



EDITORIAL

Michael Morgan

EURO-ATAXIA has a new President, Dr. Michel Koenig. He was unanimously elected at the 10th Annual General Meeting of EURO-ATAXIA, recently held in Turku, Finland, between 25 and 27 September, 1998. Inside Dagmar Kroebel gives us an abridgment of the Minutes from the business meeting on Sunday 27.

The main business was of course the science. Ewout Brunt gave a detailed account of new Medical and Scientific Developments in Dominant Ataxias, while Michel Koenig likewise gave an overall assessment of the latest Medical and Scientific Developments in Recessive Ataxias, in particular Friedreich's ataxia.

Among European countries Finland has a unique genetic disease heritage. About 30 inherited diseases are overrepresented in the Finnish population, with some of them being even totally absent in other parts of the world. A typical representative is Infantile Onset Spinocerebellar Ataxia, IOSCA, an ataxia so far found only in Finland. Inside Kaisu Nikali gives an overview of research into IOSCA, in addition explaining exactly how the concentration of rare hereditary diseases in Finland came about.

But on the other hand, some inherited disorders common elsewhere are not encountered among the Finns at all, and some are encountered only very rarely. Päivi Mustonen is one of only seven people with Friedreich's ataxia in Finland. A practising journalist, she has written of her own experiences inside, in *Living With Early Onset Ataxia*.

From a slightly different perspective, Seppo Hirvonen has written of life with Adult-Onset Ataxia, in *Adjustment*. We also include an evocative piece by Seppo, summing up as it does the Finnish love of nature. Seppo Hirvonen: *In The Woods*.

One of the strong points about the Finnish way with disability is the attention they give to the psycho-social side of life. Tarja Ketola is Clinical Neuropsychologist at the Masku Neurological Rehabilitation Centre, near Turku, which has organised adjustment training for people with neuromuscular disabilities for many years. Inside she writes specifically on ataxia, in *Psycho-Educational Activities For People With Ataxia*.

As well EURO-ATAXIA moves on. Plans are already underway for the 11th AGM next year. This will probably be held in Westende in Belgium over the weekend of 29-31 October 1999 and as it will be the last one to be held this Century it's definitely not to be missed. Be there or be forever square...

Finally we include a pen-portrait of EURO-ATAXIA board member for Germany, Peter Reussner. Peter, who hails from Hamburg, has entitled his introductory article, *Living And Working With Ataxia*.

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THE 10TH ANNUAL GENERAL MEETING OF EURO-ATAXIA, TURKU, FINLAND, 25 TO 27 SEPTEMBER, 1998 REPORT OF THE SECRETARY-GENERAL

Dagmar Kroebel

I start with a tribute. The death of Evelina Raveggi was a loss for us all. She was the visible sign that EURO-ATAXIA is a European wide federation. I miss her presence and her encouragement.

shall empower disabled people themselves to reach their full potential in society. And so to business. First, news of a change of office-holders within Euro Ataxia. Ewout Brunt has re-



An impression of the Saturday afternoon session

A year has past and we have the opportunity to meet again in Turku, Finland. EURO-ATAXIA together with its members clearly proves, that by exchanging experience and information, many things can be achieved. We must go on working even if continually we are confronted with various problems which hinder a full life in society for ataxic people. The principle of full participation and equality that inspired the 1981 Year of Disabled People has evolved towards the concept of citizenship in 1998. This evolution constitutes a major stage as it stresses the participation of disabled people as active agents in making social change. The federation's objective is to encourage the independent living of disabled people. This approach has focused on all spheres of life: mobility, culture, education, leisure activities, access, tourism, employment, etc., are all included. In this way we

linquished his position as President of EURO-ATAXIA, and is instead Vice-President. By unanimous decision Michel Koenig took over as President for 1999.

The year 1998 has also been devoted to the setting up of partnerships on concrete actions between EURO-ATAXIA and member NGO's (Non Governmental Organizations) of the European Disability Forum; on Sector mobility impairment, FIMITIC and Parking Facilities in Europe.

The AGM of the European Disability Forum (EDF) took place in Brussels on March 20-21. Its members are NGO's formed by disabled people in the European Union. The EDF has to work together with the European Commission on all aims which deal with disabled people.

The congress of the German Society of Human Genetics and the

Austrian Society of Human Genetics took place in Jena, Germany, between March 25 and 28, 1998. EURO-ATAXIA was there with a stand. This year the French Society of Human Genetics has joined up with the Societies, and will take part in next years AGM in Nuremberg, Germany, March 24-27, 1999.

The AGM of the European Alliance of Genetic Support Groups (EAGS) took place in Lisbon, May 10-12, 1998. The 11th conference was scheduled on: Community Genetic Information: Rules, Responsibilities, Power and Possibilities for Patients and their Families. (For further details on this meeting please contact me, Dagmar, in Brussels).

