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Bulletin of the European network on GENetic DEAFness: pathogenic mechanisms, clinical and molecular diagnosis, social impact

Contents:

SECTION 1 - RESEARCH CORNER

MITOCHONDRIAL HEARING IMPAIRMENT

Howy Jacobs

Mitochondria are the power stations of our cells, the structures in which biological energy is generated. Amongst the components of our cells they are also unique, in that the coded genetic instructions needed to establish and maintain them are split between the chromosomes of the cell nucleus and a separate packet of genetic information, the mitochondrial DNA (mtDNA), that is kept inside them. There are hundreds of mitochondria in each cell, and hundreds or sometimes thousands of copies of mtDNA, all containing essentially the same genetic information.

Mitochondrial DNA

MtDNA is also unique in other ways: unlike the linear chromosomes of the cell nucleus, it is a circular molecule, of just 16,569 base-pairs (the 'letters' of the DNA code). This molecule contains a great deal of tightly packed genetic information, compared with the more 'leisurely' organization of the chromosomal DNA, where a single genetic function can be spread out over one million base-pairs or more. In addition, mtDNA is inherited in a completely different fashion from the chromosomal genes. Whereas we inherit one copy of each chromosome from our mother, and one from our father, mtDNA is passed down entirely from the mother. This maternal inheritance means that we can each trace back our maternal lineage through the generations in an unbroken and unambiguous chain, whereas the reshuffling of maternal and paternal chromosomes means that, as we go back through history, at each generation the number of our 'chromosomal ancestors' doubles. Thus, we each have 16 great-great grandparents, each of whom has contributed some of their chromosomal genes to us. But we each only have one 'mitochondrial great-great grandmother', our mother's mother's mother's mother, and our mitochondrial genetic information is essentially the same as hers was.

Mitochondrial disease

Because the genes of mtDNA are dedicated entirely to bioenergy functions, genetic errors (mutations) in mtDNA result in various kinds of defects in energy supply to cells. Although these are not fully understood, what is clear is that body's organs and tissues that are the most dependent on bioenergy: the brain, the heart, muscles, and the sensory organs of the eye and inner ear, are those most affected by such mutations. Collectively, we describe the diseases caused by mtDNA mutations as 'mitochondrial disease', although this term encompasses a vast range of conditions, ranging from very severe and invariably fatal conditions that strike in infancy, to mild disabilities that may become noticeable only in late middle-age. One of the goals of our research is to understand the biological processes that underlie this enormous variability in symptoms, sometimes associated the very same mutation in mtDNA. It's as if a small mistake in the blueprint for a jet engine sometimes caused nothing more serious than a very slight loss of acceleration, but sometimes led to a catastrophic failure or mid-air explosion.

Hearing impairment, as already indicated, is a frequent component of mitochondrial disease. However, here too we encounter enormous variability. Severe disorders such as the MERRRF or MELAS syndromes almost invariably include hearing impairment as one symptom, whereas other patients can experience only a mild, late-onset hearing loss, with or without other symptoms.

Perhaps the most famous example of this is the so-called MELAS mutation, at position 3243 of the 16,569 base-pairs of human mtDNA. This mutation is found in some patients who have the full MELAS syndrome, a severe neuromuscular disease that is accompanied by stroke-like seizures, and is unfortunately usually fatal by about age 20. The same mutation can be found in other patients, however, who seem only to have one or more of a mild hearing impairment, a relatively benign form of diabetes, or problems with moving their eyes, that can begin to manifest quite late in life.

Heteroplasmy

The 3243 mutation is an example of another unusual and complicating phenomenon connected with mitochondrial disease, namely heteroplasmy. As stated above, the hundreds or thousands of mtDNA molecules found in the cells of any individual are usually identical, a situation described as homoplasmy. However, in cases of mitochondrial disease, there is very often a mixture of normal and mutated mtDNA, which is described by the term heteroplasmy. The 3243 mutation is a good example, since the severity of the symptoms depends, at least to some extent, on the relative amounts of normal and mutated mtDNA in the affected tissues. Patients with the full MELAS syndrome usually have at least three times as much mutant as normal mtDNA, whereas a 50:50 mixture of the two usually leads to much milder symptoms, though with a still puzzling amount of variability.

Mitochondrial hearing impairment in the GENDEAF project

One of the groups of scientists working on the GENDEAF project is focused on trying to unravel some of the mysteries connected with mitochondrial disease mutations that commonly cause hearing impairment. We have three major aims: first, to document more precisely the contribution that mtDNA mutations make to hearing disorders in general; second, to establish so-called 'model systems' in which we can conduct clear-cut experiments to determine the effects on cellular processes of deafness-causing mutations of mtDNA, and third, to put this knowledge together to draw up guidelines for the recognition and clinical management of patients suffering from mtDNA-linked forms of hearing impairment.

