



EDITORIAL

This edition of *Euro-Ataxia* starts on a sad note. We have to report the death of Anita Harding, Professor of Neurology at the National Hospital in London, England. Anita Harding was the central authority on hereditary ataxia world-wide, and was responsible for many of the most important advances in recent years, such as her classification of hereditary ataxias in 1983.

As usual we report on the current state of scientific research into all forms of Hereditary Ataxia. Also on the research front Dr. Sandro Banfi, of the Milan-based TIGEM Institute, gives an overview of recent progress in dominant ataxias and in particular, the characterisation of the SCA1 gene product.

Meanwhile EURO-ATAXIA has grown. Three new member groups were admitted at the 7th Annual General Meeting, held recently in Lunteren, the Netherlands. From Finland the Rare Neurological Disabilities Group and from Holland the ADCA Vereniging joined the main body of EURO-ATAXIA. Another group, elected in their absence, were the Association Strümpell-Lorrain from France. The board of EURO-ATAXIA has also changed. Manfred Van den Kerchove becomes the new President. Business of the AGM included a discussion on a set of three resolutions drawn up by Peter Cordwell (UK) to improve the efficiency of EURO-ATAXIA.

Also at the meeting in Lunteren Dr. Carolien Koopmans delivered a talk on the psychological aspects of living with a disability: *The Adventure of a Hero* is reprinted inside.

Ataxia.Net is a new section in *Euro-Ataxia* on the Internet and its implications for ataxic people everywhere.

Finally, dates for next year's EURO-ATAXIA business have been decided. On March 16, 1996 the EURO-ATAXIA board meeting will be held in Brussels as usual, while later in the year the EURO-ATAXIA AGM will come to Belfast, Ireland. It will be held over the weekend of 27-29 September 1996. For more details, booking information etc., please contact the EURO-ATAXIA secretary.

RECENT PROGRESS IN DOMINANT ATAXIAS: CHARACTERIZATION OF THE SCA1 GENE PRODUCT

During the past two years, many exciting discoveries have been made in the field of autosomal dominant spinocerebellar ataxias (SCAs). Most of the credit for this must certainly be awarded to molecular genetics. In fact, molecular genetics studies have provided us with a more precise classification of this group of disorders, based more on their causes (defective genes) rather than on their manifestations (clinical presentation). To date, at least seven forms of SCAs have been recognized from a molecular genetics point of view; in six of them, the gene

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responsible has been mapped and in two cases (SCA1 and SCA3) the gene has been isolated. It is very likely that within just a few years all the genes causing spinocerebellar ataxias will be found.

Finding a gene responsible for a given disease is an important

achievement and an obligatory step towards the understanding of the mechanisms leading to that particular disease. Sometimes, by knowing only a few features of a disease gene, for instance the kind of the protein it encodes for, it is possible to speculate about its putative function and, consequently, its involvement in the generation of the disease. However, in most cases, this is not possible because the disease gene discovered encodes for a novel protein whose function is completely unknown. In such cases, the isolation of a disease gene constitutes only a starting point and one needs to perform more experiments in order to get additional information. A typical example of the latter case is represented by the SCA1 gene.

About two years ago, researchers at the University of Minnesota in Minneapolis and Baylor College of Medicine in Houston isolated the first gene for ataxia, namely the SCA1 gene; in this journal, Dr. Laura Ranum reported on that exciting discovery. Now, I would like to continue the story from precisely that point, in order to explain what happens after a disease gene is cloned.

At that time, there were no clues about the mechanism by which an altered function of the SCA1 gene could cause ataxia. The only thing known was that the gene contained too many copies of the genetic 'word' CAG ('trinucleotide repeat' in scientific terms) in individuals with the disease compared to unaffected individuals. This increase in number of CAG repeats is shared also by other neurodegenerative disorders and, among others, spinocerebellar ataxia type 3. Actually, scientists think that most, if not all, spinocerebellar ataxias are caused by the same kind of alteration. Therefore, it is important to understand how the increase of CAG repeats in the SCA1 gene is able to cause damage in the brain and subsequently ataxia, since this piece of information might shed light on the causes and mechanisms of other forms of dominant ataxia as well.