In May we were contacted by the European Federation of Neurological Societies (EFNS) who wish to set up a Liaison Committee between themselves and patient groups. The first meeting was scheduled for September 23rd in Seville, Spain. We were represented there by Dr. José Berciano Blanco. Vote permitting, we will become members of EFNS for 3 years (at a cost of USD 160) and will strive to establish a Special Interest Group within it on hereditary ataxias.

The European Commission has recently set up a European Action Programme on Rare Diseases. This will give:

- Information On Rare Diseases
- Support To Patient Organisations
- Dealing With Clusters Of Rare Diseases.

However the budget was cut down considerably. Suggested advice is to approach your national ministers of health within your own country to pursue a larger budget at EU level for The Action Program For Rare Disorders – which is after all an initiative of the Commission.

The European Action Programme on rare Diseases

Patient groups have an important role to play in the development of legislation and the heightening of public awareness on the relevant issues. Only sufferers of rare diseases and their families can communicate what their problems and concerns are – and they should be

listened to. As every organization's dynamism depends on its ability to listen to others, our dynamism depends on us and on our ability to communicate.

And finally, deadlines for the newsletter must be respected!!!

MEDICAL AND SCIENTIFIC DEVELOPMENTS IN DOMINANT ATAXIAS

Ewout Brunt

Up to now 8 types of dominantly inherited ataxia have been genetically defined: Spinocerebellar Ataxia type 1-7 (SCA1-7) and Dentato-Rubral Pallido-Luysian Atrophy (DRPLA). Recent years have seen an impressive genetic contribution. The first gene for hereditary ataxia, SCA1, was discovered in 1993, and the gene of SCA7 was identified in 1996. Genetic mutations of SCA1, 2, 3, 6 and 7, have now been identified, and for SCA4 and SCA5 the responsible genes have been localised, waiting for identification. For the SCA's with a known genetic mutation, individual genetic diagnosis and presymptomatic testing is possible.



Thus far, all mutations for the different SCA's and for DRPLA are comparable, and consist of an unstable expansion of a CAG repeat in one of the gene's exons (reading frames). 'CAG' stands for cytosine, adenine and guanine, the names of separate DNA molecules, which form a 'triplet'. The genetic information in a gene consists of hundreds of these triplets. Each triplet codes for a single unit (amino acid) of the protein molecule, which results from the transcription of the DNA in the gene. Dominant hereditary ataxia's are not the only disorders caused by an expanded triplet repeat. Among the disorders caused by an expanded triplet repeat are Huntington's disease, Friedreich's ataxia and Myotonic dystrophy.

In the SCA's, DRPLA, and in some other inherited diseases like Huntington's disease, a normally present CAG triplet repeat is expanded and has become unstable. Translated into a protein, this expanded CAG triplet repeat results in an expanded tract of identical glutamine amino acids (a polyglutamine tract). Once the gene has been identified, the gene-products, proteins, can be studied. With one exception, SCA6, the proteins are previously unknown, novel proteins, with unknown functions. For SCA, the novel proteins are named 'ataxin' with the corresponding SCA number, e.g. ataxin1. Similarly for DRPLA the protein is called

'atrophin', for Huntington's disease the protein is called 'huntingtin' and for Friedreich's ataxia, the protein is called 'frataxin'.

SCA6 is the only Spinocerebellar Ataxia in which the function of the gene and protein is known. The protein in SCA6 is part of a voltage gated calcium channel in the cellular membrane, which plays a role in the cellular excitation. Other mutations in the gene for SCA6 can cause two other (allelic) disorders: hemiplegic migraine and episodic ataxia type II.

Combining the genetic and clinical information in the different Spinocerebellar Ataxia's, some correlations become apparent. Within groups of SCA1, SCA2, SCA3, and SCA7 patients, the length of the CAG repeat shows a strong inverse correlation with the age at onset of symptoms, and also with progression of the disease. So, people with a later onset of symptoms usually have a smaller expansion of the CAG repeat. However, at any repeat length there is a rather wide variation in age at onset, of some twenty years or so. Thus, in a single individual the repeat length does not enable prediction of the age at onset or progression. Although essentially similar during an individual's lifetime, the CAG repeat is not stable during parent to child transmission, and may thus be different in parents and children. Especially in paternal transmissions, the expansion often increases. This in part explains anticipation, the phenomenon that the mean age at onset is earlier in next generations. In maternal transmission the repeat expansion is usually more stable.

Clinical manifestations vary considerably within each genetically defined type, and with exception of SCA7 and SCA6, only few global characteristics may be discerned for the SCA's, precluding an individual clinical diagnosis.

Research on histological and pathological aspects has focused mainly on the mechanism which causes cell death and ataxia. First the presence of ataxin has been studied. As SCA's are dominantly inherited disorders, both a normal and a mutated SCA gene are present, and so both normal and mutated ataxin are expressed. It must be realised that genes are not active all the time, and that genes are turned on or off by signals according to cellular activity and specialisation. In the case of SCA1 and SCA2 for instance, it appears that ataxin1 and ataxin2 are present in many tissues and cells, and that the levels of ataxin do not correlate well with the presence of localised neurological cell death.

