If you can’t look back and you can’t look forward,
You better look up!!
The Swedish Usher interdiciplinary research group
Swedish database of deafblind syndromes

- Started 1988 in cooperation with BTNRH (Kimberling)
- Subcontractor NIH
- 26 years to complete !! ???
- All subjects examined by me
- 2014 460 families with Usher type 1,2 and 3
- Expected total prevalence in Sweden 600
- 200 families with other deafblind syndromes
Do I have a patient with a problem ??
Does this child have a syndrome?
Maybe there is a problem?
Teenage!!

I would like to be as all other young people !!!

Late diagnosis

Denial

Severe consequences
Confusion

- Why is my child deaf?
- Why is my child clumsy?
- Why has my child problems at night?
- Why don´t I like gymnastics and sports?
- Why can´t I have a drivers licence?
- Why can´t I hear, I have good hearing?
- Why can´t I go on working in the factory?
- What is Deafblindness?
How do I diagnose possible deafblindness and Usher?

- Pedigree
- Dysmorphology
- Neurology, Otolaryngology,
- Audiology
- Vision
- Balance Function
- Radiology
- Kidney
- Heart
- Tests for Viral Infections
- Genetics
Rehabilitation in children

- Examinations
- Genetics
- Information
- Shock, denial, confusion
- Crisis intervention
- Don’t forget the child and relatives
- My child is not deafblind !!! , it has a combined vision - hearing problem
Rehabilitation in adults

- Late diagnosis
- Driver license
- "wrong occupation"
- Denial vision problems
- Severe psycho-social crisis, work, spouse, children, friends
- Subjective hearing loss
- Change of communication mode
- No cooperation
The bar chart shows the number of subjects in different age groups for Usher I, Usher II, and Usher III. The subjects are divided into age groups: 0-9, 10-19, 20-29, 30-39, 40-49, 50-59, 60-69, 70-79, and 80-89. There are two categories: with diagnose and without diagnose.

- Usher I: The highest number of subjects is in the 40-49 age group, with a significant drop in the number of subjects in the 30-39 age group.
- Usher II: The highest number of subjects is also in the 40-49 age group, with a significant drop in the number of subjects in the 30-39 age group.
- Usher III: The highest number of subjects is in the 50-59 age group, with a significant drop in the number of subjects in the 40-49 age group.

Without diagnose:
- The number of subjects decreases as the age group increases.
- The number of subjects is significantly higher in the younger age groups compared to the older age groups.

With diagnose:
- The number of subjects decreases as the age group increases.
- The number of subjects is significantly higher in the younger age groups compared to the older age groups.
Auditory rehabilitation adults

- early correct diagnosis
- Adjust -change work task
- Close cooperation -hearing-vision
- Psychosocial intervention
- State of the art equipment, computers
- Discuss total communication, sign language, braille etc
- Vision rehab fails often, the most important
- Centralization to few clinics -teams??
Rehab in Usher II

- Hearing loss from birth
- Vision problems slow progression in childhood
- I don’t know how other see!
- Difficulties in school, blame it on the hearing loss
- Contrast, light sensitivity, night blindness
- Denial
- Career, children, family
- “this is not according to my plans”
- Acceptance, the light in the tunnel!
- Team support
Study 1

"Physical and Psychological Health among persons with Deafblindness due to Usher syndrome type 2"
(Wahlqvist et al. 2013)

Swedish National Public health questionnaire, "Health on equal terms"
HAD-scale

The aim was to describe physical and psychological health among persons with Usher type 2, and to explore if there was differences between men and women compared to a cross section of the Swedish population
Population

<table>
<thead>
<tr>
<th></th>
<th>USH2</th>
<th>Referens</th>
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<tbody>
<tr>
<td>N</td>
<td>96</td>
<td>5738</td>
</tr>
<tr>
<td>Age</td>
<td>55</td>
<td>58</td>
</tr>
<tr>
<td>Age range</td>
<td>18-84</td>
<td>23-91</td>
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<tr>
<td>Females(%)</td>
<td>53</td>
<td>56</td>
</tr>
<tr>
<td>Genetic diagnosis</td>
<td>59%</td>
<td></td>
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<tr>
<td>Pure tone average</td>
<td>73db</td>
<td></td>
</tr>
<tr>
<td>Visual field</td>
<td>4 (&lt;10°)</td>
<td></td>
</tr>
<tr>
<td>Visual acuity</td>
<td>0,4</td>
<td></td>
</tr>
</tbody>
</table>
**Physical and psychological bad health**

- **Percent**

- **Reference**
  - Headache
  - Tinnitus
  - Pain shoulders, neck
  - Eczema, skin rashes
  - Anxiety, worry
  - Fatigue
  - Sleeping problems
  - Concentration
  - Accomplished things
  - Unhappy and depressed
  - Constantly under strain
  - Worthless
  - Suicide thoughts
  - Suicide attempts

- **Usher type II**
  - Headache
  - Tinnitus
  - Pain shoulders, neck
  - Eczema, skin rashes
  - Anxiety, worry
  - Fatigue
  - Sleeping problems
  - Concentration
  - Accomplished things
  - Unhappy and depressed
  - Constantly under strain
  - Worthless
  - Suicide thoughts
  - Suicide attempts

*Sign p ≤ 0.05*
Study 2 USH3 (preliminary results)

The aim was to describe and analyze data on vision and hearing impairment, physical-, psychological health and social risk among persons with USH3

15 persons with USH3

Mean age 41 years

11 women and 4 men

Swedish National Public health questionnaire, ”Health on equal terms”, and HAD-scale
Psychological poor health

- Poor psychological health
- days ≥15
- Anxiety, worry
- Fatigue
- Sleeping problems
- Concentration problems
- Unable to appreciate the day
- Unable to manage problems
- Lost sleep over worry
- Unhappy and depressed things
- Incapable of making decisions
- Loosing confidence
- Constantly under strain
- Worthless
- Up to problems
- Unhappy
- Stress
- Suicide attempts
- GHQ12 Poor wellbeing
- Anxiety
- Depression

Legend:
- USH1
- USH2
- USH3
Why is it important to know the cause early?

- To get a correct diagnose
- To learn from other experiences
- To determine the *prognosis* of the disorder
  - Progression of hearing loss.
  - Usefulness of aids and/or cochlear implants.
  - Progression of vision loss.
  - Career planning
- Correct rehabilitation
- Treatments are expected to be specific to the cause
- Patients and family often want and need a reason for the problem.
Thank you !!!!