First of all, the researchers from University of Minnesota and Baylor College of Medicine decided to characterize in detail the protein encoded by the defective SCA1 gene. They were able to 'recognize' this protein in the brain; this is an important achievement because it allows one to identify other proteins possibly involved in the generation of the brain damage. In fact, scientists hypothesize that the

SCA1 mutated protein might react in an abnormal way with other proteins and this abnormal interaction could eventually cause the disease. The precise knowledge of all these molecular mechanisms will allow scientists to try to stop the damage in the future.

But the researchers in Minneapolis and Houston have not stopped at this point. Very recently, they obtained another exciting result. They were able to recreate the disease in animals by inserting the mutated SCA1 gene in mice. As a result of this, they obtained mice with symptoms that are almost identical to the ones seen in humans with SCA1. Now, it will be easier to study the mechanisms leading to ataxia in animals and eventually (everyone hopes not too far from now) try some effective therapeutical strategies.

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SCIENTIFIC ACHIEVEMENTS ON HEREDITARY ATAXIAS: STATE OF THE ART IN 1995

I. EARLY ONSET CEREBELLAR ATAXIAS

1. Friedreich's Ataxia

- a. the FRDA locus has been mapped to chromosome 9q13-21.1 (1988); the candidate chromosomal region has been narrowed to:
 - 1.000.000 base-pairs (1993);
 - 300.000 base-pairs (1994);
 - 150.000 base-pairs (1995).
- b. some FRDA candidate genes have been described:
 - X11 (1993);
 - X104=CSFA1 (ZO-2), X123 (1994);
 - STM7 (1995).
- c. clinical implications of the molecular work:
 - prenatal testing within at-risk families;
 - broadening of FA phenotype (onset may occur after 20/25 years and knee jerks may be retained in some patients).

2. Ataxia with Isolated Vitamin E Deficiency (AVED)

- a. familial isolated vitamin E Deficiency has been described (1981);

the lack of liver alfa-tocopherol-transport-protein (ATTP) is the proposed biochemical fault in AVED (1992);

- b. the AVED chromosomal candidate region spans 4.800.000 base-pairs on the long arm of chromosome 8 (1993);
- c. the gene encoding for ATTP is located on the same chromosomal region and represents the AVED candidate gene (1994); ATTP gene mutations have been found in AVED patients (1995).
- d. clinical implications: presymptomatic detection of patients in at-risk families.

3. Ataxia Telangiectasia

- a. the gene locus has been mapped to 11q22-23 (1988); the candidate region is narrowed to 500.000 base-pairs (1994).
- b. the ATM gene has been cloned and the ATM protein has similarities to phosphoinositol-3 kinase, RAD3 and MEC1 proteins (1995).
- c. clinical implications: screening of AT carriers and prenatal testing within at-risk families; screening the general population.

4. Infantile-onset Spinocerebellar ataxia (IOSCA)

the gene responsible for IOSCA is on chromosome 10q23.3-q24.1. The candidate region spans 4.000.000 base-pairs.

II. ADULT ONSET CEREBELLAR ATAXIAS

1. Autosomal Dominant Cerebellar Ataxia (ADCA) Type 1

at least five entities have been identified within ADCA type 1:

- a. *Spinocerebellar ataxia type 1 (SCA 1)*
SCA 1 gene maps to the short arm of chromosome 6 (1974); SCA1 is caused by an expanded, unstable trinucleotide cytosine-adenine-guanine repeat within a novel gene in 6p22-23 (1993); SCA1 gene has been cloned, it spans 450.000 base-pairs, the SCA1 transcript is 10.700 base-pairs and it is transcribed from both normal and SCA1 alleles (1994); the SCA1 gene product, ataxin-1, is present in brain and peripheral tissues, a longer protein is present in SCA1 patients (1995).
- b. *SCA2*
SCA2 gene was assigned to a 35.000.000 base-pair interval on the long arm of chromosome 12 (1993); the chromosomal candidate region spans 6.400.000

base-pairs (1994);

two candidate genes, NOS and HASH1, have been excluded (1995);

recombination events in the Cuban family position the SCA2 gene within a 3.000.000 base-pairs interval (1995).

- c. *Machado-Joseph Disease (MJD/SCA3)*

the MJD gene has been mapped to a 29.000.000 base-pair segment on the long arm of chromosome 14 (1993);

the chromosomal candidate region spans 3.000.000 base-pairs (1994);

the disease is caused by an expanded CAG repeat (1994).

- d. *SCA4*

SCA4 gene has been localised to the long arm of chromosome 16 (1994).

