

הכנס השנתי של החברה הישראלית למחלות מטבוליות

19-20 בינואר 2026, מלון אלמא, זיכרון יעקב

THE MEETING OF THE ISRAEL SOCIETY FOR METABOLIC DISEASES (ISMD) Monday- Tuesday, January 19-20, 2026

Monday – 19.1.2026

11:30-12:30 Registration & Refreshments

12:30-14:15 Part A: Opening Lectures

Moderator: Prof. Yair Anikster, Sheba Medical Center

12:30 - 12:40 Welcome Greetings

ISMD committee

12:40-13:25 Gene Modifiers in Lysosomal Storage Disorders

Prof. Andy Klein

Universidad del Desarrollo, Chile

Prof. Andy Klein is an expert in lysosomal storage disorders (LSDs) and Parkinson's disease, focusing on the genetic and cellular basis of phenotypic variability. His research aims to identify novel biomarkers of severity and personalized therapeutics. His work in preclinical models includes the discovery of potential treatments for Niemann-Pick C and neuropathic Gaucher disease, using FDA-approved drugs

13:25-13:45 Heat Shock Dysfunction In HIKESHI-Related Hypomyelinating Leukodystrophy

Dr. Ayelet Zerem

Tel Aviv Medical Center, Israel

13:45-14:00 Disordered Eating in IEM Patients

Dr. Yael Finezilber

Sheba Medical Center, Israel

14:00-14:15 A Human Model Of NAD⁺ Depletion Reveals Metabolic Pathways Driving Neurodegenerative Vulnerability

Galit Gean-Akriv

Sheba Medical Center

14:15-15:00 Lunch

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15:00-16:50

Part B: Lysosomal Storage Diseases-A

Moderator: Dr. Stanley Korman, Rambam Medical Center

15:00-15:30

Complicated Pathway of MPS Diagnosis, Monitoring and Treatment

Prof. Orna Staretz-Chacham

Soroka Medical Center, Israel

(Sponsored by Medison)

15:30-15:45

Multorgan Fibrosis in Gaucher Disease Type 1: An Unmet Goal of Enzyme Replacement Therapy

Dr. Hanna Rosenbaum

Clalit Health Services Community Division, Israel

15:45-16:00

The Challenge of Immunomodulation in CRIM Negative Pompe Disease

Dr. Yoav Zehavi

HaEmek Medical Center, Israel

16:00 - 16:45

Lysosomal Mitochondrial Axis Dysfunction as Final Common Pathway in Lysosomal Diseases- Lessons From N-Acetyl-L Leucine in Niemann Pick Type C Disease

Prof. Marc C Patterson

Mayo Clinic, USA

(Sponsored by Neopharm)

Prof. Marc Patterson served as Professor of Neurology, Pediatrics and Medical Genetics at Mayo Clinic through his retirement in December 2024. Dr Patterson served as Editor for the Journal of Inherited Metabolic Disease and JIMD Reports, Editor-in Chief of the Journal of Child Neurology and Section Editor (Pediatric Neurology) for Up-To-Date through December 2024. His research and practice have focused on rare inherited metabolic disorders, with special interests in Niemann-Pick disease, type C (NPC), other lysosomal diseases, and congenital disorders of glycosylation, areas in which he has published more than 250 peer-reviewed papers and book chapters. Dr Patterson has served on the scientific advisory boards of several rare disease foundations and chaired the scientific advisory board for the International Niemann-Pick Disease Registry

16:45-17:15

Coffee Break

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17:15-18:40	Part C: Local Clinical Experience Moderator: Prof. Ann Saada, Hebrew University of Jerusalem
17:15 - 17:30	Update on COQ5 -Related Primary Coenzyme Q10 Deficiency Prof. Yair Anikster <i>Sheba Medical Center, Israel</i>
17:30- 17:45	The Challenge Of Dietary Treatment in Classical Homocystinuria Mrs. Miriam Behar-Shtriker <i>HaEmek Medical Center, Israel</i>
17:45 - 18:00	Glycomics And Confirmatory Method for Diagnostic Dilemmas in Congenital Disorders of Glycosylation Dr. Josh Manor <i>Sheba Medical Center</i>
18:00-18:15	GHF-201, A Novel Oral Compound for Adult Polyglucosan Body Disease (APBD) Demonstrates Promising Safety and Efficacy in A Compassionate Use Program. Prof. Or Kakhlon, <i>Hadassah Medical Center</i>
18:15 - 18:45	Pan-Ethnic Gaucher Disease: The Experience of a Single Center for Rare Diseases in Northern Israel Prof. Ronen Spiegel <i>Emek Medical Center, Israel</i> (Sponsored by Sanofi)
19:00-20:00	Dinner (For Hotel Guests)
21:00-23:00	Social Event

"ישראל במלחמה – היסטוריה נכתבת בתמונות" הרצאתו של זיו קורן

הצלם זיו קורן מציג תיעוד חזותי נדיר מרגעיה הדרמטיים של ישראל מאז מתקפת 7.10.23 ועד הלחימה מול איראן במבצע "עם כלביא". בהרצאה הוא משתף כיצד הצליח ללכוד את אותם "רגעים מכריעים" בשטח, ללא פילטרים, באופן שמאפשר לצופה לחוות את המציאות הישראלית כפי שהייתה בזמן אמת.