In the rest of this article I shall outline some of our methods and achievements in pursuit of these three goals, and where they might lead us in future. Our long-term aim is obviously to prevent or perhaps even reverse the commonly observed long-term worsening of the condition of mitochondrial disease patients. However, we are still a long way away from these goals. We first need to establish the basic parameters of the problem: how commonly do mtDNA mutations cause hearing disorders in the European population, what cellular pathways in bioenergy production do they interfere with, and how does the organism try to respond to the stress created? Only with this knowledge can we hope to design workable therapies or preventative strategies, or even provide appropriate advice and counselling in the shorter term.

The epidemiology of mitochondrial hearing impairment

Our first aim has been to document how frequently mtDNA mutations are the cause of hearing impairment of different kinds. We already know that mtDNA mutations cause disorders like MELAS, that include many different types of symptom. However, we also know that many such families also have other members who have the same mutation at position 3243, but who suffer only from a progressive hearing impairment. We therefore set out to collect families where the *only* symptom is hearing impairment, which are obviously much more common than MELAS families.

What we wanted to find out was how commonly mtDNA mutations are the cause of deafness in such families. We collected families as well as so-called sporadic cases, where only one member of a family is affected (though more distant relatives may also be hearing impaired in such cases). In the UK and Italy we collected only those cases that are said to be postlingual, in other words, where hearing impairment is only detected after the child begins to speak. This is because the genetic causes of profound congenital deafness are already largely known, and mtDNA seems to contribute only to a few cases of this type of 'prelingual' deafness, at most just 1-2% of cases. In Spain we collected all cases, whether prelingual or postlingual, and in Finland we collected a completely different group of persons, suffering from age-related hearing loss, the type that afflicts people beginning only in late middle age.

The results of our screening have been highly informative. In different populations, mtDNA mutations seem to be the causative factor in at least 5-10% of cases of postlingual hearing impairment, although this does not apply to those with onset in late middle-age, which are not associated, as far as we can tell, with any of the previously recognized mutations in mtDNA. This makes mtDNA the second commonest genetic cause of hearing disorders, and indicates that it should be routinely screened for, even though at present no therapy can be offered, to delay or slow down the progressive worsening of the condition that we see in such cases.

Inheritance of mitochondrial hearing impairment

In all such families, the inheritance of the condition is exactly as expected for a mitochondrial gene mutation, in other words it is passed maternally, along with the mtDNA, from mother to child, but never from father to child. Men and women are equally affected by mitochondrial hearing impairment, but only women can pass it on. However, in some cases, the causative mutation seems to have arisen anew in a single individual, or the offspring of one woman who herself may be unaffected. This is mainly due to a new mutation: an error during the process of copying mtDNA in the egg cell that grew into a single individual, or in a single cell that divided many times to generate all the egg cells of one woman.

Another explanation that seems sometimes to apply, at least in some cases, is that of heteroplasmy. Some members of a family all of whose members have a mixture of normal and mutated mtDNA, have just too little of the mutated form to cause any detectable problem. But those family members who carry a more substantial amount of mutated mtDNA can manifest the disorder which it causes. It seems that only a few copies of the mtDNA are randomly assigned to each egg cell, which then are copied many times over to produce the final number of copies found in the whole egg cell, which is over 100,000. But this 'random assignment' process leads to the different children of one woman who is herself heteroplasmic having very variable amounts of the mutated mtDNA in their own tissues. Sometimes, this amount can become so high that a severe disease like MELAS results, but in the majority of cases, hearing loss is the only symptom that is found.

To understand this process, imagine that a mother gives her children five sweets (candies) drawn blindfold from a bag containing a mixture of equal numbers of red and green ones. Assuming there are a large number of sweets in the bag, so that those given to the first child don't significantly affect the selection available for the next, we can easily understand that whilst most children will get 3 of one colour and 2 of the other, some will receive four red sweets and only one green, or *vice versa*, and sometimes, even though it will occur only seldom, all five sweets drawn from the bag will be of the same colour, just by chance. The child with five red sweets would be equivalent to the rare cases of MELAS, the ones with 3 or 4 red sweets would have only hearing impairment, and the children getting mainly green sweets would be unaffected. Although this concept over-

simplifies the situation a little, it is actually quite a good model for how heteroplasmy evolves over the generations.

