul Golumbek, MD (Washington University, St. Louis), Brian Sommerville, MD (Washington University, St. Louis), Leo Wang, MD (Washington University, St.

Louis), Anna Wojcicka-Mitchell, MD (Washington University, St. Louis), Andrew Godbey, MD (Washington University, St. Louis), Matthew Harms, MD (Washington University, St. Louis), Arun Varadachary, MD, PhD (Washington University, St. Louis), Stanley Iyadurai, MD, PhD (Washington University, St. Louis), Luisa Rojas, MD (Washington University, St. Louis); Susan Iannacone, MD (Children’s Medical Center, Dallas), Chaiyos Khonghatithum, MD (Children’s Medical Center, Dallas); Douglas Sproule, MD (Columbia University, New York), Darryl De Vivo, MD (Columbia University, New York), Andre Constantinescu, MD, PhD (Columbia University, New York); Craig McDonald, MD (University of California, Sacramento), Jay Han, MD (University of California, Sacramento); Ben Renfroe, MD (Northwest Florida Clinical Research Group, Gulf Breeze); Barry Russman, MD (Oregon Health Sciences University, Portland),

Michael Sussman, MD (Oregon Health Sciences University, Portland); Stephanie BurnsWechsler, MD (Duke University, Durham), Vern Juel, MD (Duke University, Durham), Lisa Hobson-Webb, MD (Duke University, Durham), Edward Smith, MD (Duke University, Durham).