קורן הוא מצלם הדוקו הבולטים בישראל ובעולם, זוכה עשרות פרסים בינלאומיים, שצילומיו פורסמו בעיתונות המובילה והוצגו במאות תערוכות. בהרצאה הוא יחשוף סיפורים מאחורי הקלעים של עבודתו ויציג את כוחו של התיעוד לשמר זיכרון ולהשפיע על התודעה הציבורית.



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Tuesday – 20.1.2026

07:00-08:30

Breakfast (For Hotel Guests)

08:45-10:30

Part D: From The Lab To The Clinic and Vice Versa

Moderator: Prof. Orna Staretz-Chacham, Soroka Medical Center

08:45-09:10

First Diagnostic Mitochondrial Lab in Israel Present and Future

Dr. Rachel Rock

Sheba Medical Center, Israel

09:10-09:45

Genetics In Newborn Screening Program

Dr. Shlomo Almashanu

Ministry of Health, Israel

09:45-10:05

Precision And Velocity: Updates In Confirmatory Biochemical Services from Sheba Medical Center

Dr. Elena Dumin

Sheba Medical Center, Israel

10:05-10:25

Pitfalls In Clinical Metabolic Laboratory

Dr. Stanley Korman

Rambam Medical Center, Israel

10:25-10:40

From Metabolism to Genome: Is This the End of Biochemistry?

Prof. Eyal Reinstein

Maccabi Health Services, Israel

10:40-11:10

Coffee Break

11:10-13:00

Part E: Novel Treatments in Metabolic Diseases and Mitochondrial Diseases

Moderator: Dr. Galit Tal, Rambam Medical Center, Israel

11:10-12:00

Recent Advancement in Management of Phenylketonuria (PKU)

Prof. Maria Giżewska,

(Sponsored by Neopharm & PTC)

Head of the Department of Pediatrics, Rare Diseases and Metabolic Medicine at Pomeranian Medical University, Szczecin, Poland

Professor Maria Giżewska is Head of the Department of Pediatrics, Rare Diseases and Metabolic Medicine at the Pomeranian Medical University in Szczecin, Poland. A specialist in pediatrics and pediatric metabolism, she serves as a regional consultant for inborn errors of metabolism. Her research focuses on early diagnosis and treatment of metabolic disorders, particularly phenylketonuria, and she contributed to the European PKU guidelines. She has authored over 130 publications and has lectured worldwide. Prof. Giżewska has also co-led

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major Polish-German newborn screening projects and she is a promotor of the National Center of Science grant on the impact of lifelong low-phe diet on renal function in patients with phenylketonuria.

- 12:10-12:20 **Update On Two Novel Treatable Metabolic Disorders: DNAJC12 And PPCS Deficiencies**
Prof. Yair Anikster
Sheba Medical Center, Israel
- 12:20-12:40 **PKU Drug Repurposing for a Rare Cytoskeletal Disease**
Dr. Josh Manor
Sheba Medical Center, Israel
- 12:40-13:00 **Splice-Switching Oligonucleotide Rescues GFM2 Pseudo-Exon Inclusion in Fibroblasts of Individual with Combined Oxidative Phosphorylation Deficiency**
Dr. Nava Lotan, *Hadassah Medical Center*
Dr. Shira Gross, *Hadassah Medical Center*
- 13:00-14:00 Lunch**
- 14:00-15:50 Part F: Lysosomal Storage Diseases- B**
Moderator: Prof. Hanna Mandel, *Ziv Medical Centre, Zefat Israel*
- 14:00-14:20 **Incidence And Characteristics of Lysosomal Storage Diseases in the Negev Population: A Comparative Study Between Two Ethnic Populations**
Prof. Orna Staretz-Chacham
Soroka Medical Center, Israel
- 14:20- 15:00 **The Experience of Treatment Change in Pompe Patients from Two Centers In The South and the North**
Dr. Galit Tal, *Rambam Health Care Campus, Israel*
Dr. Merav Attias, *Soroka Medical Center, Israel*
- 15:00-15:30 **Modifiers Of Gaucher and Gcase Activity: Implications for Gaucher Disease Subtypes and Parkinson's Disease**
Prof. Andy Klein
Universidad del Desarrollo, Chile
- 15:30-15:50 **Hyperuricemia In Type 1 Gaucher Disease: Is Uric Acid A Biomarker for Disease Severity**
Dr. Zufit Hexner Erlichman
Clalit Health Services Community Division, Israel
- 15:50-16:00 **Closing Remarks**, Prof. Yair Anikster

16:00 End of Conference

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