St John Eye Hospital Group

Current Research Projects (September 2020)

(1) Retinal Genetics Research

Dr Altalbishi, Dr Alsweiti

The Inherited retinal Degenerations research Unit was established in St John Eye Hospital in 2017 following a generous grant from the European Union and in collaboration with Hadassah Hebrew university medical centre.

Currently more than 420 families with Inherited retinal degenerations are recruited, around 50% of them are genetically diagnosed, and work is on-going to search for the diagnoses in the remaining families.





4 peer-reviewed papers have been published from this work, covering a spectrum of retinal inherited diseases and their genetics. As of August 2020, a further 2 papers have been submitted for publication.

Furthermore, there are also candidate genes which are thought to be causing retinal degenerations not hitherto reported, which our research team are working on.

On-going efforts are underway to enrol the hospital's genetics research unit in gene therapy trials; SJEHG has a large pool of patients who are good candidates for such trials.

Publications:

- 1. Khan M, Cornelis SS, Pozo-Valero MD, et al **(incl AlSwaiti Y, Al Talbishi A).** Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. *Genet Med.* 2020;22:1235-1246.
- 2. Al Talbishi A, Zelinger L, Zeitz C, et al. TRPM1 Mutations are the Most Common Cause of Autosomal Recessive Congenital Stationary Night Blindness (CSNB) in the Palestinian and Israeli Populations. *Sci Rep.* 2019;9:12047.
- 3. Abu Diab A, **Al Talbishi A**, Rosin B, et al. The combination of whole-exome sequencing and clinical analysis allows better diagnosis of rare syndromic retinal dystrophies. *Acta Ophthalmol.* 2019;97:e877-e886.
- 4. Khalaileh A, Abu-Diab A, Ben-Yosef T, et al **(incl. Al Swaiti Y).** The Genetics of Usher Syndrome in the Israeli and Palestinian Populations. *Invest Ophthalmol Vis Sci.* 2018;59:1095-1104.

(2) Glaucoma Research

Dr Shawar, Dr Muhsen

- **1.** Evaluation of the demographic profile and subtypes of glaucoma patients at St John Eye Hospital. Pending publication.
- 2. The effect of phacoemulsification cataract extraction with posterior chamber lens implantation on intraocular pressure in different glaucoma types . Pending publication.
- Outcomes of transscleral diode laser cyclophotocoagulation in the treatment of refractory glaucoma. In progress
- 4. **Chronic angle closure glaucoma in spherophakia (case report).** Pending publication.
- 5. **Brimonidine induced anterior uveitis (case report) .** Pending publication.
- 6. **Pseudoexfoliation an autophagy disorder (literature review) .** Pending publication.

(3) Rapid Assessment of Avoidable Blindness (RAAB)

The RAAB study was launched at St John Eye Hospital in August 2018, and fieldwork was completed in May 2019. This study is the second of its kind in the Palestine. The first, conducted in 2008, was a vital tool in identifying the priorities for tackling preventable blindness across the country . (Publications pending).

(4) Uveitis Research

Dr Erakat

1. Vogt Koyanage Harada spectrum of disease and treatment options in collaboration with Moorfields (analysis stage)

2. Juvenile Idopathic Arthritis and uveitis (Submission stage)

3. Retinal Vessels occlusion in Behcet disease (Submission stage)