

Rivka Carmi, Ben Gurion University of the Negev: “Lessons on Genes (and Genies) Learned from the Negev Bedouins”

ABSTRACT: The Negev Bedouin community is a tribal, traditional society in transition from a semi nomadic life style to a sedentary, urban one for the past 4 decades. Consanguinity is deeply rooted in the Bedouin culture and consequently rare recessive diseases are highly prevalent. For almost 3 decades I studied genetic disorders in the Bedouin community. Before the Human Genome Project era, those studies involved mainly observations and clinical delineation of known and unknown syndromes. After HUGO the research was set at a systemic identification of recessive new genes/mutations in known genes for various genetic diseases in that community, first by linkage analysis and later by applying new genomic technologies to identify gene mutations. This research has resulted in the findings of numerous new genes and mutations in known genes and also, new insights and hypothesis related to genes functions and roles in bringing about certain phenotypes.

In parallel to bench work, we were involved in challenging community based projects to promote accessibility to genetic testing, both prenatal and carrier detection, in a highly traditional society. Those programs were aimed at various audiences within the community, using different methodologies dependent on the specific target population.

Those projects were followed by a socio-anthropological research that provided deep insights into approaches and attitudes towards genetic knowledge and usage of genetic diagnosis among community members, based on cultural believes and perceptions.

This talk will discuss insights gained on both the biological and the sociological aspects of the genetic research in the Negev Bedouin community.