

EUROPEAN SEMINAR OF THE ACQUIRED DEAFBLINDNESS NETWORK,
NOV 1-5, 2006

ABSTRACT

Usher syndrome - new findings concerning prevalence, diagnosis, habilitation, rehabilitation, treatment and eventually cure

Usher syndrome is a heterogeneous disorder with three clinical types.

- Type 1 Congenital profound deafness, congenital bilateral vestibular areflexia (balance problems) and early onset of retinitis pigmentosa.
- Type 2 Congenital moderate to severe hearing loss, normal vestibular function, retinitis pigmentosa as in type 1.
- Type 3 Progressive bilateral hearing loss, progressive vestibular dysfunction and retinitis pigmentosa.

At present ten different genetic forms are known. Some forms are much more common than others. The prevalence of Usher syndrome varies between different countries but is probably much larger than previously estimated. New epidemiological and genetic findings on Usher syndrome in humans and in animals will be presented.

These genetic findings have led to a better understanding on what goes on in the inner ear and in the eye. These findings and the possibilities of early diagnoses have led to better habilitation and rehabilitation. New treatment modalities such as cochlear implants, vitamins and antioxidants will be discussed. New animal research on other treatments and maybe even in the future a cure will also be presented. Usher syndrome is the most common disorder among people with deafblindness (50 per cent). The genetic research in Usher syndrome is at present performed all over the world. These findings will not only be important for people with Usher syndrome but in the long term for people with other eye-disorders and hearing loss. A baby born today, with Usher syndrome, in developed countries, will probably have a possibility of being cured.

Claes Möller, Professor
Department of audiology
Sahlgrenska University hospital
Gothenburg, Sweden