The 1555 mutation and aminoglycoside antibiotics

One particular mutation, at position 1555 of the mitochondrial DNA, seems to be present at a hugely variable frequency amongst patients in different countries. In most countries it is relatively rare, being seen at about the same frequency as the 4-5 other most commonly detected mtDNA mutations associated with hearing disorders. In Spain and some other countries, however, it is found much more commonly amongst the hard of hearing. Puzzlingly, it also shows an apparently complex and unpredictable inheritance pattern, even though it is not one of the mutations that commonly shows heteroplasmy, nor does it seem to occur afresh at an unusually high frequency

A clue as to what is going on is provided by the fact that the mutation has been known for some time to predispose to severe and rapid hearing loss in patients treated with a specific class of antibiotics, called aminoglycosides, which includes streptomycin, gentamucin and neomycin[1]. These drugs are sometimes prescribed to treat a variety of common infections. One possibility, therefore, is that the mutation is really at a similar frequency in different populations, but that it is antibiotic use that is extremely variable. In countries where this particular class of antibiotics are, or have in the past been very widely used, there would be expected to be a much greater occurrence of severe hearing loss associated with the combination of the mutation and the drug. In non drug-treated individuals the mutation may produce only a very mild hearing loss that is not usually brought to the attention of clinicians. Therefore, in countries where this type of antibiotic has not been widely used, the frequency of the mutation amongst the hearing impaired would be low.

In order to test this idea we are analysing random individuals from the population in different countries (Spain, Italy, Finland and elsewhere) to see if, indeed, the 1555 mutation is at a similar frequency in different countries. If so, the antibiotic hypothesis would be strongly supported, and we would need to investigate this issue in more detail.

Testing and counselling for mtDNA mutations

Mitochondrial hearing impairment, although superficially less 'severe' than other forms, presents unique features that make it particularly important to be diagnosed. For a start, the patient must be made aware that other symptoms could develop. For example, carriers of the 3243 mutation should be regularly tested for diabetes. In addition almost all cases show progressive worsening of the hearing loss with age, which means that the patient should be regularly tested by an audiologist, and must plan for the possibility of a more severe disability in the future. Carriers of the 1555 mutation should be made aware that they are at significant risk of a rapid worsening of their hearing loss if treated with aminoglycoside antibiotics, and any doctors treating them must be made aware of this as well, so that this group of drugs can be avoided. Finally, carriers of mtDNA mutations need to be aware of the implications for their actual or potential children. Affected males will never pass on the condition to their children, but affected females will always do so. Heteroplasmy complicates the issue, however. As already explained, a heteroplasmic mother will usually have heteroplasmic children, but the relative amount of mtDNA they inherit can be highly variable. A child might be much less severely affected or alternatively the same or much more severely affected than the mother. Prenatal or even preimplantation diagnosis (requiring *in vitro* fertilization) can be offered in such cases, but there is never a 100% guarantee that the child will be completely healthy.

In the long term, we obviously hope that our use of model systems will allow us to understand more about how mtDNA mutations cause hearing impairment and other disorders. This, hopefully, will lead us in the direction of an effective treatment for a conditions that currently can only be managed, e.g. by fitting of a hearing aid, not actually prevented or cured.

I hope, in a future issue of the GENDEAF bulletin, to present some of our findings from model systems, such as fruit flies, in more detail.

If you have been prescribed antibiotics by your doctor do not stop taking them. Should you have any concerns, please contact your doctor.

SECTION 2 - LATEST NEWS

PSYCHOSOCIAL ASPECTS OF GENETIC HEARING IMPAIRMENT REPORT FROM WP6

Dafydd Stephens, Lesley Jones

This reports covers most of the work achieved in the first half of the GENDEAF programme, which was to review the literature in the field. It is now being produced as a book, and the present account is an edited version of the introduction/preface of the book, entitled "Genetic Hearing Impairment – Its Impact" It will be published by Whurr publishers, London, in the latter half of 2004.

This book sets out to examine the social and psychological effects of genetic deafness, hearing impairment and the genetic interventions associated with them. There have been enormous developments in this area over the last twenty years, and this book reflects some of those changes in the lives of deaf people and those who live and work with them. Scientific progress in the study of genetic deafness has meant that ethical, political and economic dilemmas have entered the field of what was once seen as an area of objective, value-free endeavour. These issues cannot be avoided and we seek to address them in a straightforward way by making use of different perspectives within the book: medical, psychological and sociological as well as personal and political.

The book arises from the European Union GENDEAF Thematic Network project, and is a product of the Psychosocial Working Group of that Project, which attempts to bridge the gap between those concerned with clinical and molecular genetics and the service users organisations.

