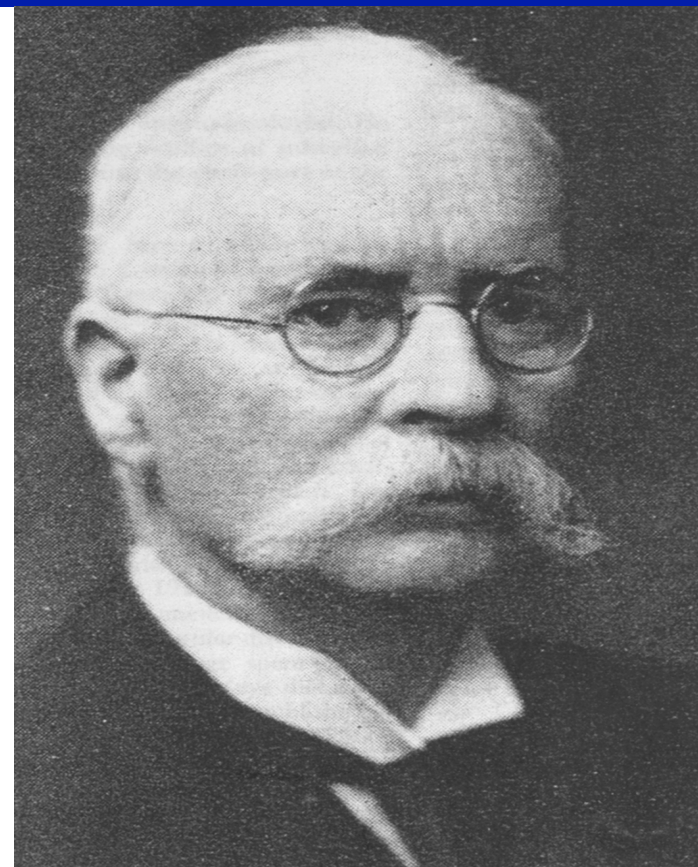


If you can't look back and  
you can't look forward,  
**You better look up!!**



# The Swedish Usher interdisciplinary research group



# Swedish database 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

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- 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

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- 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



