

# Delivering industry research in advanced therapies for rare diseases in ophthalmology

Marcela Votruba MA BM BCh PhD FRCOphth FLSW  
Professor of Ophthalmology & Hon. Consultant Ophthalmologist  
Welsh Specialty Lead for Ophthalmology  
University Hospital Wales & School of Optometry & Vision Sciences, Cardiff University

Hayley Westwood  
Senior Research Co-ordinator  
Ophthalmology  
University Hospital Wales, Cardiff

## Purpose

The overall purpose was to increase interactions with and foster trials in ophthalmology in the rare disease advanced cell & gene therapy space.

## Method

- The first step was to actively engage with and build links with biotech and pharmaceutical companies scoping cell and gene therapy applications and trial readiness across a range of ocular genetic rare diseases.
- The second step was to actively bid for clinical trials in this space.
- The third step was to make use of a Cardiff University NMHII secondment to a commercial partner to initiate a two-way process of knowledge sharing, collaboration and development of opportunities and ideas

## Conclusion

Highly rewarding ongoing and increasing engagement and results on 3 levels:

1. personal learning and development,
2. increasing trials and especially entering the gene and cellular therapy space,
3. access to trials for Welsh patients increased providing equity.

## Results

In 12 months:

**Step 1-** scoping and network building with 6 companies.

**Step 2-** 4 trials- publications arising from two studies currently under review in major neurology journal, one study recruiting to target, one study bidding for site selection complete.

**Step 3-** successful secondment to Cambridge based start-up developing an RNA platform for protein upregulation in ocular haplo-insufficiencies and neurodegenerative disorders.

- . Generation of, and interpretation of preclinical data from disease models for its lead ocular programme.
- . Mapping out the regulatory/preclinical/clinical path for its lead ocular programme, engaging with KOLs and regulators.
- . Evaluating and selecting targets for the company's future indications in the ocular and neurodegenerative space.