Cochlear Implant & Usher Syndrome

Why early diagnosis matters and why it is relevant for the CI community and for European health policies

Genetic diagnosis – Why?

- Perspective parent
- Perspective first contact & family advisor
- Perspective patient representative & advocate
- National & International
- European & Austrian perspective

Genetic diagnosis – Why?

- Personal/family
- Individual -> CI & wider patient community
- Beyond -> other stakeholders ->
- Patients & families

Usher Syndrome

- Combined hearing loss & vision loss -> deaf blindness
- Vestibular dysfunction some subtypes
- Rare genetic (recessive)
- Congenital HL profound (USH1), moderate or light (USH2, USH3)
- VL caused by RP (Retinitis pigmentosa)
  - Night blindness, ↓ visual field, ↓ visual acuity
  - Early childhood (USH1), adolescence & adults (USH2, 3)

Genetic diagnosis – Why?

- Diagnostic journey
- All stages of diagnosis
  - No diagnosis at all at birth (1996)
  - 1 symptom (1997)
  - RP -> presumed, suspected, likely (2005)
  - Genetically confirmed (2012 at age 16)

Genetic diagnosis – Why?

- Relief!
- Cause, explanation, certainty, no guilt
- before: uncertainty
  - Deafness
  - Night blindness
  - Delayed motor skills – balance
  - Vertigo, seizures from age 11
  - What else??
- More targeted & effective actions

Personal/family

- Parents responsible <- teenagers, adults
- Create adequate environment: school, hobby, career, family, friends, …
- Connect to patient groups
- Targeted coping strategies – reduce individual burden
- Build individual resilience & support children thriving

Personal/family

- USH: treat hearing disorder -> CI, HA -> compensate VL
- USH1: vestibular dysfunction -> balance training -> relevant with VL
- Create identity – belong somewhere
- **Family planning & carrier screening – how likely…?**
- Access experts, support (medical, financial, social, disability services)
- Treatment, management, research, clinical trials

CI Community & Usher Syndrome

- 1/3 genes hearing loss -> USH
- 10% congenital HL/deafness -> USH1
- > 50% combined vision &hearing loss/deaf blindness -> USH1, USH2, USH3
- < 40% all USH -> USH1, < 60% - USH2, < 3% USH3
- 25% USH1b, 50% USH2a
- USH1 CI candidate as a baby
- USH2 or 3 CI candidate sooner or later in life

Wider patient community

- Build network & patient/clinician & research partnerships
- Peer to peer consulting to families
- Exchange & build knowledge and coping strategies
- Create powerful community
  - Patients & families
  - HC professionals
  - Support services
  - Clinicians, researchers, scientists
  - Health policy makers & stakeholders

Meta level – What else is in stake?

- Physicians, clinicians, researchers, scientists, HC systems
- Create knowledge – **education** of future experts
- Patient pathways & patient referrals
- European Reference Network for Rare Diseases: ERN-Eye, ERN CRANIO (ENT disorders)
- Informed, more **targeted treatment & disease management** & reduce ineffective … **Reduce burden for HC systems** & social

Meta level – What else is in stake?

- Genetically confirmed diagnosis –> **coding** ORPHA, ICD10 or ICD11 (to be applied during all stages of diagnostic pathway!)
- Applied to data sets -> **tracking & visibility in systems**
- Build registries: ERNs, EHDS, national health data hubs (e.g. hospital level)
- Basic -> translational research, build individual cohorts, clinical trials, innovative treatments

Meta level – What else is in stake?

- Sustainability HC systems, information for health planning & social systems
- Planning of HC, research, education
- Identify & access/visit relevant patient data
- Identify burden of disease: individual & society
- Estimate costs, access & allocate **appropriate funding to patients, hospitals, research**

Conclusion

- Value of early genetic diagnosis far beyond individual/family aspects -> allows to
  - Inform health & social systems
  - Estimate costs, plan resources, allocate funding
  - Access expert knowledge (ERNs, CBHC)
- Define educational & other actions, psychosocial support needed
- Give timely access to clinical trials & (upcoming innovative) treatments

Call

- Only early genetically confirmed diagnosis at MUC?
- Also refer adolescents & adults in any (ENT) settings (also non-clinical) with hearing disorder & signs of vision loss to genetic diagnosis?
- Identify ALL individuals with USH, code?
- Give certainty & visibility, inform health systems & social systems, inform research & science -> better disease management, integrated care, innovative treatments?

Actions?

- How can Euro-CIU and CIA and the wider CI community support this?
- What can I do to support this within Euro-CIU and CIA and the wider CI community?