Although the study and practice of medical genetics and of the genetics of hearing impairment might be seen as essentially socially neutral topics of scientific endeavour, the actual effects have by no means been neutral. This applies particularly to the way in which the findings are used and applied by clinicians, teachers, politicians and other professionals involved with people with hearing impairments.

Such misuse of the scientific findings related to genetic hearing impairment goes back a long way, and one of its greatest perpetrators was a key figure in the field of education of the Deaf, Alexander Graham Bell. Despite having a deaf wife himself, he very much espoused the Eugenic concepts of Galton, aimed at eliminating such "defects" from the population. This was carried to extremes

under National Socialism in Germany in the 1930s with initially sterilisation and later “elimination” of individuals with congenital deafness.

It is not surprising; therefore, that many Deaf people are suspicious of developments in of genetic hearing impairment which, they feel could, via prenatal diagnosis and subsequent termination of pregnancy or by gene therapy, lead to the elimination of people with congenital deafness. This is reinforced by the statements of some clinicians within genetics, otology and audiology categorically proposing the elimination of genetic deafness.

The concepts held both by Deaf people and by some professionals are false in that only some fifty percent of congenital hearing impairment is genetically determined. Furthermore, over 36 different genes, mutations of which can result in non-syndromal hearing impairment, have so far been identified and many more localised. Approximately half of these are generally associated wit recessive inheritance. In addition, the entrenched views of some professions ignores the social impact of human rights legislation, the growth of the political disability movement and the increasing recognition and status of the Deaf community and of National Sign Languages. Within this book, we try to maintain a balance between the medical and social models of disability by not focussing on the impairment as “the problem” but by taking into account the impact of the barriers to communication and access created by others. Obviously each author has their own perspective and the readers must make up their own minds. We simply present informed views within the rapidly changing field of genetic interventions in hearing impairment in the hope of providing some insight into improving policy and practice in an important area.

Our research in this area has also highlighted the dearth of knowledge and understanding about the impact of having genetic or familial hearing impairment on individuals and their families. As previously mentioned, only a very small proportion of genetic hearing impairment is congenital, accounting for some fifty percent of congenital hearing impairment. This amounts to less than one per thousand of the population. However some 15 – 20% of the population have a significant hearing impairment, mainly of late onset, and recent twin studies indicate that at approximately 50% of that is also genetically determined. Furthermore, while a little is known about the impact of having a family history of hearing impairment in children, there is a complete dearth of knowledge of such an impact in adults. This book was intended originally to be an overview of the psychosocial effects of genetic hearing impairment in particular, as opposed to hearing impairment in general. However, because of the absence of information available specifically on that, the four key chapters on the impact of genetic hearing impairment in children, working age adults, elderly people and the deafblind have had to concentrate on the impact of such conditions in general, while highlighting any specific evidence of the impact of genetic conditions.

In addition to these four key areas, we have also included a consideration of otosclerosis, one of the few genetic causes of hearing impairment which is amenable to surgical treatment, and of Neurofibromatosis 2, which is a condition causing hearing impairment but which is also a potentially lethal condition.

Finally we included two autobiographical chapters outlining the experiences and impact of having a genetic hearing impairment and genetic deafblindness on the lives of the authors.

The book begins with two chapters on responses to genetic deafness and the growth of genetic intervention. The first, by Lesley Jones, a social scientist who has worked for many years in the field of Deaf studies, examined the social aspects of genetics and deafness and particularly the response of the Deaf Community, highlighting the historical and political basis of their attitudes.

Anna Middleton, a genetic counsellor, then reviewed her own and other studies on the attitudes of Deaf and hearing parents towards genetic interventions, including new data from her own studies. These indicate a limited demand for prenatal diagnosis, with those seeking it, regarding it as a way to help them cope with and adjust to having a deaf child.

We then included two methodological chapters providing the structure for the overviews on the impact of the hearing impairment and related conditions covered in the main part of the book. In the first of these Dafydd Stephens, an Audiological Physician, and Berth Danermark, a Sociologist, present the relevant parts of the World Health Organization's "International Classification of Functioning, Disability and Health" (ICF) which we have adopted as the framework for examining the impact of genetic hearing impairment. This model attempts to bridge the medical and social models of disability, so seemed particularly appropriate to our approach.

The second, by Berth Danermark, Sophia Kramer, a Psychologist, and Dafydd Stephens, outlined the general methodological approach adopted in the three review chapters on the impact of hearing impairment in general, and of genetic hearing impairment in particular. This was important to ensure that all key work was highlighted, many such studies often not being accesses by standard databases. Bibliographies based on these chapters are found on the GENDEAF website under the heading "Psychosocial group members". While these were initially password-protected, any visitor to the website can now access them.