- e. *SCA5*

SCA5 gene has been mapped to the centromeric region of chromosome 11.

2. Cerebellar Ataxia with Retinal Degeneration (ADCA type 2)

ADCA type 2 (or SCA7) maps to 3p12-p21.1 and the candidate region has been restricted to an 8.000.000 base-pairs interval (1995).

3. Dentatorubropallidoluysian Atrophy (DRPLA)

DRPLA gene has been mapped to the short arm of chromosome 12; the identified gene contains an expanded,unstable trinucleotide cytosine-adenine-guanine repeat in Japanese families (1994); DRPLA transcript is 4.500.000 base-pairs (1994);

DRPLA mutation has also been found in European families (1994);

DRPLA gene product has been identified in human brain and a longer protein is specifically present in DRPLA brains (1995).

4. Familial Periodic Cerebellar Ataxias (FPCA)

- a. a potassium channel gene (KCNA1) in chromosome 12p13 is responsible for FPCA with Myokymia (1994);

- b. the gene responsible for FPCA without myokymia has been mapped to chromosome 19p (1994); the candidate region has been narrowed to 19.000.000 base-pairs (1995).

Clinical implications:

Prenatal and predictive test by direct mutation analysis in SCA1, SCA3 and DRPLA and by linkage analysis in the others.

Alessandro Filla

7th EURO-ATAXIA AGM: AN IMPRESSION

Lunteren, in the Netherlands, provided the setting for the 7th Annual General Meeting of EURO-ATAXIA, held over the weekend of 27-29 October last. To get to it I first flew into Schiphol, to be met by a welcoming committee in the shape of Robert-Jan Schoonenberg, who was to bring us safely via Minibus to our destination. The Dutch have an admirable way of combining efficiency and casualness, it seemed to me, not even the grim experience of the Friday-evening rush hour traffic around Amsterdam caused any panic. After a late dinner we rushed to join the main body of EURO-ATAXIA, which had already opened the conference. I was just in time to hear Alessandro Filla's final speech as EURO-ATAXIA chairman. Not that there was anything sinister about this, just that Alessandro had completed his two-year stint and it was time to move on. After a short break Peter Cordwell addressed the meeting and introduced three important resolutions for us all to mull over during the weekend. Then the bar opened and work was soon all but forgotten...

The morning session was devoted, in keeping with EURO-ATAXIA tradition, to the Scientific programme. First, though, there was a minute silence in memory of Professor Anita Harding, who had died few weeks previously. Alessandro Filla then gave a general overview of research into hereditary ataxia's. My impression was that genetic research was closing in on the location of many ataxia's, and that of Friedreich's ataxia is getting tantalisingly close. Maybe next year? Dr. Ewout Brunt gave an inserting talk on clinical aspects of dominant ataxias. Too much science though is inimical to a good AGM. It remains fairly abstract to many ataxic people, who prefer to hear issues on living with ataxia, which is a reality with which they are familiar. To this end EURO-ATAXIA divides the AGM into 'scientific' and 'psycho-social' parts, normally separated by lunch. A slight rescheduling brought Dr. Carolien Koopmans's excellent paper on the psycho-social aspects of living with a disability, entitled *The Adventure of a Hero*. It also, to an extent, made the rest of the days presentations – on rehabilitation – seem a bit dull by comparison. Then, having done with the formal business, it was time to get down to the more serious business of the AGM: relaxing, making and renewing friendships. By 2 a.m. a new ataxia was in evidence – Heineken Ataxia.

Sunday morning brought the boring bureaucratic bits – the reports of EURO-ATAXIA officers, election of a new President and Committee, lightened up by the admission of new members – from Finland the Rare

Neurological Disabilities Group and from the Netherlands the ADCA Vereniging – and a lively discussion on Peter Cordwell's resolutions, introduced on Friday night. Then it was all over ... for another year.

Michael Morgan

THE ADVENTURE OF A HERO

When I was asked at the last board-meeting of EURO-ATAXIA to write a speech about the psycho-social aspects of having a disability, I didn't yet know in which way I would handle the subject. I wanted to write about the subject in an optimistic manner because the stories you usually hear about disabilities are very sad and negative. Very depressing indeed. My favourite approach is an alternative approach that compares a time full of problems with the adventure of a hero. Hans always teases me that this approach is just a matter of woolly socks and open sandals, but it is not! It is such a beautiful way to look at life, that I love to tell you about it. Well, here is the alternative point of view.