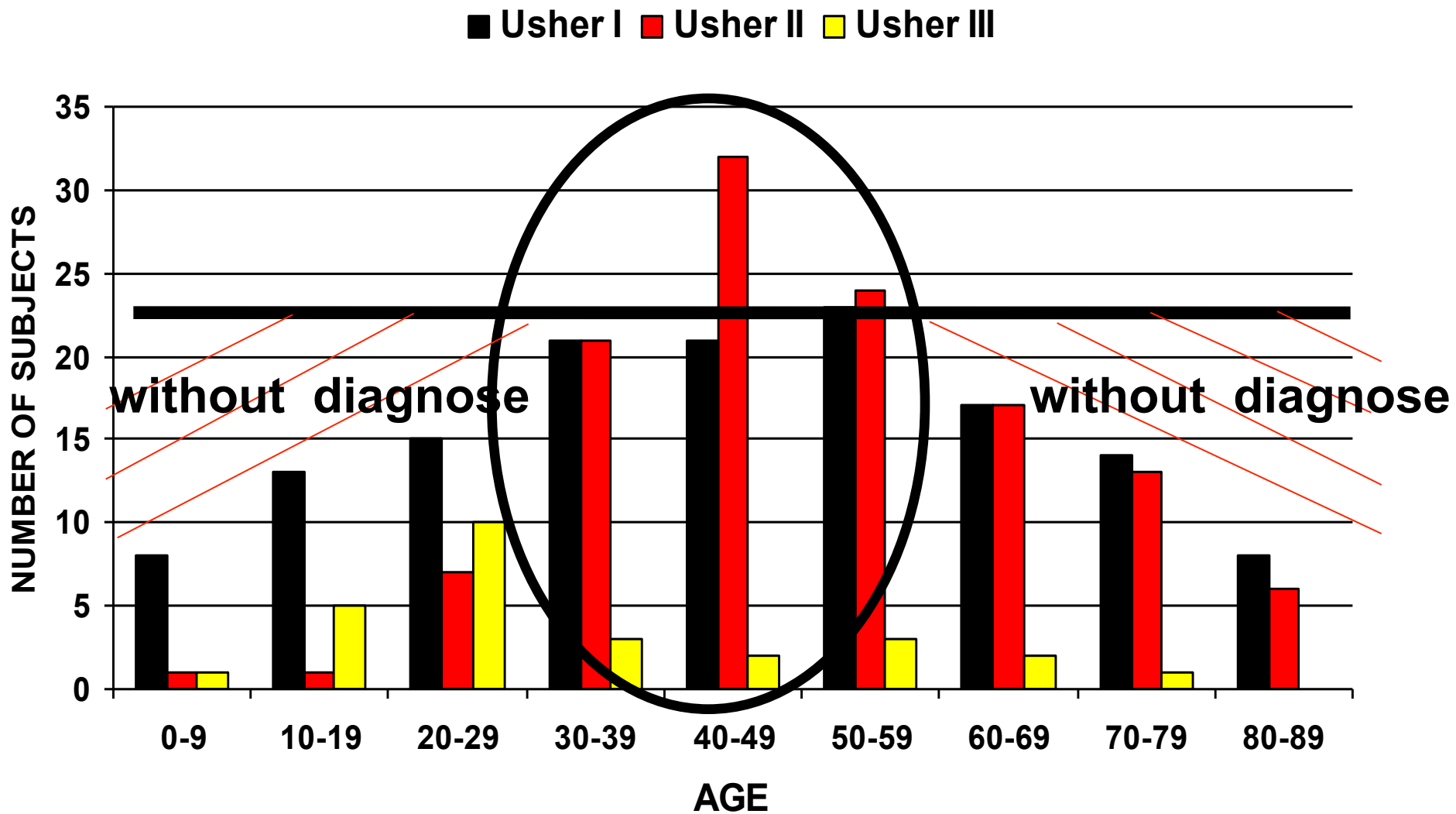


# Rehabilitation in adults

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- 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







# Auditory rehabilitation adults

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- 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

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- Hearing loss from birth
- Vision problems slow progression in childhood
- I don't know how other see!
- Difficulties in school, blaim 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



