



Wissam Saleh
Molecular Geneticist
St. John Eye Hospital Group

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The molecular genetics research lab at St John Eye Hospital was established in 2017. Our research team is comprised of two clinicians specialized in retina and genetics (Dr. Alaa Al Talbishi & Dr. Yahya Swaiti) and two molecular geneticists (Wissam Saleh & Manar Salameh). We mainly focus on studying the genetics of inherited retinal dystrophies (IRDs) in the Palestinian population. IRDs are a group of disorders characterized by the loss or dysfunction of retinal cells and/or the choroid with a high genetic and clinical heterogeneity. There's a high prevalence of these disorders in Palestine compared to other studied populations due to the high rate of consanguineous marriage in the Palestinian community.

Although there are currently no treatments available for these disorders, the genetic testing that we do in our lab has multiple benefits. Firstly, it helps confirm the clinical diagnosis provided by the clinician. Secondly, the genetic result is critical for genetic counseling, risk assessment and family planning. Thirdly, our research work contributes to the large number of research studies being done worldwide to help better understand the disease mechanisms of these disorders which will translate into the development of treatment options in the future, including gene therapy approaches.

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At the moment, we have around 700 Palestinian families with IRDs recruited for our study. These patients are referred to our retinal dystrophy clinic where the clinician provides a provisional diagnosis based on the clinical examination, diagnostic testing and Imaging done at the hospital. When the patient is recruited, we explain the purpose and benefits of this research study, collect necessary data and blood samples and obtain an informed consent from the patient or their guardians to participate in the study.

After that, we extract DNA from collected blood samples and perform genetic screening for known mutations associated with these diseases. In some cases, we send our samples to collaborating labs in order to perform a more comprehensive and advanced genetic testing using next generation sequencing (NGS) techniques.

We analyze the NGS data and confirm the results in our lab to solve these cases and determine the underlying genetic cause for each case. We are currently in the process of preparing a research article for the Palestinian cohort of IRD families in collaboration with the Hebrew University. This paper will serve as an important reference for better diagnosis and understanding of IRDs in Palestinian patients.

For our future goals, we are aiming to expand the research work that we do in our lab to include cell culture and protein experiments and perform functional studies. We would also like to provide better training in bioinformatics for our team members to help further improve their data analysis skills. Another important goal of ours is to organize more IRD awareness campaigns in the Palestinian community especially in high disease risk regions. We also hope to recruit a genetic counsellor to join our team in the future. This is necessary to help provide genetic counseling services to patients after receiving their genetic diagnosis to determine the probability of having affected offspring and explore available prevention options. Lastly, we hope to participate in gene therapy clinical trials to contribute to the advancement of scientific research as well as provide our patients the opportunity to receive treatments once they are available in the future.



London Support had the pleasure of meeting with Wissam in her laboratory in Jerusalem on a recent visit, where we were able to discuss the vital importance of the work carried out.