Being a child is wonderful. At least for me – and for the majority of people I know – it was. The world is fascinating for a child, full of things to be explored, everything is new and interesting. Time is an endless flowing stream, not yet split up into years, months, days, hours, minutes and seconds. When I think about my childhood it seems to me like an endless summer-holiday. As a child you've got no – or little – problems on your mind; you could say that your heart and your intellect speak the same language.

The world in which we have to live as grown-ups is far less self-evident than the world looked like when you were a child. Society is very artificial and has nothing natural or obvious anymore. People are separated from each other by very unnatural borders. Our behaviour is ruled by lawbooks. Machines and computers do the work for us. Money is regarded as the most important thing there is. For the sake of money everything has to be sacrificed. When he grows up, the child has to learn to play his future role in this artificial society. As a result of this the child's heart and intellect each go their own way. Sometimes even in opposite directions. It is difficult to explain what I mean by this but I'm sure that everyone present here will have experienced situations in which your heart tells you to do something and your intellect says not to do it. Or the other way around, of course: situations in which your heart says 'no' and your intellect says 'yes'.

Well, a problem that every human being faces, is to

get his heart and intellect in line again. He has to fight a lonely fight to restore the balance of his mind. Most people are afraid to fight this lonely mental fight and just act like there is nothing wrong. To fight this mental fight means that you have to ask yourself questions. Questions like: why do my heart and intellect argue? The Swiss psychiatrist Carl Gustav Jung called the fight to get the mental balance back again the individuation-process, the process one has to go through to change from child to an individual. A real individual, not a person that can't be distinguished from the crowd.

Normally the individuation-process takes place between childhood and adulthood. However, in life all sorts of things can happen. You may experience situations in which your heart and your intellect react in a different way. To give you a few examples: you can lose your parents, partner or one of your children; you can get cancer and are informed that you have only a short time left to live; a married couple can want to start a family but find out that they are unable to get children; you can get a progressive disease; or a pitiful accident can happen that leaves you with a serious disability. Such nasty things do happen – in fact they are happening all the time. Under such circumstances the individuation-process may also start.

There can be told an innumerable lot of stories about the difficult time a person has to go through to become a true individual. Stories about the adventure of a hero tell about this difficult time in a metaphorical way. According to the anthropologist Joseph Campbell the stories of the adventure of a hero are to be found in cultures from all over the world and from all ages. In each culture these stories are told over and over again, from generation to generation. The principle of these stories has stayed the same through all times and places. They not only serve as a pass-time, to be told just for fun around the campfire, but in each culture all the adventures of heroes together function as a means of education. One of the things people can learn from these stories is that courage pays.

The form of the stories of the hero are the same each and everywhere. The hero-to-be almost always makes a long journey to an unknown area. He has to make the trip upon his own, so it's a pretty lonely trip. During that journey he experiences some dangerous situations. He has to find a way out of these dangerous situations. After having undergone these dangers the hero succeeds in returning home safely.

This theme of a journey must be well-known to all of you. It's a theme in a lot of fairy tales, take for instance Hans and Gretchen, Little Red Riding Hood, Snowwhite and the Seven Dwarves, Tom Thumb.

Fairy tales are in fact stories of a hero for little children. In them the danger is simple, the journey is short and the problems are few. Fairy tales always have a happy end; they always close with the sentence: '...and they lived long and happily ever after'. Fairy tales have a pedagogic effect; they show children that solutions can be found for even the most difficult kind of situation. Fairy tales tell children to have courage, never to despair and never to give up hope.

The same theme of a journey to be made comes back in tales that are told and books that are written for the somewhat elder children and the grown-ups: for example the Odyssey, the Knights of the Round Table and the Search for the Holy Grail, Robinson Crusoe, Journey to the Centre of the Earth. Nowadays a lot of movies are built around the adventure of the hero too. A lot of modern books and movies don't have a happy end anymore. That really is a sign that a lot of people have lost the hope that all is going to end up well with the world. But I am an optimist, so let's go on with my tale.

As told before, the stories about the adventures of heroes can be regarded as stories about the mental fight a person has to fight to change from a child into an individual or a true grown-up. Has Jung called that troublesome struggle the individuation-process, Joseph Campbell calls it the fight with the dragon inside yourself. During that time you have to get your heart and intellect in balance again, the way it was when you were a child. The only way to reach that goal is to ask yourself questions. Questions about matters that you never thought about as a child. That travelling of your mind can indeed be compared with a journey to an unknown area. Those questions to which you have to find an answer look very awful in the beginning; they even look frightening at first.