In the first of these chapters, Dafydd Stephens examines the impact of hearing impairment on children, particularly those with moderate to profound congenital disorders, who have been the most extensively studied. There is also, however, increasing evidence of a significant impact of mild and unilateral hearing impairments. While there is little information on the impact of genetic deafness per se, there is a body of literature indicating that the children of deaf parents, probably mainly with autosomal dominant inheritance, experience the impairment differently, with less impact on their self esteem than the deaf children of hearing parents.

Berth Danermark then provided an extensive overview of the effect of hearing impairment on working age adults, hitherto a very neglected area, and is able to identify only very limited information on the impact of a family history in this respect. The impact on their work situation is particularly important in this context.

Similarly, Sophia Kramer, drawing together the very diverse results on the effects of hearing impairment in elderly people, was unable to identify any studies on the impact of such a family history. She did, however, highlight many methodological weaknesses of existing studies, emphasising the need for a more systematic approach if we are to have a realistic understanding of the problems experienced by elderly people with hearing impairments.

Kerstin Möller, a health services researcher, reviewed the situation with regards to deafblindness, an area bedevilled with definitional problems. These stem, in part, from the variability of both the hearing and the visual impairments, their age of onset and their progression. This applies in particular, to the genetic condition Usher syndrome of sensorineural hearing impairment and retinitis pigmentosa, which recent genetic studies have shown covers a range of mutations in different genes, with very variable phenotypes. However, despite this she has admirably drawn the literature together and highlighted the deficits in our knowledge.

Nele Lemkens, an academic otologist, looked at the impact of Otosclerosis, and found that patient expectations are very much conditioned by the possibility of surgery in the condition. However, despite this being a common condition, there has been little work on the impact of otosclerosis on

those with the condition, probably stemming from the fact that most surgeons have little interest beyond the functional results of their intervention.

Wanda Neary, a Paediatric Audiological Physician, addressed the effects of Neurofibromatosis 2 (NF2). This has multi-system effects, resulting from tumours in different parts of the central nervous system, including those affecting both cochlear nerves. Surprisingly the psychosocial impact of the condition has been almost completely neglected and the chapter has had to concentrate on the background of the condition and the impact of related disorders.

From here, we move on to two important autobiographical studies by individuals with hearing impairment who have themselves become professionally involved in the field. The first, by Jill Jones, a Social Worker and researcher, gives a personal perspective on genetic deafness caused by Treacher Collins syndrome. The second, by Patricia Lago-Avery, a Counsellor, deals with the development of different components of Usher 2 syndrome and her experience of having a cochlear implant. Valuable insights are gained from these two accounts which could inform and improve practice.

The book concludes with a glossary, which all authors have attempted to stick to throughout their overviews, deviating only when quoting the work of others.
We hope this overview will be useful in stimulating more focussed work in this field. Already, most of the authors have been conducting specific studies to fill some of the gaps, but much more needs to be done by all interested researchers and clinicians. We are convinced of the value of multidisciplinary work in this area in order to address the issues raised by the rapid changes taking place in the experience, theory and practice of genetic hearing impairment. Research in the field cannot be performed in isolation from the very real psychosocial effects on the daily lives of those people with genetic hearing impairments if it is to adequately inform the policy and practice of the relevant professionals.

We hope that this book will go some way towards doing that by drawing together some important elements of the relevant work.

SECTION 3 - ASSOCIATION CORNER

INHERITANCE OF USHER SYNDROME

Christina Fasser

Together with the diagnosis people with Usher Syndrome (or their parents) learn that they suffer from a genetic condition. Immediately a number of questions arise. The first is always a simple, but evident one: My parents and nobody in my family have similar symptoms or are known to suffer from Usher syndrome. How can it be that this is a genetic disease? Siblings of an affected person may ask themselves if they would in future develop the disease or what is the risk for their own children. The most common questions are:

How is Usher Syndrome inherited?

All forms of Usher syndrome are inherited the same way, which is called “auto-somal-recessive”. Every human being has two copies of each gene. One gene is inherited from the father, one from the mother. In order to get the disease a person needs to get two copies of the gene with the mutation causing Usher Syndrome.

What is a carrier?
If somebody receives only one gene copy with the mutation for Usher syndrome and a second, healthy copy than that person is a so-called carrier and presents no signs of Usher Syndrome.

My parents and nobody else in the family has Usher Syndrome. Why can it be inherited?
Carriers of Usher Syndrome gene present no symptoms of the disease. They only learn that they are carriers when one or several of their children are born with Usher Syndrome.