To explain the discrepancy between the presence of (mutated) ataxin and cell degeneration, interaction with other, possibly cell specific proteins has been suggested, and indeed, in Huntington's disease and various SCA's, aggregation with proteins has been demonstrated. Also, a tendency has been demonstrated for mutated ataxins to (self)aggregate, and proteins containing an expanded polyglutamine stretch have been found to be toxic for cultured cells.

Last year it was reported that in Huntington's disease and in SCA3 or Machado-Joseph Disease, certain brain cells showed inclusion bodies within the cell's nucleus. These inclusion bodies contained partially degraded molecules of mutated huntingtin and ataxin. Normally, huntingtin and ataxin are present in the cytoplasm of these cells, that is the fluid surrounding the nucleus, but not within the nucleus. The presence of intranuclear inclusion bodies seemed to correlate with degeneration of these neurons. As these inclusion bodies also contained a protein degrading molecule, named ubiquitin, these inclusion bodies seem to indicate that the protein aggregate which they contain is resistant to degradation.

As for this year's progress, one of the outstanding scientific achievements is the development of a transgenic fruit-fly (*Drosophila*) model for SCA3/MJD. Following the development of transgenic mice for SCA1,

SCA3/MJD and Huntington's disease, changes may be studied over weeks to months, but in the *Drosophila* model pathologic mechanisms can be studied within a relative short time span of days. As in humans, the *Drosophila* model demonstrates that normal and mutated ataxin3 are widely expressed, but cell degeneration is rather localised, and involves eye retina cells. In the transgenic *Drosophila*, retina cells develop normally and intranuclear inclusions build up before the cells degenerate. The cells subsequently die through apoptosis. Apoptosis is the cell's suicide programme, which destroys the DNA, and results in shrinkage of the cell. In the stage that intranuclear inclusions are present, prevention of apoptosis can delay but not prevent cell death, indicating irreversible cell damage as a cause for apoptosis.

To summarise, following genetic identification and correlation with clinical manifestation, study of the expression of ataxin and its role in cellular changes has substantially increased our understanding of these neurodegenerative disorders. All of this has been made possible by the development of animal models. We are privileged to live in an exciting time of rapid scientific progress. Hopefully this progress will soon enable us to help those affected by inherited disorders like Spinocerebellar Ataxia.

MEDICAL AND SCIENTIFIC DEVELOPMENTS IN RECESSIVE ATAXIAS

Michel Koenig

Friedreich's ataxia is the most frequent recessive ataxia occurring among white populations. But there are other recessive ataxias as well. They include: Ataxia-Telangiectasia. This is an early onset ataxia with dilated veinules and telangiectasia. AT as it is known is almost as frequent as Friedreich's ataxia.

Familial Vitamin E Deficiency – of which there are two forms, with or without fat malabsorption. The form without fat malabsorption is more common in the North African population, but rare everywhere else. Vitamin E supplementation is an effective treatment. Refsum Disease (ataxia with neuropathy and retinitis).

Infantile Onset Spinocerebellar Ataxia, or IOSCA, a Finnish disease unknown in other world populations. Spastic Ataxia of Charlevoix-Saguenay. This is found mostly in Quebec, Canada.

Other rare ataxias that don't have a name because they are not well characterised.

In recessively inherited ataxias only the ataxia patient manifests the disease because he/she has 2 copies of the ataxia defect or mutation, one received from each parent. The parents of the patient are not affected because they carry only one copy. For all recessive dis-

eases, the frequency of healthy carriers is much higher than the frequency of patients, because the risk that 2 carriers of the same ataxia mutation marry is relatively low. For example, for Friedreich's ataxia, the frequency of healthy carriers is 1 in 120 while the frequency of patients is 1 in 40,000. We have shown that almost all Friedreich's ataxia carriers and patients have the same ancestor (who lived most likely more than 10,000 years ago). This explains why Friedreich's ataxia is more prevalent in white populations and is almost non-existent among Japanese and black African populations.

The mutations lie in genes, which serve to make proteins, the building blocks of all living matter. The identification of the Friedreich's ataxia gene showed that the mutation is the same in almost every patient (in agreement with the fact that they are the descendants of a same ancestor). It is an expansion of a repetition of the trinucleotide GAA. Unlike the trinucleotide expansions found in dominant ataxias, the GAA expansion is very large and does not contain the information to make the protein (called frataxin). How then does this expansion cause the disease? The expansion causes much less frataxin to be made, about 10 to 20 times less. The important point here is that there is still a little bit of frataxin made. This distinguishes the expansion mutation from other mutations, referred to as truncating mutations and frequently found in other recessive diseases, which completely