In reality the mental fight often begins when something unusual happens. For instance the death of a husband or wife, the diagnosis that one has a killer-disease, the realization that one can't have children, the beginning of a serious progressive disease. Such events ask for a change in your attitude towards life. You have to find an answer to questions like the following. What is the sense, the meaning and the value of life? What is the sense, meaning and value of death? Why do I go on living if I feel like dying and my life doesn't seem to have any sense anymore? Who and what am I?

Because every person is unique, the problems and dangers each hero-to-be has to face are different. I now want to give you an example of such a story of a hero. I have chosen the story of Buddha. Buddha is one of the great heroes of history and I don't know if everything told about his life has really happened or

is only fiction. But that isn't important at the moment. It's a beautiful story and one can learn a lot from it. I quote the story from Joseph Campbell's and Bill Moyers's *The power of myth*.

“The story of the Buddha's childhood is that he was born as a prince and that, at the time of his birth, a prophet told his father that the infant would grow up to be either a world ruler or a world teacher. The good king was interested in his own profession, and the last thing he wanted was that his son become a teacher of any kind. So he arranged to have the child brought up in an especially beautiful palace where he should experience nothing the least bit ugly or unpleasant that might turn his mind to serious thoughts. Beautiful young women played music and took care of the child. And there were beautiful gardens, lotus ponds, and all. But then one day the young prince said to his chariot driver, his closest friend, ‘I'd like to go out and see what life is like in the town.’ His father, on hearing this, tried to make everything nice so that his son, the young prince, should see nothing of the pain and misery of life in this world. The gods, however, saw to it that father's program for his son should be frustrated. So, as the royal chariot was rolling along through the town, which had been swept clean, with everything ugly kept out of sight, one of the gods assumed the form of a decrepit old man and was standing there, within view. ‘What's that?’ the young prince asked his charioteer, and the reply he received was, ‘That's an old man. That's age.’ ‘Are all men then to grow old?’ asked the prince. ‘Ah, yes,’ the charioteer replied. ‘Then shame on life,’ said the young prince, and he begged, sick at heart, to be driven home. On a second trip, he saw a sick man, thin and weak and tottering, and again, on learning the meaning of this sight, his heart failed him, and the chariot returned to the palace. On the third trip, the prince saw a corpse followed by mourners. ‘That,’ said the charioteer, ‘is death.’ ‘Turn back,’ said the prince, ‘that I may somehow find deliverance from these destroyers of life – old age, sickness, and death.’ Just one trip more – and what he sees this time is a mendicant monk; ‘What sort of man is that?’ he asks. ‘That is a holy man,’ the driver replies, ‘one who has abandoned the goods of this world and lives without desire or fear.’ Whereupon the young prince, on returning to his palace, resolved to leave his father's house and to seek a way of release from life's sorrows.”

It takes some courage to start a journey with an unknown destination. Courage or trust in yourself. In daily life it indeed seems to take some courage to philosophize about life and death. Asking yourself

questions is a lonely experience. You have to be prepared to row against the stream for a while, walk alone on your own way, leave the crowd for a few years. It is such a lonely experience because so little people dare to face nasty problems. That's the great problem of our time. Most people rather do what other people expect them to do, always prefer to walk with the crowd and are afraid to attract negative attention.

When the hero has reached the most far away spot of his journey, he can start his travel homeward again. In contrast with the journey away from home, the return back home is surprisingly easy and quick. When you have reached the bottom of the pit, you are up again in no time. The hero has learned a lot by facing the dangers of the first part of the trip. He now immediately sees the solution to problems and is able to take the shortest way home.

After his return home it becomes clear that the traveller has undergone a transformation. He has become a real hero. During his journey he had to face some dangers. By finding solutions for difficult problems he has gained insight or cleverness and now has the power of true wisdom. It is a characteristic of a hero that he wants to share his wisdom, let others profit of his insight too. The hero will do the best he can to prevent other people from having to go through the same misery he has undergone.

