ההסתדרות הרפואית בישראל
Israeli Medical Association

הכנס השנתי
של איגוד הגנטיקאים הרפואיים בישראל

תאריך	שעה	מיקום הכנס
26.11.2020	8:30-15:30	מקוון

Israeli Society of Medical Genetics
Scientific program 26/11/2020

08:30-08:40

Prof. Hagit Baris Feldman: Director of the Genetics Institute Tel Aviv Sourasky Medical Center and Chair of the Israeli Society of Medical Geneticists, Israel
Dr. Amihood Singer: Head of the community genetics department, Ministry of health, Israel

08:40-10:00

Chairman **Dr. Riki Sukenik Halevy** - Chosen abstracts 1:

- **Ms. Noy Azoulay** - Significant rate of abnormal findings in Prenatal Exome Trio in apparently normal fetuses. The Genetic Institute of Maccabi Health Services.
- **Dr. Idit Maya** - Is it time to report carrier state for recessive disorders in every microarray analysis? – A pilot model based on hearing loss genes deletions. Rabin Medical Center.
- **Dr. Yuval Weigl** - Variant re-classification in Shamir Medical Center: lessons from 290 analyses. Genetics Institute Shamir Medical Center.
- **Dr. Rachel Michaelson-Cohen** - Breast cancer risk and hormone replacement therapy amongst BRCA carriers following risk-reducing salpingo-oophorectomy. Shaare Zedek Medical Center.

10:00-10:30 | Coffee break

10:30-11:15

Chairman **Prof**. **Hagit Baris Feldman** –

Prof. **Gili Kenet**: Director of the Israel National Hemophilia Center and Thrombosis Institute and **Dr. Sarina-Levy** Mendelovich: Sheba Medical Center.

Treatment of genetic diseases in the new era-the future is here!

11:15-12:45

Chairman **Dr. Amir Peleg** - Chosen abstracts 2:

- **Ms. Vered Offen Glasner** - Whole-exome sequencing – a proposed first tier diagnostic test in fetus with central nervous system malformations. Sourasky medical center.
- **Dr. Lena Sagi-Dain** - Chromosomal microarray in 562 singleton pregnancies with polyhydramnios – do the degree and week of diagnosis matter? MOH.
- **Dr. Rivka Sukenik Halevy** - Should we report 15q11.2 BP1–BP2 deletions and duplications in the prenatal setting? Rabin Medical Center.
- **Dr. Odelia Chorin** - Transcriptome sequencing identifies a noncoding, deep intronic variant in CLCN7 causing autosomal recessive osteopetrosis, Institute for Rare Diseases, Tel Hashomer.

12:45-13:15 | Lunch break

13:15-14:40

Chairman **Dr. Riki Sukenik Halevy** - Chosen abstracts 3:

- **Dr. Jonathan Rips** - Parental exome analysis identifies shared carrier status for a second recessive disorder in couples with an affected child. Department of Pediatrics, Hadassah Ein-Kerem Medical Center.
- **Dr. Nitzan Gonen** - Pathogenic variants in the fourth zinc finger domain of Wilms' tumor 1 (WT1) gene are associated with 46,XX testicular/ovotesticular DSD, Dr. Nitzan Gonen, Bar-Ilan University.
- **Dr. Reeval Segel** - A defect in GPI synthesis as a suggested mechanism for the role of ARV1 in intellectual disability and seizure, Shaare Zedek Medical Center.
- **Dr. Lior Cohen** - SETD5 Gene Haploinsufficiency in Patients With Suspected KBG Syndrome. Barzilai Medical Center.

14:40-15.30

Chairman **Prof. Hagit Baris Feldman** –
Prof. Ran Balicer, MD, PhD, MPH: Founding Director of the Clalit Research Institute. Predictive proactive care in practice.