alter the production of the protein. The identification of the Friedreich's ataxia gene now allows the study of the function of frataxin, which should shed light on how its reduction causes the disease. Frataxin is a protein of the mitochondria, the structures of a cell where its fuel is produced from food. This energy conversion requires electron transport with iron containing proteins. Frataxin is present in tissues that are rich in mitochondria, which include the tissues that are affected in patients (dorsal root ganglia of the spinal cord, heart ...). Frataxin is present in every living organism, including fungi and yeast, because they also contain mitochondria. In yeast, complete absence of frataxin causes iron accumulation within the mitochondria. In Friedreich's ataxia patients, a similar defect is also likely to apply, but to a lesser extent since there is still some residual frataxin present. Evidence for this are iron deposits and defect of some iron containing proteins in heart of patients. Mitochondrial iron accumulation seems therefore to be the culprit, since excess of iron is well known to stimulate the production of toxic compounds called free radicals or reactive oxygen species. The toxic process is called oxidative stress. Some drugs and vitamins, called anti-oxidants, protect against oxidative stress. One of them is vitamin E, therefore suggesting that reduction of frataxin or of vitamin E may cause ataxia through a final common mechanism. The use of anti-oxidants appears attractive but should be approached with caution, since some anti-oxidants may have an effect opposite to the one intended. The use of animal models is vitally important here, in order to assess the effectiveness of different therapeutic approaches. A 'knock-out' mouse has been specially bred, its DNA altered to inhibit frataxin production and so mimic Friedreich's ataxia. The difficulty here is to create a mutation in the mouse that would have the same

consequence as the expansion mutation in patients: a reduction but not a complete extinction of frataxin expression. As suggested by Daniela Iser, an alternative therapeutic approach would be to prevent the expansion mutation from reducing frataxin production. The faulty process here seems to

lie in the reading of the gene (elongation of transcription), and more precisely the reading of the expansion of the GAA repeat, as shown by the groups of M. Pandolfo (Montreal) and P. Patel (Houston). More work is warranted on this aspect of the disease.

INFANTILE ONSET SPINOCEREBELLAR ATAXIA

Kaisu Nikali

About 30 inherited diseases are overrepresented in the Finnish population, with some of them being even totally absent in other parts of the world. These diseases are mostly recessively inherited, meaning that both gene copies need to be defective in order for the disease to present itself. The concentration of rare hereditary diseases in Finland can be explained by the founder effect resulting from a small number of original settlers, genetic drift and long-term regional isolation. The original settlers of Finland, just hundreds or thousands in number, migrated slowly and periodically within a relatively large country. This almost unique population movement led to the formation of regional isolates and several subpopulations. Also, geographical and linguistic reasons meant that the entire country remained isolated, with practically no immigration from the neighbouring areas. Even migration between internal subpopulations was limited, resulting in an uneven distribution of genes. This regional clustering of heterozygous disease gene carriers and consanguineous marriages have led to the enrichment of genetic disorders of the people's own. On the other hand, some inherited disorders common elsewhere are not encountered among the Finns at all, since the respective disease genes were either lacking or lost from the gene collection of the original inhabitants. Due to the homogeneity of the Finnish gene pool, most of the

Finnish diseases can be expected to be caused by a single major mutation, a feature offering great advantages in genetic research. Indeed, the passionate genetic studies of recent years have resulted first in the assignment of several Finnish disease loci and even further in the isolation of these genes and identification of the mutations.

A typical representative of the diseases found so far only in Finland is autosomal recessively inherited infantile onset spinocerebellar ataxia, IOSCA. It was encountered for the first time in two pairs of siblings at the Hospital for Children and Adolescents, University of Helsinki, in the early 1970's. To date, 21 IOSCA diagnoses have been made, the patients originating from 15 distinct nuclear families. The gene frequency of this disorder among the 5 million Finnish population can thus be estimated to lie around 10⁻³. The very first clinical symptoms of IOSCA – acute or subacute ataxia, hypotonia, atetosis and loss of deep tendon reflexes – manifest themselves during the second year of life in previously healthy infants. They are followed by ophthalmoplegia and deafness, developing by school age, and sensory neuropathy, optic atrophy, and female primary hypogonadism, in teenage years. Epileptic attacks, which are unresponsive to or even exacerbated by antiepileptic drugs, are a late manifestation and can eventually

be fatal. The typical central nervous system (CNS) pathology in IOSCA is a continuum of progressive degeneration of the spinal cord, brain stem, cerebellum and its connections, and at a late stage, the cerebral cortex. In addition to the degeneration in CNS, an early-onset, rapidly progressive sensory axonal neuropathy with a severe loss of mainly large myelinated fibres presents a prominent neuropathological finding. Among other hereditary ataxias, therefore, IOSCA most closely resembles Friedreich's ataxia.

Neither biochemical defects nor chromosomal rearrangements are observed in IOSCA patients, the etiology of the syndrome thus remaining unknown. Positional cloning is a strategy used to clone a hereditary disease gene solely on the basis of the location of the gene in the genome. It was thus the method of choice for the search for the gene underlying IOSCA. In positional cloning, the primary requirement is a sufficient collection of samples from families carrying the inherited disease in question. In essence, positional cloning then consists of assigning the chromosomal location of the disease gene, restricting this locus as well as possible, constructing a physical map of the locus, identifying and analyzing positional candidate genes, and finally characterizing the mutation(s).

