EUR-USH Consortium

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PI's second kick-off meeting
Mainz, 26.09.2014

Further information

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Permanently updated:
http://eur-ush.eu/

European young investigators network for Usher syndrome

EUR-USH

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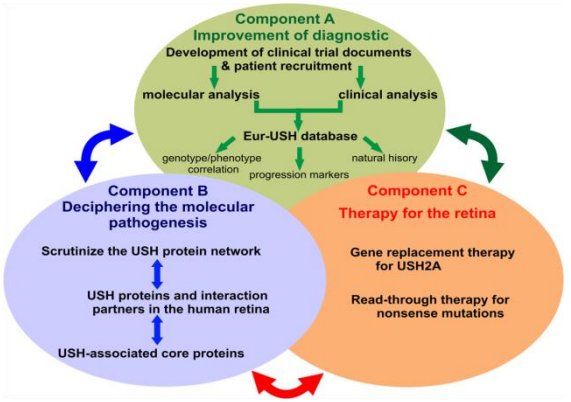
2013 - 2016

European young investigators network for Usher syndrome



- Usher syndrome (USH) is a rare (~3/100.000) genetic disease with autosomal recessive pattern, resulting **major handicap** for patients, since it affects two major senses, vision and hearing.
- Currently, there is **no treatment available** for Usher syndrome. The auditory deficit in USH can be compensated by the use of hearing aids and cochlear implantation, but the retinal problems remains unsolved and leads to a loss of peripheral vision.
- First, USH is a **clinically and genetically heterogeneous** disorder, making diagnosis and treatment challenging. Second, to date existing **databases** and rare material collection **are local, small** and not accessible or standardized**.** Third, **natural history** of USH ophthalmic component is **not fully understood**.

EUR-USH : the three components are each interconnected to make up the full project

- *The combination of state-of–the-art clinical examinations, molecular analyses (e.g. NGS) and developed database will improve diagnosis.*
- *Basic research (proteomics, imaging) will enlighten the mechanisms of the molecular pathogenesis.*

If you/your family members are affected by Usher syndrome and/or willing to be included in EURUSH clinical trial, please contact: evicrnet@aibili.pt

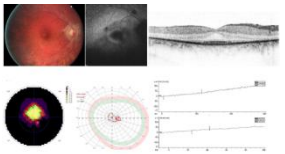
If you are interested in European wide database and/or looking for cooperation partners: ieva.sliesoraityte@inserm.fr

Component A

European wide genotype/ phenotype related database development (P4).

Up to ~500 USH patient data will be uploaded

Usher syndrome diagnostic platform with significant markers elaboration for clinical therapy trials (P3, P4, P5).



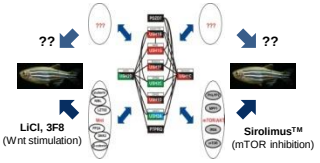
http://clinicaltrials.gov/show/NCT01954953

EURUSH study currently recruiting patients (P3, P4).

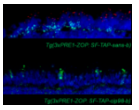
Component B

Scrutinize the USH protein interactome and assess the role of USH proteins in retina development and function (P6).

Protein complexes isolated from HEK293T cells

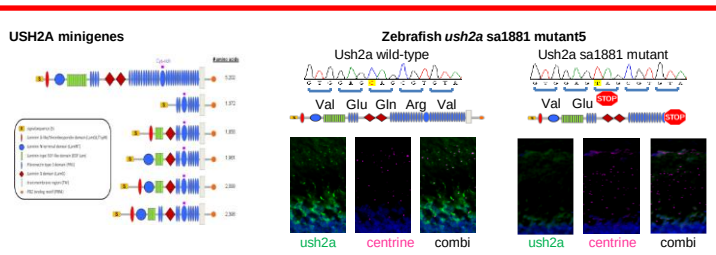


Expression, subcellular distribution and co-localization of USH proteins and novel USH network patterns in human retina (P1).



Retinal expression of Strep-FLAG-tagged sans_b and cip98_b under the control of an enhanced zebrafish opsin promoter.

If you are searching for well described USH patient cohort or potential partners for therapy trials in future: nagelwol@uni-mainz.de erwin.vanwyk@radboudumc.nl



Component C

Gene replacement therapy for USH2A related retina degeneration (P6).

Read-through therapy for the retinal phenotype (P1).

- *Gene-based therapies will be evaluated: gene augmentation and translational read-through, to treat the progressive retinal degeneration*