I think that some people here, and certainly the persons with ataxia, will recognize the hero in themselves. The fact that most members of EURO-ATAXIA are heroes, that's what I think Manfred Van den Kerchove meant when he told us some years ago that we were special persons, different from other patients. Alessandro Filla must have pointed to the same thing when he wrote me that being president of EURO-ATAXIA gave him the chance to meet some extraordinary people. Well, here's my compliment back again: it takes one to know one. If you can see the hero in us, you must be a hero yourselves.

In stories of a hero anything is possible. They are not ruled by the laws of logic. Magical things can happen, like people that change into animals and back again, witches that fly on brooms or dead people that speak and give advice. Magicians and wizards often play an important role. This is to show that in reality things that seem to be impossible are possible. If – when I was 15 years old – someone would have told me how bad my physical condition would be when I would be almost 40 and in spite of that would still lead a satisfying life and would still be happily married, I wouldn't have believed him. I would have called him a liar or an idiot. In several myths the hero is helped or guided by a bird that can speak. That is some kind of magic too. It points to the fact that you can be

helped by persons you never expected help from or people of whom you thought they could never be of any help.

I want to end my speech with a beautiful myth I have read in *The hero with a thousand faces* of Joseph Campbell. When I first read it, I felt a warm feeling of recognition. I sincerely hope that all of the ataxic people can see that there is some truth in it. The story shows that it can make a lot of difference if you meet a kind, open person or are confronted by a square-minded person who thinks that physical handicapped are automatically mentally handicapped too. From the first moment I met Hans, I felt that my disability didn't really matter, was only of secondary importance. I do not say that it was love at first sight – it took us seven years to get together – and the first thing we did when we first met was quarrel. But Hans brings out the good sides in me and I really feel like I'm another person when I have his company. The story is from a few centuries ago, from a time as far back as the Middle Ages. The *Zeitgeist* or spirit of the time was quite different in those days. What we should call nowadays a bit naive. Well, here's the story.

“A story, for example, is told of the five sons of the Irish king Eochaid. One day the five brothers went out hunting and somehow got lost in the woods. After a while they got thirsty. They set off, one by one, to look for water. Fergus was the first to find a well, but it was guarded by an old woman. A very ugly old woman who was described in the following way. Her grey wiry mass of hair came out of the surface of her scalp. She had a greenish tusk which curled from her mouth till it touched her ear. Blackened and smokebleared eyes she had. Her nose was hooked and with wide nostrils. She had a wrinkled and freckled belly, very ugly legs with knotty knees. The woman looked indeed disgusting. The woman said to Fergus that he could have some water if he gave her a kiss. He answered that he would rather of thirst than give her a kiss. Then the young man went back to the place where his brothers were, and told them that he couldn't find water. His brothers Olioll, Brian and Fiachra went on the quest for water and equally came to the same well. Each asked the old woman for water, but they were all afraid to give her a kiss. Finally it was Niall who went, and he came to the very well. ‘Let me have water, woman!’ he said. ‘I will give you water,’ she said, ‘but first give me a kiss.’ He answered: ‘Not only will I give you a kiss, but I will even hug you!’ Then he bended to embrace her, and gave her a kiss. At that moment the ugly old woman suddenly changed into a beautiful young woman. In the whole world there was not a young woman whose

gait was more graceful or whose semblance was fairer. Every part of her was queenly: forearms, fingers long and taper, straight legs. A galaxy of charms. Of course the beautiful young woman was a princess, and Niall and she married, and lived happily ever after.”

Carolien Koopmans

GENETICISTS CHALLENGE CHINA TO CHANGE EUGENICS LAW

Two main Genetic Groups in Europe have joined forces to urge the People's Republic of China to drop plans for compulsory childlessness on genetic grounds from their new law on Maternal and Infant Health Care.

In an open letter to the Chinese Government, passed through the Chinese Ambassador in London, Mr. Ma Yuzhen, Professor Marcus Pembrey of the European Society of Human Genetics and Alastair Kent of the European Alliance of Genetic Support Groups – which together represent the broad mass of both medical geneticists and people who suffer from genetic disease throughout Europe – challenge para. 10 of the new law on Maternal and Infant Health Care as an ‘unacceptable misuse of genetic information and a violation of human rights’. Para. 10 reads:

“Physicians shall, after performing the pre-marital physical check up, explain and give medical advice to both the male and the female who have been diagnosed with certain genetic disease of a serious nature which is considered to be inappropriate for child-bearing from a medical point of view; the two may be married only if both sides agree to take long term contraceptive measures or to take litigation operation for sterility...”