In the course of the molecular genetic study, IOSCA was first shown by linkage analysis not to share its disease locus with any known hereditary ataxia, and thus to be distinct from other inherited ataxias. Subsequently, the search for shared chromosomal regions with homozygosity, using samples from just four affected individuals, resulted in the assignment of the IOSCA locus to the long arm of chromosome 10. Studying extended disease haplotypes, ordered collections of marker alleles reflecting the ge-

netic constitution of the chromosome, revealed old genetic recombinations, greatly restricting the critical IOSCA region. The haplotypes also suggested that all recent IOSCA chromosomes do originate from a single founder chromosome. Haplotype and so-called linkage disequilibrium analyses combined with the genealogical study, tracing IOSCA ancestors back to the 1500's, suggested that this founder chromosome was introduced into the Finnish population some 30-40 generations ago and spread over the country during the major east-west migration of the people from the county of Savo. The genomic clones covering the IOSCA locus have also now been isolated and the physical map of the region constructed, resulting in the length estimation of a 160 kilobase-pair IOSCA region. Three interesting genes could be localized close to this critical IOSCA region. Of these, a 66 kD neurofilament is a novel form of mammalian neurofilaments putatively involved in early axonal transformation, CYP17 presents a cytochrome with a major role in steroid biosynthesis, and paired-box protein 2 (PAX2) participates in the development of the CNS and neural crest derivatives as a transcrip-

tion factor. However, none of these genes proved to be defective in IOSCA patients, although the association of PAX2 with IOSCA cannot be conclusively excluded yet.

The IOSCA gene still remains unknown, within a region containing some five genes on average. However, it has now been genetically shown to represent a distinctive disease entity, its gene locus on 10q24 has been characterized in detail, and optimal tools for the eventual identification of the disease gene have been created. The results enable genetic counselling, as well as patient, carrier and prenatal diagnosis for the families possessing affected offspring. The future isolation of the IOSCA gene and the identification of the mutation(s) are a prerequisite for the precise DNA diagnostics for the disease and will enable studies for developing gene therapy. Moreover, the unravelling of the molecular defect in IOSCA will be a further step towards the complete understanding of the pathogenic mechanism underlying this neurodegenerative disease and will give new insight into the molecules essential for the normal function of the nervous system.

LIVING WITH EARLY ONSET ATAXIA

Päivi Mustonen

I was diagnosed at the age of seven and my clumsiness was labelled as Friedreich's ataxia. Knowledge of the disease didn't change my life at all, that of my parents perhaps. Friedreich's ataxia is such a rare diagnosis in Finland – only seven people have been diagnosed with it – that I have almost been able to consider it as a personal characteristic, as an inseparable part of my person. As my ataxia progresses and I become unable to do things that I used to be able to do, for example to write by hand, reach out to a

light switch and actually touch it, I have to work towards developing alternative ways of functioning. Applications made by other disabled people are a great help. In my early youth I didn't know any disabled people other than paraplegics, and that's why I can't move or do other things like ataxic persons, but do them in the same way as paraplegics. In my opinion, there should be far more disabled people in public life, because public figures serve as role models and work on public opinion, making it more tolerant.



The further my speech difficulties progress, the more time and trouble I have to take to convince people of my credibility. Unclear speech is associated with unclear ability to think – at least in Finland. Ataxia affects breathing, but at this stage I have still attempted to make use of breaks in speech and slow down my speaking as a way of varying my speech. The sharp 's' and rolling 'r' in the Finnish language sometimes seem impossible to produce, but long vowels and diphthongs are naturally easy for me.

Nowadays I work as a journalist, sometimes lazily and at other times even more lazily. The Finnish welfare state takes care of my basic livelihood, and thus it is all the same to society whether I work or not. To me, not working and being excluded from society sounds like a punishment. Like most other people with a progressive disease, I too cling to everything to do with life with all my strength. I'm taking lowest grade degrees in university in everything interesting such as philosophy and history. I can only progress slowly in my studies, but my own ambitions are the only limit to my studies. Gaining new knowledge is a thrilling feeling, but sometimes I wonder whether I'm involved with the university more because of the social environment.

I work on a regular basis for the

magazine published by the disabled citizens' organisation Kynyns. Sometimes it feels strange to say that I'm working because I would be writing the same articles even if I was not paid for it. On the other hand, I get enough hours of help from an assistant and the required computer hardware on the grounds that I need them for my work or vocational studies, which I would not be entitled to only on the basis of all-round education or having writing as a hobby.