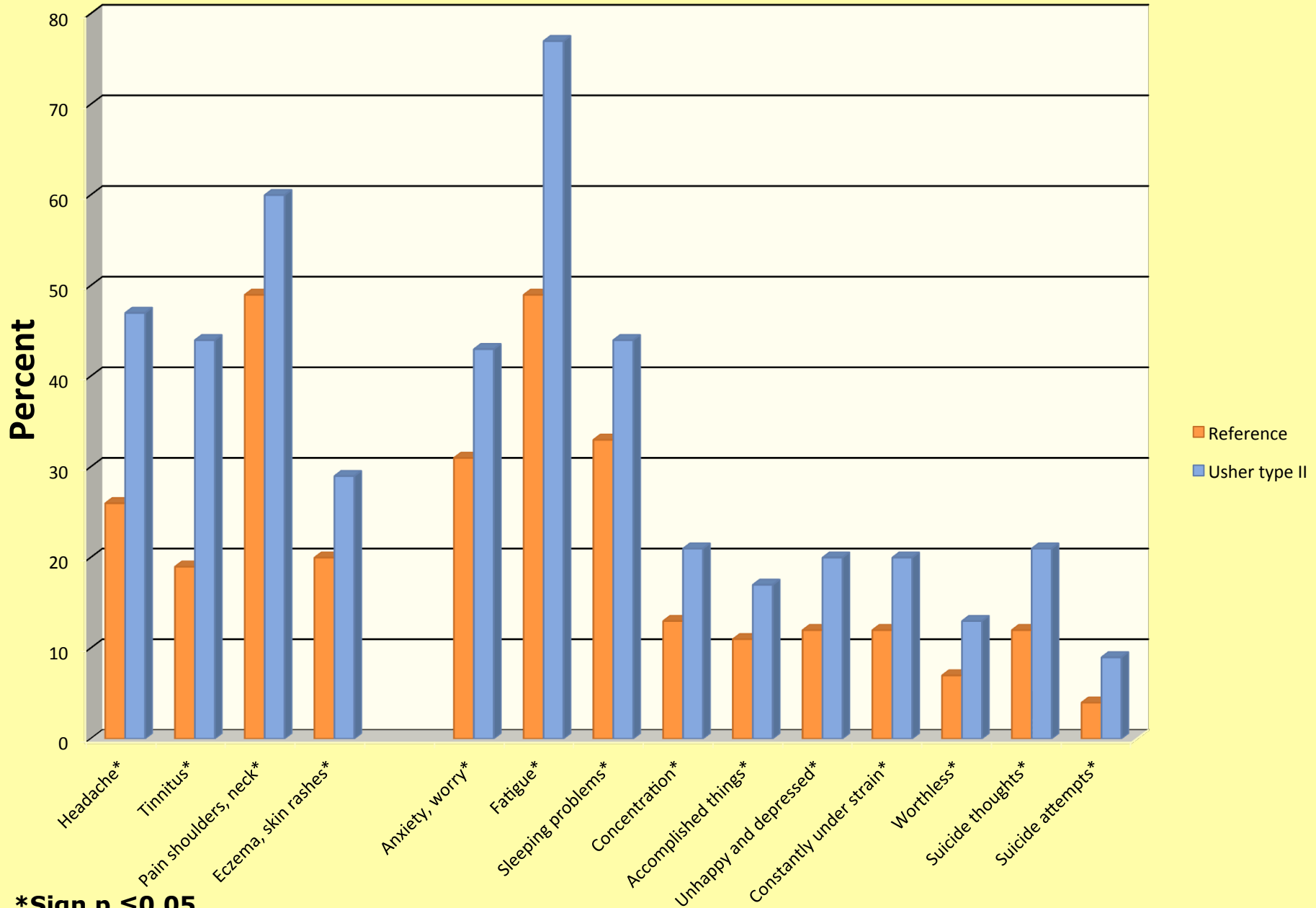
# Population

	USH2	Referens
N	96	5738
Age	55	58
Age range	18-84	23-91
Females(%)	53	56
Genetic diagnosis	59%	
Pure tone average	73db	
Visual field	4 ( $<10^{\circ}$ )	
Visual acuity	0,4	





