Therapy for retinal degeneration in ciliopathies

Pharmacological therapy to slow significantly retinal degeneration linked to protein trafficking defect inducing a Unfolded Protein Response

**KEYWORDS**
- Photoreceptor cell,
- Retinitis pigmentosa,
- Cellular protein overload,
- Endoplasmic reticulum stress,
- Apoptosis,
- rare diseases

**TECHNOLOGY**
- Impaired intraciliary transport observed in Bbs12-/- murine model
- Results in protein retention in the endoplasmic reticulum
- This leads to a proapoptotic unfolded protein response (UPR) triggering death of the photoreceptors
- BiP, and PERK-mediated phosphorylation of eIF2a have been identified as 2 key UPR actors (Mockel et al., 2012)
- GV-ReT is a combination of clinically validated molecules (Valproic Acid and Guanabenz) for the treatment of cilia-related retinal degeneration

**APPLICATION**
- Pharmacological therapy for ciliopathy related retinal degeneration

**INNOVATION ADVANTAGES**
- A combination of two approved drugs, with a well defined MoA
- Effective in protecting retinal cells from dying and significantly increasing light detection capacity
- Aims at preserving the vision loss and slowing the retinal defects to cure secondarily with a more targeted therapy
- Positions on early onset of the retinal degeneration
- Applicable to different paradigms of retinal degeneration: UPR-inducing ciliopathies, retinal degeneration linked to protein trafficking defect inducing a UPR

**DEVELOPMENT STATUS**
- Compelling ex vivo and in vivo data establishing the efficacy of this drug combination
- Efficacy assessed on different cilia-related retinal degeneration (Bardet Biedl syndrome, Leber Congenital Amaurosis)

**PATENTS**
- WO2013124484

**INVENTORS**
- H. Dollfus
- V. Marion
- A. Mockel
- Laboratory of Medical Genetics
- Unistra / INSERM

**CONTACT**
Nathalie Lenne
Business Developer Healthcare
Phone: +33 (0)6 09 79 06 13 - nathalie.lenne@satt.conectus.fr