The basic foundations of my life consist of the Social Insurance Institution (SII), the municipal social administration, the municipal health centre and the hospital district. All manual aids that are not fixed on the ceiling or walls, for example, a manual wheelchair, tongs for gripping, specially made shoes and all aids for eating and cooking, I get from the health centre. The Social Insurance Institution provides me with computer hardware, financial support for vocational studies and support for maintaining physical fitness, such as physiotherapy. I'm entitled to two one-hour sessions of physiotherapy per week and in addition to a two-week institutional rehabilitation session twice a year paid by the SII. The SII also takes care of my basic livelihood in that if my medical expenses exceed 2000 Finnish marks, the SII pays for the remainder. The hospital district acquires all the electrical and motorised aids for my use, such as my electric wheelchairs (one for indoors and one for outdoors) and my motorised bed.

It is the duty of the municipality to ensure that all its residents have appropriate accommodation. Thus, I too have accommodation in which I can get assistance if necessary. This so-called service housing is paid for by the municipality.

I need additional assistance to be able to work and study. I apply for

the assistance and for fixed assistive devices mounted on the ceiling and walls – such as handles and grab rails – from the municipal social services on the basis of the Act on Services for the Disabled. The Act comprises two parts. First, there are subjective rights, such as home modification work and service housing. Second, there are services tied to appropriations, such as assistants' wages. In Finland, the local authority is obliged to take into account future needs of residents when drawing up the budget, and thus if I feel that my application has been rejected on incorrect grounds, I can appeal to the provincial administrative court for amendment.

I have applied for the expenses for hiring a personal assistant for 100 hours per month from my local authority. The local authority reimburses me for the wage expenses for 20 hours per week, that is, 80 hours per month. I am relatively happy with the amount. When I pay my assistants' wages, withholding taxes to the tax office and employer's social security contributions and statutory insurance premiums to the insurance company, I collect all the sums together and charge them the act on services.



ADJUSTMENT

Seppo Hirvonen

I have had many years of good health. For about forty years I was allowed to lead a happy life without the distress brought on by disease. The disease I suffer from today has cast a shadow over my life for some ten years, but I have learned to live with it. I have enjoyed my family, myself and my work both before and after I was diagnosed. I have learned to live with my disease; someone might put it in a more sophisticated way and say that I have 'adjusted'. I was diagnosed with the disease as an adult, which means that I have had to adjust to it, in other words, I have had to accept certain things about my life.

My life

I suffer from a disease which will get worse and for which there is no cure. The disease is not even interesting from the point of view of medical research, because there are so few patients suffering from it and the potential income and profits for the pharmaceutical industry are low. This means that I have had to accept some unpleasant things about my life, such as the fact that

- the state of my health will deteriorate continuously
- the quality of my life will remain satisfactory as long as I have the energy and will to take care of myself. Physical therapy and outdoor recreation help to maintain my fitness at a satisfactory level.

Life around me

The scope for living a normal life is becoming narrower. It is restricted mostly by other people. Some people don't accept that a person who needs assistance goes to the same places as healthy people.

The people closest to me, especially the members of my family, worry about how my disease will progress. I can see very clearly that they are scared about how I

am going to be able to cope in life. People often think that I've had too much to drink, because I stagger when I walk and speak indistinctly. I think this is characteristic of Finns – it doesn't even enter their minds that staggering might be caused by something other than alcohol.

Many sufferers get a lot of support from others with the same disease and like to spend time together. For them, the various club activities for sufferers are important and rewarding. I can't and don't want to be involved in sufferers' clubs.

In all the activities that interest me, I make an effort to be with healthy people. I can cope with many things alongside the healthy. I need to use aids such as a cane, walking aid rollator or wheelchair, but that doesn't matter. Many of my friends are glad to help. I'm good for them – through me they get a chance to do good; to help someone who needs help. Last winter I was President of our local Rotary Club, although I was in a wheelchair.

Working life

I am still also involved in working life, although I would gladly already have given it up many times. So many people close to me have hoped that I would stay working with them as a symbol of joy of life and hope, that I have stayed on.

I love my work and the people I meet through my work. My workplace is comfortable and special equipment has been constructed for me. My colleagues are also very nice and always willing to help me.

Something very important should be noted here. As an older employee, I had reached a good position while I was healthy. When I was diagnosed with the disease, my colleagues were shocked that a good friend had become ill. They have done everything possible to help me cope with my duties. I have been given all the necessary aids that I have requested – my

word has been sufficient to sanction the purchase. Had I been seeking my job as a young, new sufferer, I would hardly have got it, because health and beauty are admired everywhere. Even I got to my position while I was healthy.

Services provided by society

Finland has good legislation on the basis of which a disabled person can get what he or she needs to be able to live as normal a life as possible. The benefits can be divided roughly into a few main groups:

1. Benefits making it possible to work and have hobbies. These may include special equipment and travel services.

2. Health care. Provision of: one institutional rehabilitation session lasting three to four weeks or two two-week sessions per year, for example at the Masku Neurological Rehabilitation Centre.

non-institutional rehabilitation at the place of domicile, for example in my case at the Imatra spa, where I exercise twice weekly on different devices for half an hour, then have physical therapy with a professional therapist for one hour, and then finally go swimming and relax for about one hour. I also have speech therapy once a week for 45 minutes at a time.