# Physical and psychological bad health



\*Sign  $p \leq 0,05$

## 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

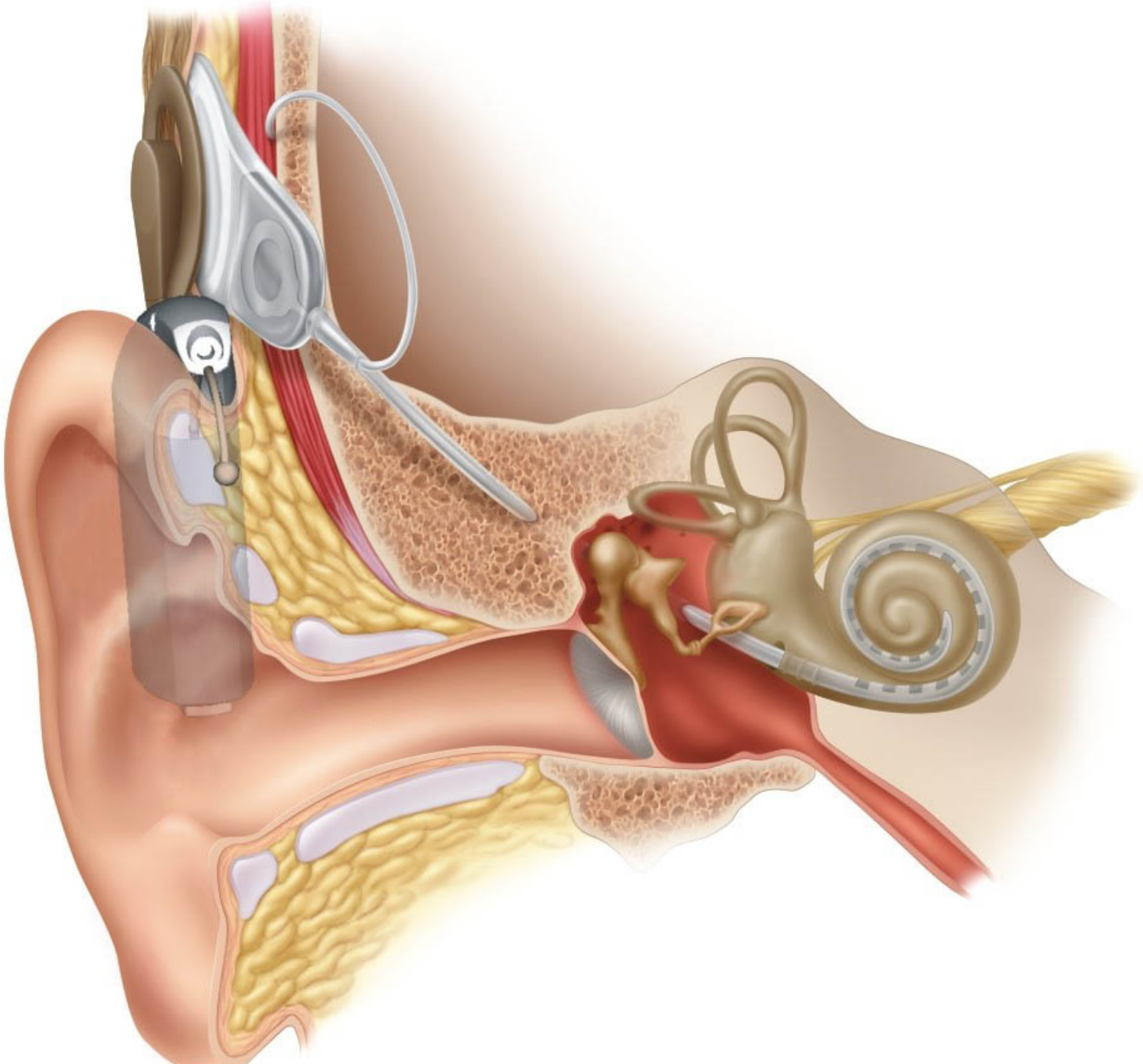
