

FOR IMMEDIATE RELEASE Kathy Moran, MBA <u>kmoran@acmg.net</u>

ACMG Foundation/PerkinElmer Diagnostics Travel Award winner is Cynthia S. Gubbels, MD, PhD

BETHESDA, MD - April 3, 2018 | Cynthia S. Gubbels, MD, PhD was honored as the 2019 recipient of the **ACMG Foundation/PerkinElmer Diagnostics Travel Award** at the American College of Medical Genetics and Genomics (ACMG) 2019 Annual Clinical Genetics Meeting in Seattle, Washington.

Dr. Gubbels was selected to receive the award for her platform presentation, "Rapid turnaround whole exome sequencing for critically ill neonates."

Cynthia Gubbels is a Research Fellow in the Division of Genetics and Genomics at Boston Children's Hospital, where she is studying the implementation of new genomic techniques in clinical medicine, to help facilitate earlier diagnosis and better treatment in children and adults with genetic disorders. She has an MD from Maastricht University (the Netherlands), did clinical residencies in pediatrics and clinical genetics, and obtained her PhD working on the reproductive system in classic galactosemia. She enjoys combining her clinical background with her main research interests, neurodevelopmental and metabolic disorders.

"I am deeply honored to have been awarded the 2019 Perkin Elmer Travel Award. It is such a privilege to join the ranks of distinguished awardees, and I would like to thank the selection committee and the ACMG Foundation for this great opportunity. To see my work validated in this way gives an enormous boost to my career and research and confirms that we are continually improving the way we diagnose and treat our patients by advancements made in research such as this. I aspire to continue to contribute to the field of medical genetics as both a physician and a scientist, and to be part of the future of medicine. I would like to thank my mentor and Principal Investigator, Dr. Timothy Yu, for the tremendous support that he has given me and my project. It has been a wonderful experience to work with and learn from him; he is truly a great teacher, scientist and person. I am also grateful for the support of Dr. Pankaj Agrawal for his guidance on this project and the opportunities that I have by working with him and the Manton Center for Orphan Disease Research at the Division of Genetics and Genomics at Boston Children's Hospital," said Dr. Gubbels.

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This award was created in 2008 by Signature Genomics to recognize an ACMG member and first author of a platform presentation abstract for the scientific program. The ACMG Program Committee selects the Travel Award recipient based on scientific merit. In recognition of the selected presentation, PerkinElmer Diagnostics covers the travel costs for the recipient to the ACMG Meeting.

"Travel to present their results and network with colleagues is a critical component of the training of medical genetics researchers. We greatly appreciate Perkin Elmer's continuing commitment to support of the Travel Award," said Bruce R. Korf, MD, PhD, FACMG, President of the ACMG Foundation for Genetic and Genomic Medicine.

The ACMG Foundation for Genetic and Genomic Medicine (ACMGF) is a community of supporters and contributors who understand the importance of medical genetics and genomics in healthcare. A non-profit organization established in 1992, the Foundation supports the American College of Medical Genetics and Genomics (ACMG) mission to "translate genes into health"; to foster charitable giving, promote training opportunities to attract future medical geneticist and genetic counselors, to share information about medical genetics, to sponsor important research and much more.

To learn more and support the ACMGF mission to create "Better Health through Genetics", please contact Nicole O. Bell, ACMG Foundation Manager, at <u>nbell@acmg.net</u> or 301-718-9604 or visit <u>acmgfoundation.org</u>.

Note to editors: To arrange interviews with experts in medical genetics, contact Kathy Moran, MBA, ACMG Director of Public Relations at <u>kmoran@acmg.net</u>.

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