3. Travel services

I am entitled to use an ordinary taxi or invalid taxi for travel relating to work and recreation. For these services I myself only pay the equivalent of the cheapest bus fare.

I'm annoyed by the way some people think that if you use an aid, there's something wrong with your head, too, although in most cases this is not so. After all, I suspect that most of us here today are quite healthy in general, if you exclude the fact that our speech falters and legs don't carry us very well – otherwise we are quite OK.

Finally, I would like to say that I lead a good life. Disability imposes some limitations on my life, but difficulties are meant to be overcome. So the daily battle goes on.

IN THE WOODS

Seppo Hirvonen

Some people think that the disabled are excluded from the mainstream of life. I refuse to accept this view and in many ways can show that I am quite able to handle most things. I believe that I am the only one entitled to assess my achievements, and if they satisfy me and make me feel good about myself, then they are good achievements.

Many people have heard that Finland has four very distinct seasons. The present time of year, autumn, is a time when nature is at its best. A popular activity is, for example, walking in the woods and collecting wild mushrooms. I, too, can do a lot of things in the woods, although for some activities I need assistants and aids. However, the only thing restricting my activities is my own resourcefulness. I love being in the woods; you can listen to the silence and enjoy the beauty of nature.

In late summer and autumn the woods in my home district in eastern Finland are full of different kinds of products of nature just waiting to be picked and enjoyed. I'm thrilled when I find berries or mushrooms, but they are only of secondary importance in my trips into the woods. The most important thing is to be able to wander around in the woods, where I experience a wonderful feeling of freedom.

One of the fundamental rights of Finns is to be able to wander in the woods and gather the yield of the forest. Luckily, there are many trails and narrow roads in the woods along which you can go even by car. It also makes me very happy that today there are such good opportunities for outdoor recreation in the woods and on lakes and rivers even for the disabled.

During the past summer, I was able to try out all kinds of equipment that make it easier to move about. In my present situation I have found best a crawler with which I can go to and move around in the woods. This vehicle also makes enough noise to scare off bears, of which there are plenty where I live.

My way of moving about in the woods may seem strange and laborious. Once I've arrived at a suitable place, for example with the crawler – previously with my car – I go out and try to stagger a few meters around my car. Knee pads help in crawling and most times I find something to take home. I enjoy the stimulation of mind and body brought about by being outdoors -I'm not after a big catch.

It will soon be winter. In Finland it is a dark and cold time of year when the days are short. Many people find the conditions – whirling snow and icy roads – hard to bear.

In winter people enjoy being indoors. I'm in the habit of gathering together with friends and family to spend enjoyable evenings with good food and hot drinks. Hot punch warms you up and conversations in candlelight are relaxing.

Soon after Christmas it is spring again. It is still very cold but the darkness begins to give way slowly. It is like a starting signal for many men who take off for lakes which are still covered in thick ice. They are amateur fishermen who bore holes in the ice and lower their fishing tackle into the water. Then they sit and wait for a perch, burbot or some other fish to get caught on the hook. The atmosphere is so electric that those who are nearby come even closer in the hope of catching some fish themselves. I have experienced this excitement myself – my friends have taken me along a few times, although I cause quite a lot of inconvenience. Last winter I caught a burbot which is an extremely ugly but really delicious fish.

In Finland, if you dare, there's a lot you can do in the snow in winter. A lot of people ski, skate and go downhill skiing. Disabled people can take part in most of these activities, although they'll need special equipment. When using them, you can really enjoy a lot of things.

Spring is a wonderful time of year: it is full of hope, the migratory birds return and the air is filled with their joyous singing. Nature awakens – trees grow new leaves and flowers bloom. The woods are full of flowers and everything looks beautiful with people busily planting new plants for the summer. In spring it is worthwhile to be very quiet in the woods because then you can hear the birds singing and the trees sighing. I'm absolutely spellbound if I'm on top of a hill and can see a far-away lake reflecting the blue sky on its surface. The silence and clean air feel good – almost as if new energy was flowing into my body.

By describing these small experiences I want to emphasise to everyone that it's not worth sitting and grieving over your situation, instead take joy in the things you can do and enjoy nature, which belongs to all of us.

PSYCHO-EDUCATIONAL ACTIVITIES FOR PEOPLE WITH ATAXIA

Tarja Ketola

The Masku Neurological Rehabilitation Centre, which is owned by the Finnish MS Society, has for many years organised adjustment training for persons with rare neurological diseases, such as ataxia, and their families. The training focuses among other things on information on the disease and social matters, fears relating to the disease and family members' thoughts and expectations, bearing in mind the special needs of a minority group of the disabled.