15 persons with USH3

Mean age 41 years

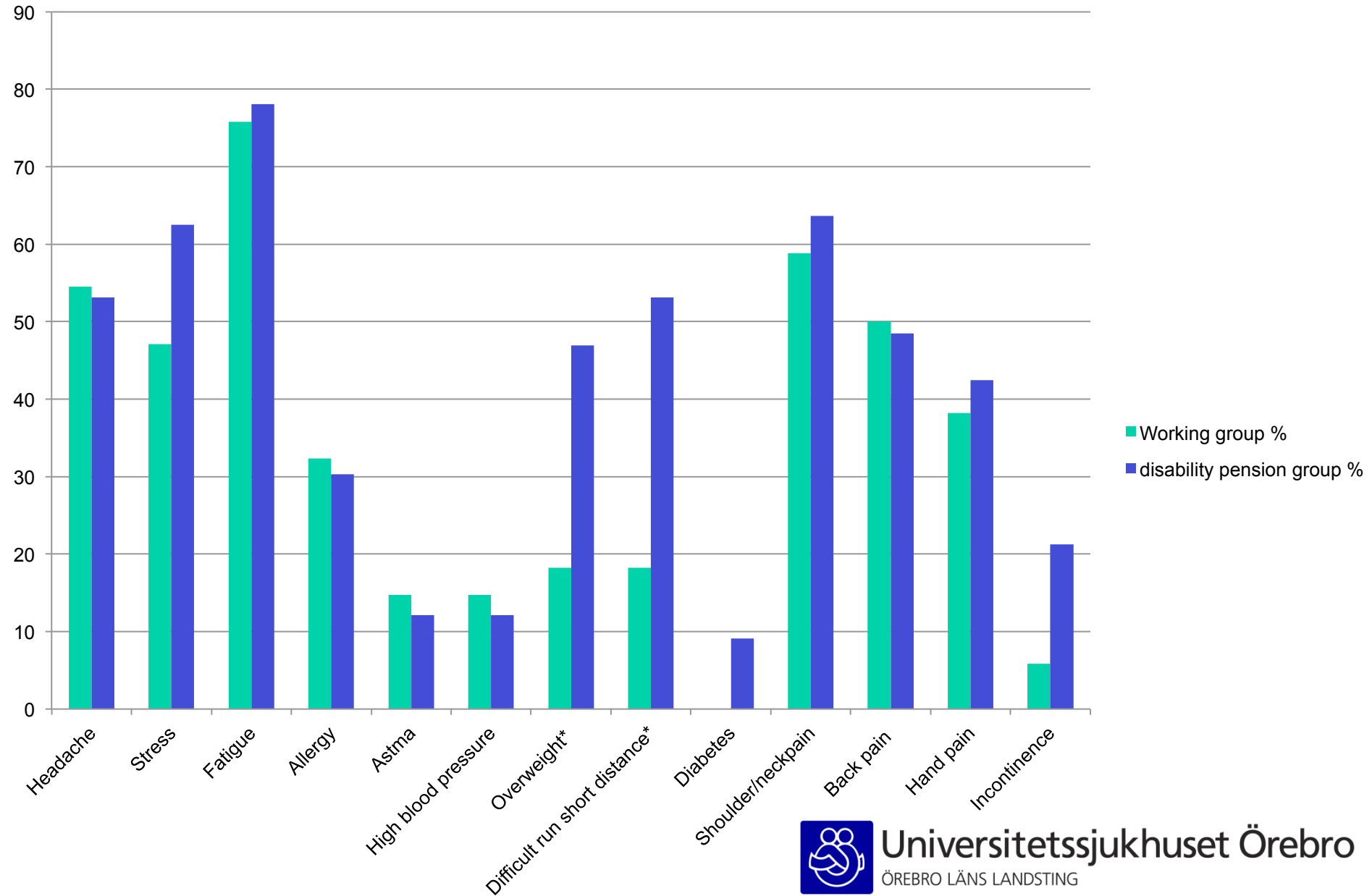
11 women and 4 men

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

