

Rare / orphan genetic diseases: From phenotype to gene and molecular pathways

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Wilhelmina Cohen Auditorium, Pediatric Division, Soroka Medical Center, Beer-Sheva

May 17, 2018

09:00 – 09:30	Welcome and light refreshments
09:30 – 09:45	Opening remarks and introduction of the National Knowledge Center for Rare/Orphan diseases
09:45 – 10:15	Monogenic diseases: from phenotypes to genes, molecular pathways and disease prevention. Ohad Birk , <i>Genetics Institute, Soroka Medical Center and Shraga Segal Department of Microbiology, Immunology and Genetics, Ben-Gurion University.</i>
10:15 – 10:45	Genetic diseases in Israel - an overview. Joël Zlotogora , <i>Faculty of Medicine, Hebrew University of Jerusalem.</i>
10:45 – 11:15	From the genetic disease to the mechanism causing it: example from cardiomyopathy. Ruthi Parvari , <i>Shraga Segal Department of Microbiology, Immunology and Genetics, Ben-Gurion University.</i>
11:15 – 11:45	Coffee break
11:45 – 12:15	<i>C. elegans</i> as a model for human rare diseases: the role of fusion in shaping the mitochondrial network. Anat Ben-Zvi , <i>Department of Life Sciences, Ben-Gurion University.</i>
12:15 – 12:45	Induced pluripotent stem cells (iPSCs) as “disease in a dish” models for human inherited diseases. Rivki Ofir , <i>BGU-iPSC Core Facility, Regenerative Medicine & Stem Cell Research Center, Ben-Gurion University.</i>
12:45 – 13:15	The role of transcriptional enhancers in neurodevelopmental disorders. Ramon Birnbaum , <i>Department of Life Sciences, Ben-Gurion University.</i>
13:15 – 14:15	Lunch break
14:15 – 14:45	Elucidating the use of the fruit fly, <i>Drosophila melanogaster</i> , as a model for human rare diseases. Uri Abdu , <i>Department of Life Sciences, Ben-Gurion University.</i>
14:45 – 15:15	Mitochondrial-nuclear gene expression co-regulation and its role in health and disease. Dan Mishmar , <i>Department of Life Sciences, Ben-Gurion University.</i>
15:15 – 15:45	Quantitative views into the tissue-selectivity of hereditary diseases. Esti Yeger-Lotem , <i>Department of Clinical Biochemistry & Pharmacology, Ben-Gurion University.</i>