**Edmond J. Safra Center for Bioinformatics**

Jointly with

**School of Computer Science**

**DISTINGUISHED SPEAKER SERIES SEMINAR**

Prof. Ting Chen

Department of Computer Science, Tsinghua University

*"*Developing AI-Assisted Diagnosis Systems for Rare Diseases*"*

Wednesday, March 20 at 11:15

School of Computer Science, Check Point Building, Room 420

**Abstract:** Rare diseases, affecting approximately 350 million people worldwide, pose significant challenges in clinical diagnosis due to the lack of experienced physicians and the complexity of differentiating between numerous rare diseases. (1) To address these challenges, we developed PhenoBrain, a fully automated AI pipeline. PhenoBrain utilizes a BERT-based NLP model to extract phenotypes from clinical texts in electronic health records and employs new diagnostic models for differential diagnoses of rare diseases. The AI system was evaluated on diverse, multi-country rare disease datasets, and compared to specialist physicians. The results strongly support PhenoBrain’s seamless integration into the clinical workflow to narrow down candidate rare diseases and improve physicians’ diagnostic accuracy. (2) In addition, we developed AI based dialogue agents using reinforcement learning, which are capable of interacting with patients to perform automatic disease diagnosis.

Host: Prof. Ron Shamir, School of Computer Science, Tel Aviv University