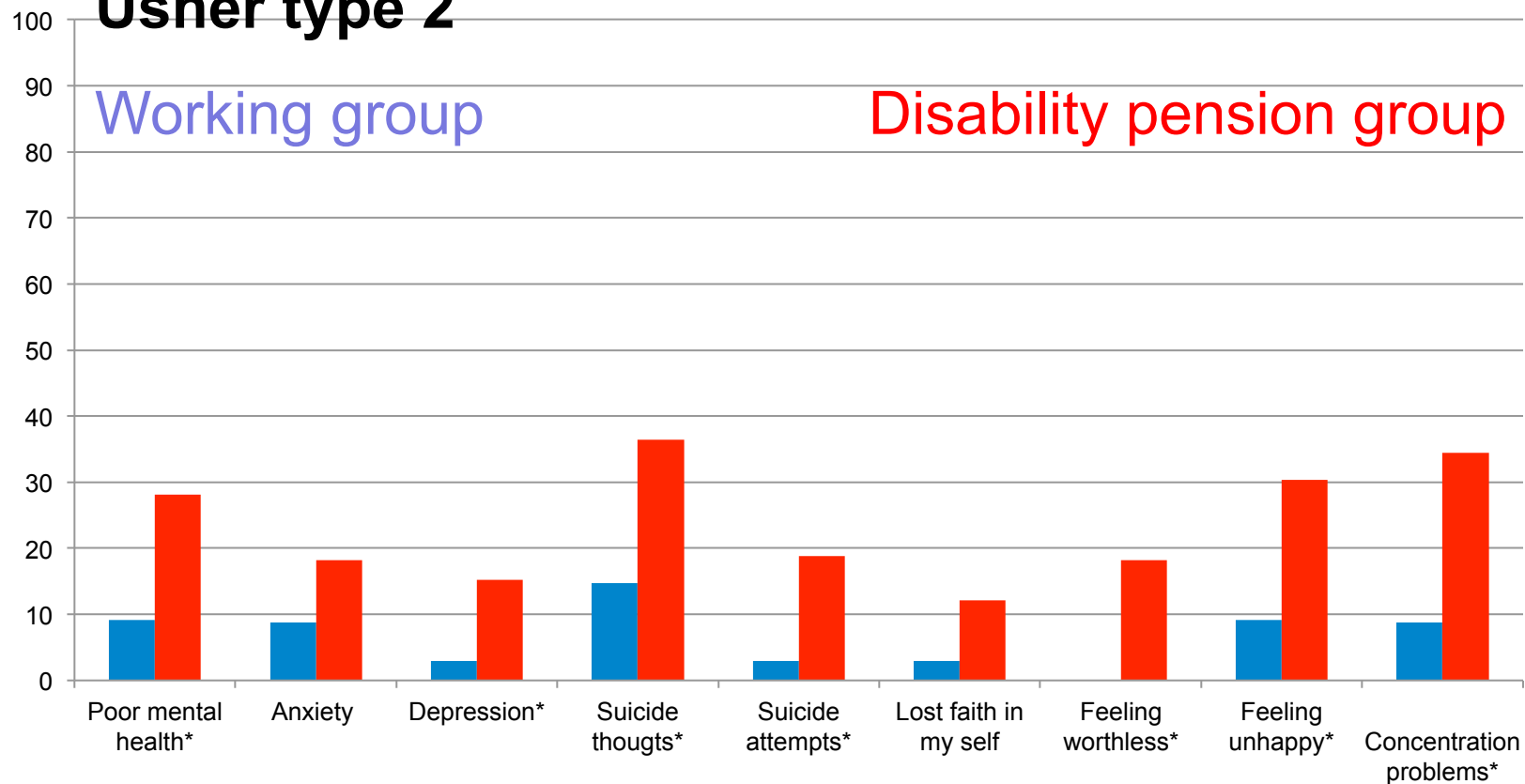




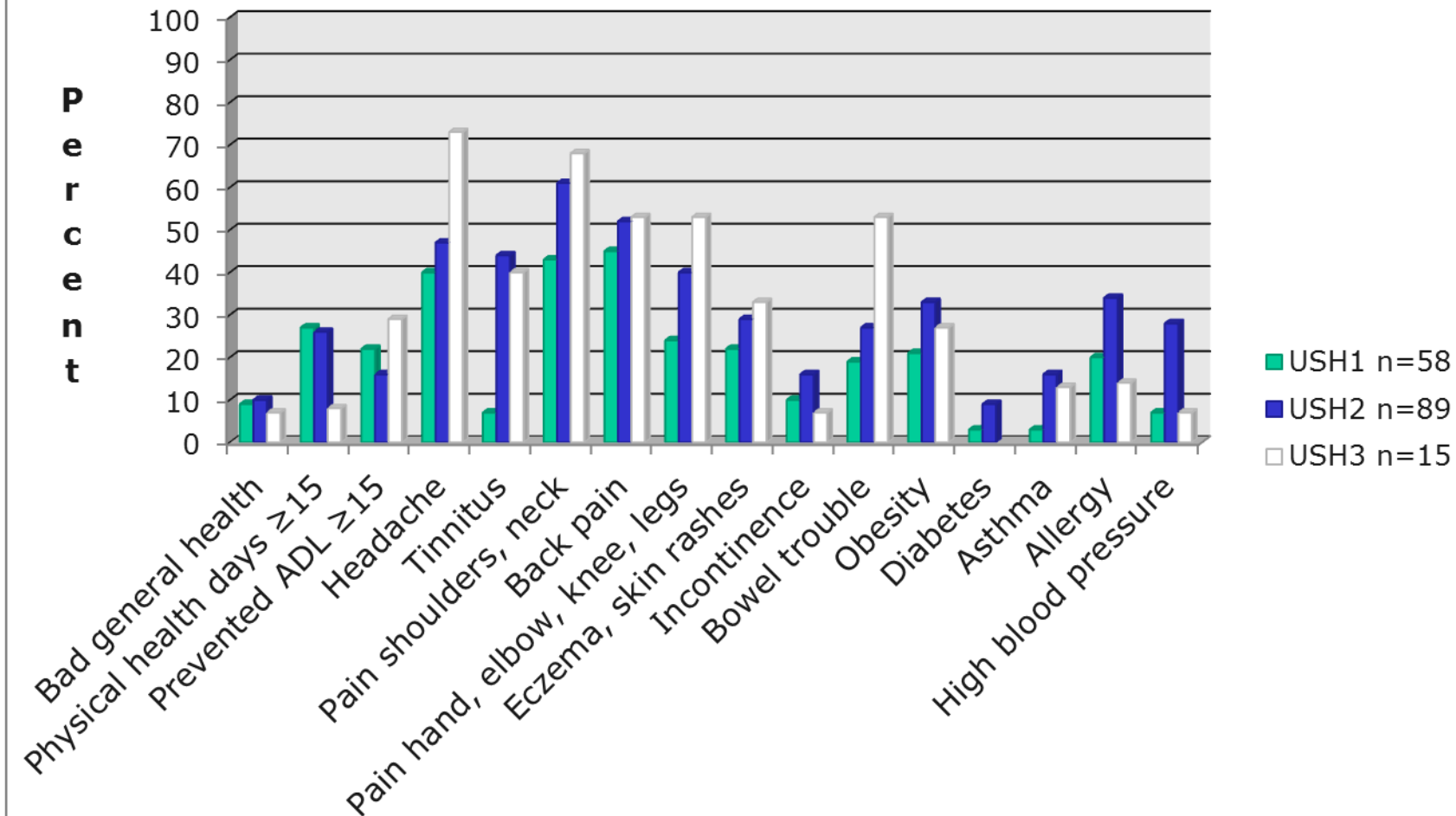




# Usher type 2

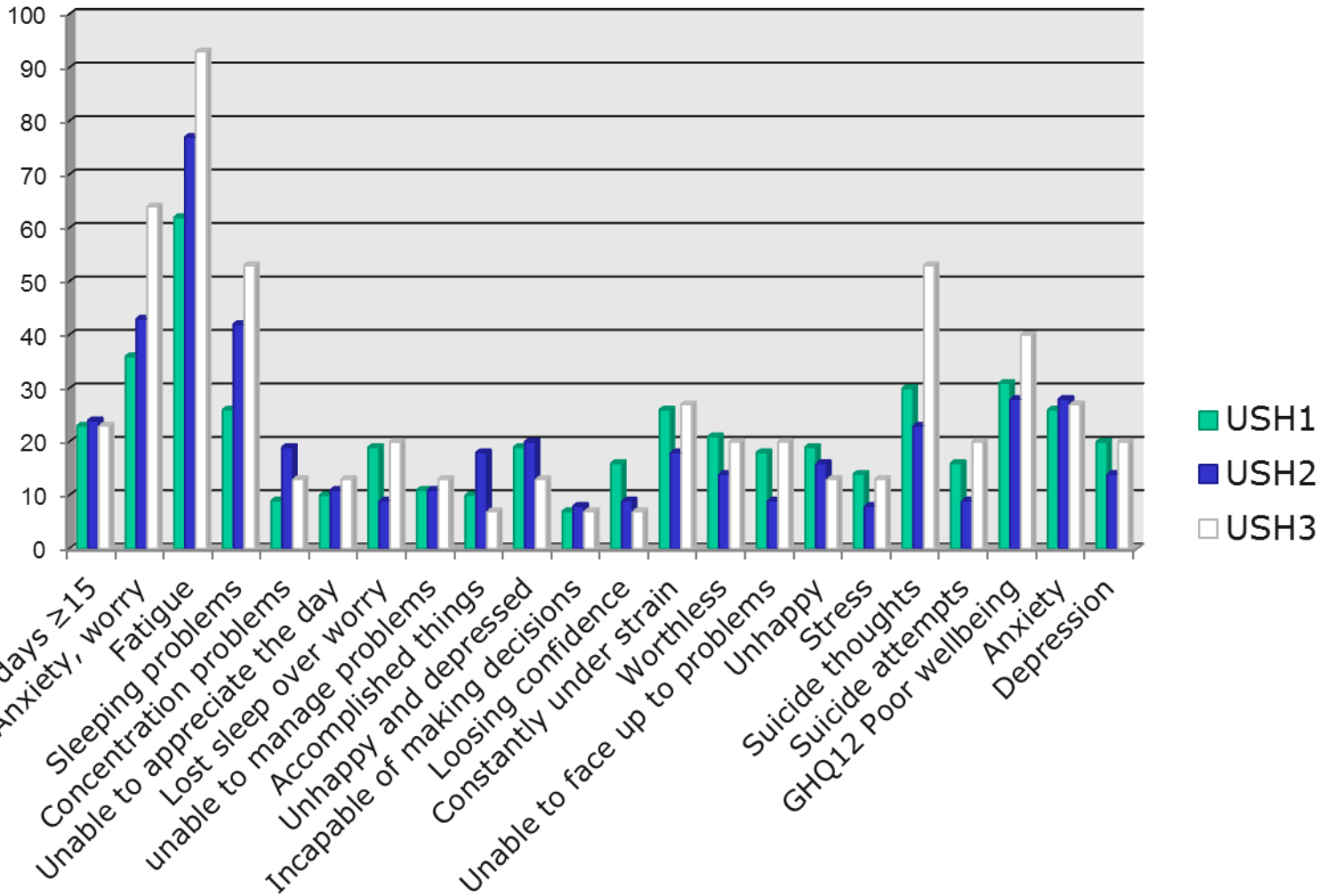


## Poor physical Health



## Psychological poor health

P  
e  
r  
c  
e  
n  
t



■ 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 !!!!