I have Usher Syndrome. Will my children have Usher Syndrome? A person with Usher Syndrome will give a copy of the gene with the mutation to all his or her children. However, if the other parent has no
mutated gene for Usher Syndrome, the children will be carriers and will not develop the disease.

My brother or sister has Usher Syndrome. I have no symptoms. Will my own children have Usher Syndrome?
An unaffected brother or sister of a person with Usher Syndrome has a 50 % chance to be a carrier of the disease. If he or she is a carrier, they will give with a chance of 50 % the mutated gene to their children. These children will be carriers and present no symptoms of Usher Syndrome, provided that the other partner is not a carrier of the Usher Syndrome gene.
What happens to my children if my partner has also Usher Syndrome?
Theoretically all the children of these parents will have Usher Syndrome. Since there are a number of different mutations in different genes causing Usher Syndrome, to receive a precise answer to this question, these couples should consult with a geneticist.

Is genetic testing available?
A number of mutations in different genes are known to cause different forms of Usher Syndrome. At present not all gene mutations causing Usher Syndrome are known. Therefore, genetic testing at present is only possible for cases with a known mutation.

Where can I get genetic counselling?
The basis for good genetic counselling is a well established diagnosis by your ophthalmologist and ortho-rhingologist. They will refer you to the appropriate genetic counsellor.

When do I need genetic counselling?
There are different moments in the life when people may wish to get genetic counselling:
- If a child with Usher Syndrome were born to a family and the parents would like to have more children.
- If an adolescent person with Usher Syndrome wishes to know more about the genetics of his or her form.
- If a person with usher Syndrome wishes to get married and wants to know with his or her partner about the risk for their children to have Usher Syndrome.
- If a sibling of a person with Usher Syndrome wants to know his or her risk, that their children may get Usher Syndrome.

Important:
Nobody can be forced to get genetic counselling or be told not to have children. This is always the choice of the parents to be. However, a genetic counsellor can clarify the inheritance pattern and present the risks and probabilities, thus helping to make an informed decision

SECTION 4 - MISCELLANEOUS

DEAFBLINDNESS

Claes Moller

From workgroup 3 (Ushers Syndrome group)

I went to the doctor and he told me that I will go deaf and blind. He doesn't know when, but it might be in the near future. Then the doctor abruptly left the room. No! Not my hearing, not my vision! It is not fair! How could God do this to me? Why wasn't I told until I was grown up? I have had vision and hearing problems for as long as I can remember and no one told me. This is not fair. Somebody help me!"

Deafblindness are a heterogeneous hearing and vision disorders, which can be caused by trauma, diseases, or inherited syndromes. To be fast and reliable, communication between human beings relies on vision and hearing. These two senses are complementary and enhance each other. A deafblind person can be profoundly deaf and completely blind, completely deaf with visual impairment, completely blind with hearing problems, or have hearing and vision dysfunction. Since vision and hearing interact, deafblindness means that in this instance, one plus one equals three.

Congenital deafblindness (blindness and profound deafness) is extremely rare. Causes of congenital deafblindness include genetic problems, premature birth, and infections. Children with congenital deafblindness very often have other problems such as mental retardation or cerebral palsy. Many different genetic syndromes are known and genes and mutations have been identified for some.

The hallmark is extreme difficulty in communication and relies heavily on tactile sign language and input via the remaining senses. In congenital deafblindness very promising achievements have been made, especially by using cochlear implants.

Acquired defablindness is a very heterogeneous group of disorders, which affects many more people than does the congenital disability. There are at least 50 different syndromes that cause acquired deafblindness. Most of them are rare. The most common cause of aqired defablindness is Usher syndrome, which accounts for almost half of all people with deafblindness. Usher syndrome is an autosomal recessive disorder and in most countries, the prevalence is 8–10 per 100 000 newborn babies.

Usher syndrome is clinically divided into three main types, with several genes implicated in different types of the syndrome.

Type 1. Congenital profound deafness, balance problems due to vestibular areflexia and retinitis pigmentosa (RP), (a disease which gradually destroys the retina causing night blindness and tunnel vision) In type 1, up till today seven different genes have been localised. One of the most common types is Usher type 1b, which has a gene mutation called myosin VIIA. This gene is expressed in the hair cells of the cochlea and in retinal photoreceptors.
Type 2. Congenital moderate to severe hearing loss, normal balance function and RP as in type 1. In Usher type 2, mutations are found in a gene called usherin (type 2a). This gene is also expressed in the cochlea and in the retina but it seems to cause disturbances in supporting cells instead of the hair cells.
Type 3. Congenital moderate hearing losses, normal balance function during childhood, which deteriorates and RP as in type 1 and type 2. Type 3 is quite common in Finland but can be found in other countries as well. The gene for type 3 has been localised as well.

