

## Concept for genetic testing in CI candidates in Switzerland

### Introduction:

The guidelines for cochlear implants [ref SGORL] recommends genetic testing in deaf adults and children in the work-up of implant candidacy since 2018. Genetic analysis can have a clinical impact on decision making and follow up strategies especially in undetected syndromic patients. With the availability of the first gene therapy for deaf patients affected by a mutation of the otoferin gene [ref.] genetic testing is a prerequisite to identify potential candidates and to inform deaf patients looking for information.

Therefore, the CICH confirms the recommendation for genetic testing in deaf patients in the work-up of cochlear implant candidacy. The following procedure is proposed:

1. A solid and permanent collaboration with the local geneticists needs to be established in each CI center to initiate adequate patient counselling before and after molecular testing. The creation of a genome board between geneticist and ORL is typically needed for adequate interpreting of molecular results and elaboration of the final molecular diagnosis.
2. The medical staff of the CI center encourages deaf patients or their parents for genetic testing. For basic information about the procedure, a leaflet can be provided.
3. The geneticist sees the patients and their families before molecular testing and obtains the financial authorization for reimbursement by the insurance companies.
4. The medical staff of the CI center can help to obtain the needed blood sample particularly in children, where they are ideally obtained during general anesthesia during the CI preoperative procedure such as MRI or AEP.
5. The geneticist informs the patients about the molecular result (inheritance mode, other affected organs in syndromic cases).
6. The Swiss CI centers work together to anonymously store the molecular diagnosis in the CICH databank along with the functional results.

### Actual proposed molecular test:

- There are different options available, which need to be discussed with the local geneticist.
- In Geneva, whole exome sequencing is performed (WES)
- Standardized panels should be updated on a regular basis and recommendations of the geneticists should be provided to the CICH group.

28. Mai 2024

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CICH Genetic testing, Version 1, Last update 28.5.2024