On the actual adjustment training course most of the work is done in groups. Participants are divided into two groups of five persons, usually aiming at as great a level of homogeneity as possible within each group. The factors taken into account when forming the groups are diagnosis, duration of illness, age, family relations and sex. In the case of rare diseases, it is not always possible to form a group where everyone has the same diagnosis. If there are several different diagnoses within one group, it is important that the diseases have a sufficient number of common features such as heredity, rate of progression and prognosis, range of symptoms and age of onset.

Physical and cognitive symptoms must be kept separate

The contents of adjustment training consist firstly of information on each person's individual disease and, for example, on social benefits. Secondly, management of the patient's fears and assumptions concerning the disease and its consequences is important, as is the problems faced by being a patient and client (or subject) of social welfare services. The fears or unrealistic expectations reflected by one's immediate surroundings, particularly by the reactions of family members and close friends, are also discussed. Utilising the sufferer's own psychological resources and strengths is also of vital importance.

It is important to learn to distinguish between things caused by the disease, which we can either do something about or nothing about, and things which have nothing to do with the disease. In connection with ataxias it is particularly important to make clear to both the sufferer and his or her close relations that physical and cognitive symptoms are distinct from each other. There is no connection between inappropriate, uncontrolled movements and unclear thoughts or feelings. These are completely separate areas of activity. It is essential to reinstate the person as a subject of his or her own life. It's easy for the person to slip into being an object when going through the health care system, and it's therefore critically important to return to the person the responsibility and freedom that belongs to him or her.



Riitta Rinne and Tarja Ketola

Also if adjustment training does not take into account family members, it can never be as efficient as when family members are involved to the same extent as the patients. Patients cannot be expected to train their families or to manage family members' crises in addition to their own. Rehabilitation and adjustment training are two distinct areas of work with the disabled that support each other. Successful adjustment enhances the effect of rehabilitation.

Adjusting to ataxia is made particularly difficult by the fact that the patients cannot themselves decide when they are ready to tell family and friends about the disease. Ataxia is always socially visible and demands comment. Since the symptoms are obvious to everyone, anyone might ask questions about the condition. It might be more difficult not to reply to questions asked when one is not yet ready to say anything about something that does not show.

Ataxia often makes social life difficult. A person with ataxia may need an assistant or special arrangements from an early stage, which means that social interaction may be curtailed.

For an ataxic person himself it is often most difficult to accept the fact that he or she knows that he or she has the strength but cannot use it because he cannot control his or her movements. Thus an ataxic person has to accept help from others at a stage which often seems too early. Learning to accept help is difficult for almost

anyone. Therefore every possible and even impossible means of coping by oneself are tried before accepting help. Sometimes people are willing even to give up things that are important to them just to be able to avoid receiving help from anyone else. Learning to accept help often includes displays of temper which are usually directed at the people closest to the patient as well as at the patient him/herself. When ataxia is very severe, some people may even think that complete paralysis is a better alternative than ataxia. Ataxic persons are also often ready to try almost anything if there is even the smallest hope of alleviation of the symptoms. It is in fact difficult to find another disorder that would make people go into such trouble, take risks, defy the unknown or to invest both time and money.

Heredity – an issue of its own

In hereditary ataxias, the sufferer's own problems often remain somehow secondary for a long time. Instead, concern for the future of children worries both the sufferer and his or her partner.

The increasing accuracy of genetic tests is continuously producing new information in this field. However, people often find it difficult to accept that parents are not permitted to have their underage child undergo genetic testing to find out whether the child carries the gene potentially leading to the disease. To be able to appreciate the sense of this rule, people must put themselves in the child's position. One must understand that the child's possibilities of being a normal child end with the knowledge of the future disease, and not with the onset of symptoms, which would be the case without that knowledge.

When a child falls ill with hereditary ataxia, parents have to work hard at getting rid of feelings of guilt, although everyone understands that no parent would intentionally cause their child to fall ill. Parents are unable to choose their child's genotype, and so cannot be blamed for their child being afflicted. But understanding this emotionally, alas, is a very long process during which most people need outside help.

The Editors of
Euro-Ataxia
wish you all a
very happy 1999

LIVING AND WORKING WITH ATAXIA

Peter Reussner

I have had ataxia for 24 years now. Presently I am at a moderate stage, although I must use a walker/rollator to move around. At first, the ataxia was diagnosed as Friedreich's ataxia, then as OPCA, and finally as ADCA type 1, as I have some family history in ataxia. Genetically, however, my case is not clear: 'my ataxia-type' has not yet been cloned. I live quite well with



my ataxia. My therapies are: physiotherapy, horse-riding, swimming, my work and other activities.

I am an engineer in a large company and I like my work. It keeps me physically and mentally active. In addition I am surrounded by my colleagues and have numerous social contacts within my company. I have been employed in the company for 32 years now and take my job really seriously, so I don't find any problem in staying in work because of my disability. Although, as my ataxia progresses I am finding it harder to work at my full effort, so for the past eight years I've been working on a part time basis. I hope to be able to continue working like that until my retirement.

**CLOSING DATE FOR
THE NEXT ISSUE
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