Collaboration between researchers within Gendeaf and Boys Town National Research Hospital, Omaha, USA, has yielded many new insights to Usher syndrome, and has resulted in advances that will allow early and accurate diagnosis. Hopefully, in the very near future, improved understanding might develop new treatment modalities for Usher syndrome. Other syndromes that might cause deafblindness include CHARGE association, Alström syndrome, Alport syndrome as well as many other rare syndromes. Due to activities within the Gendeaf consortium many deafblind syndromes have been clinically characterised and genetically localised.

Identification of the precise mutation of genetic disorders might, in the future, lead to medical and genetic treatment. In addition, progressive loss of hearing and vision might be preventable with drugs such as antioxidants (e.g., vitaim A, vitamin E or vitamin C) or growth factors. Another possibility is stem-cell treatment.

One goal in habilitation, rehabilitation and treatment of deafblindness is to reduce isolation. New knowledge in other fields can today reduce the effects of impairment associated with deafblindness. Computers and the Internet, such as the Bulletin for example, have reduced the isolation of people with deafblindness. New software developments have also enhanced communication through Braille, speech synthesisers, and Magnivision among others. Many networks exist already both within the European community and in other parts of the world that connect deafblind people.

The new insights in genetics, combined with more advanced diagnostic tools for assessment of vision and hearing, make early and correct diagnosis possible. These advances are important, since an early diagnosis will result in accurate prognosis. Informing a patient about the aetiology and the prognosis of a disease will reduce fear and misunderstanding, create realistic expectations, better rehabilitation and, hopefully in the future, treatment.

“Nothing is impossible. The impossible just take a little bit longer time”. Helen Keller.

USHER SYNDROME CONTACTS

For more information, conctact the organisation in your country:

Clinics with rehabilitation/treatment of persons with Usher Syndrome

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Deafblind organisations engaged in Usher Syndrome

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Switzerland	Christina Fasser The International Retinitis Pigmentosa Association Ausstellungsstrasse 36 CH-8005 Zurich Switzerland cfasser@e-link.ch www.retina.ch	SZB Beratungsstelle fur Hörsehbehinderte und Taubblinde St. Galler-Ring 79 SE- 4055 Basel www.szb.ch
UK	Mary Guest The National Deafblind & Rubella Association Usher Research Sense 11-13 Clifton Terrace, Finsbury Park N4 3 SRLondon UK mguest@sense.org.uk www.sense.org.uk	

Rehabilitation centres working with Usher Syndrome

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Norway	Roar Meland Eikholt Senter for døvblinde Helen Kellersvei 3 Drammen N-3031 Norway roar.meland@eikholt.no www.eikholt.no Wenche Andersen Regionsenteret for døvblinde, Skådalen Pb13, Slemdal N-0710 Oslo Norway wenche.andersen@statped.no	Sigmund Spetalen Huseby kompetansesenter Gamle Hovsetervei 3 Oslo 768 Norway sigmund.spetalen@statped.no

Sweden	Claes Nilsson Resurscenter Mo Gård 612 93 Finspång Sweden mogard@mogard.se www.mogard.se Gun-Britt Milioris Nationellt experteam för diagnostisering av dövblinda Resurscenter Mo Gård Nora Strand 24 182 38 Danderyd Sweden expertteamet@mogard.se	Lena Hammarstedt Nationellt kunskapscenter för dövblindfrågor Box 570 182 15 Danderyd Sweden lena.hammarstedt@mogard.se
Switzerland	SZB Beratungsstelle für Hörsehbehinderte und Taubblinde St. Galler-Ring 79 Basel 4055 Switzerland www.szb.ch	

Genetic centres working with Usher Syndrome in humans

Denmark	Thomas Rosenberg National Eye Clinic for the Visually Impair 1 Rymarksvej DK-2900 Hellerup Denmark roseeye@visaid.dk www.visaid.dk	
Netherlands	C. W. R. J Cremers University Medical Center Hospital Nijmegen Department of Otorhinolaryngology/EnT dept/UMCA St Radboud P O Box 9101 6500 HB Nijmegen Netherlands c.cremers@kno.umcn.nl www.humangenetics.nl	F P M Kremer University Medical Center Hospital Nijmegen Department Human Genetics P O Box 9101 Nijmegen 6500 HB Netherlands h.kremer@antrg.umcn.nl www.humangenetics.nl
Norway	Öivind Nilssen University hospital of Tromsö Department of Medical GeneticsTromsö Norway oivind.nilssen@unn.no www.unn.no/medisinsk-genetisk/index.html	

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Sweden	Claes Möller Sahlgrenska Universitetssjukhuset Center of Genetic Hearing Losses Department of Audiology and Genetics/SweGene 413 45 Göteborg Sweden claes.moller@audiology.gu.se	

Centres working with Usher Syndrome

Denmark	Thomas Rosenberg National Eye Clinic for the Visually Impaired 1 Rymarksvej DK-2900 Hellerup Denmark roseeye@visaid.dk	Henrik Ribber The Information Center for Acquired Deafblindness Generatorvej 2 A, DK-2730 Herlev Denmark hri@dbcent.dk www.dbcent
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Database - Patient association network

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Database – Clinical Research

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Database - Basic research human

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SECTION 5 - LETTERS

DEAFBLINDNESS, THE UNIQUE DISABILITY

Dear Patrizia,
I am very pleased to be able to tell you that deafblindness was recognised as a 'unique disability' on 1st April (http://www.europarl.eu.int) The campaign to achieve this was launched at the meeting held at the European Parliament on 6 January, 2004. 80 people representing the Edbn (European Deafblind Network), the DbI (an international association for deafblind people) and other national institutions for deafblind people, were present as well as some members of the European Parliament. We have been successful and I would like to thank the Lega del Filo d'Oro, which, like other institutions working for or with the deafblind supported this initiative, and my thanks also to those working on the GENDEAF project because through their website we were able to launch an appeal on internet too.

William Green
DbI President

Dear William,
This is an important achievement for the deafblind who could benefit from a law recognising that they have a 'unique disability'. Not only will the deafblind themselves benefit but also their families and all the services who work with them now and in the future.
The appeal launched by the Lega del Filo d'Oro and the appeal on the Gendeaf Project website, were taken up by my colleagues and partners and by others whom I would like to thank for their valuable contribution to the successful outcome of this campaign.

Patrizia Ceccarani
Education and Rehabilitation Director, Lega del Filo d'Oro

SECTION 6 - MEETINGS AND EVENTS

International Symposium on Hearing Disorders in Early Childhood etiology, screening, diagnostic and rehabilitation, Prague (CZ), May 6-8 2004. For more information: http://www.orl.lf2.cuni.cz/symposium - E-mail: contour@volny.cz

The International Conference on Newborn Hearing Screening, Diagnosis and Intervention, Villa Erba, Cernobbio (Como Lake), Italy May 27 – 29

2nd European Family Conference "Listen to me 2 – in Denmark", Slettestrand, Northern Jutland. DK, 20 – 26 June 2004

7° World Congress IFHOH, Helsinky, Finland 4-9 July 2004. For more information: http://www.ifhoh-helsinki2004.org

9th International Conference on Computers Helping People with Special Needs, Universitè Pierre et Marie Curie, Paris, France 7 – 9 July 2004. For more information: www.icchp.org

SECTION 7 - NEW OPPORTUNITIES TO COLLABORATE IN THE GENDEAF PROJECT

Can you help us?

As part of the GENDEAF programme we are interested in the effects that having a family history or genetic cause of hearing loss has had on you or other members of your family. The present literature in the field is very limited and we are keen to discover how people themselves think about this. In order to help us in our work we would like to hear from as many viewers of this website as possible.

Do you yourself or any of your children have a hearing impairment due to genetic causes or have other people in your family with hearing loss? if YES **Please read on.**

1. If **you yourself** as well as other members of your family (parents, brother and sisters, children) have hearing problems please let us know of any ways that having other people in your family with hearing problems has affected **your** reaction to **your own** hearing problems.

Please e-mail a list of these problems to us at StephensD@cf.ac.uk writing down as many effects as you can think of. Please let us know whether you would be agreeable to answering other questions about hearing problems and their consequences.

NB. In the subject label of your e-mail please insert the word GENDEAF

Thank you very much for your help.

2a. Does your child or children have a **genetic** hearing impairment or deafness ? ☐ YES ☐ NO

b. Does/do they have any additional handicaps? ☐ YES ☐ NO

c. If YES, what other handicaps do they have?

d. Are you hard of hearing or Deaf yourself? ☐ YES ☐ NO

e. Did or does the fact that your child or children's hearing impairment was due to **genetic causes** affect you in any way? ☐ YES ☐ NO

- if **YES**, in which way or ways.

Please list as many effects as you can think of and e-mail them to us at StephensD@cf.ac.uk putting in the subject label the GENDEAF
Thank you very much for your help.