The new law on Maternal and Infant Health Care came into effect on 1 June 1995 in seeming breach of the Universal Declaration of Human Rights, article 16: ‘men and women of full age without limitation due to race, nationality or religion have the right to marry or found a family.’

The new law may not even be workable in practice for pre-marital selection would seem to constitute a clear breach of professional ethics. Ironically para. 34 of the same law re-affirms the confidentiality of the relationship between doctor and patient. But such a relationship would inevitably be breached if Doctors were compelled to operate within the new law. They would in effect be acting as genetic policemen,

reporting back to the state what should be confidential between them and their patients.

Protests so far had been met with a 'deafening silence' from the Chinese Government, according to Alastair Kent. 'They are aware that patient groups in Europe are unhappy but have made no official comment as yet.'

BOOKREVIEW

Former President of the Human Genome Organisation Walter Bodmer has teamed up with Robin McKie, the Science correspondent of *The Observer* newspaper to write *The Book Of Man*, a highly-readable yet informed analysis of the current state of Genetics. 'DNA literacy' is the underlying aim of *The Book Of Man* – the necessity of creating a society fully conversant in Genetics, able to engage in rational debate about new genetic technology, rather than allowing themselves to be carried away by the Frankenstein's Monsters scenario peddled by alarmists.

The result of this marriage of science and journalism is a fast-paced, gripping, though occasionally breathless, account of Modern Genetics. It does so with style, using innovative and novel approaches. Ferniehirst Castle in Sixteenth century Scotland provides an introduction into the intricacies of inheritance through a discussion of the left-handed Kerr family.

All the main Personalities in Genetics are here: from Mendel to Watson and Crick, through to the present 'Lords of the Genome', while pathbreaking developments such as DNA sequencing., recombination and the discovery of PCR (polymerase chain reaction) are made accessible to all. As expected Bodmer himself has written the chapters on the Human Genome project and gene mapping.

What also comes across is the sheer pace of change in modern genetics, where revolutionary developments occur on almost a yearly basis. Even the geneticists of only forty years ago might find difficulty in recognising themselves among some of today's 'Lords of the Genome'. A description of Herbert Wayne Boyer in 1981 is reported thus: 'In his faded jeans and open leather vest, a can of *Budweiser* in his hand, he looks just like a leftover from the 1960s...' Style-conscious ataxia researchers please note!

Not everything in the book is well-treated though. The last chapter is concerned with the ethical dilemmas raised by genetic technology – entitled, 'The Slippery Slope'. But what Bodmer and McKie actually do is to rush through this complex area, dealing with honestly-held concerns somewhat brusquely, even at

times dismissively. For instance voluntarism in Genetics – such as the right of individuals alone to decide on genetic testing – is generally held to be sacrosanct, yet Bodmer and McKie simply overturn this by drawing an analogy with the involuntary character of seat belt legislation. Elsewhere they cheerily report that setting up Genetic Testing Centres for Cystic Fibrosis only costs £80,000, compared with the £125,000 needed to keep one CF individual alive. Invoking utilitarian, economic and 'involuntary' arguments as they seem to do they might be thought to be already skating freely down the slippery slope...

Nevertheless, *The Book Of Man* is an excellent overview of Genetics and comes recommended. The Authors are quite right: DNA literacy is important – for everyone.

Michael Morgan

The Book Of Man, by Walter Bodmer and Robin McKie is published by Abacus Books in the UK, price £8.99.

ATAXIA.NET

Ataxia.Net is a new section in *Euro-Ataxia* on the coming of the mighty Internet and its implications for ataxic people everywhere. We'll aim for a hype-free environment, cutting through the jargon whenever possible, and offer new addresses and sites of interest as they become available. If you have any contributions, suggestions, etc., please get in touch.

What is the Internet and why is it/will it be so important?

Basically it's a communication device, both global and virtually instantaneous. You can communicate with any other Internet user anywhere within seconds. It's supposed to be cheap, although you'll need a computer, a modem and to open an account with a service-provider to get connected. None of these are cheap and you'll also need to watch the phone bills... The second part of the question is a little trickier to answer. Because everybody says so is the obvious answer! Never in the field of human endeavour has so much hype surrounded any new gizmo... Yes, I can see its uses (especially for scientists and journalists and others who shunt around information on a regular basis), but some of the more extreme claims by 'techno-hippies' for instance are off the Californian wall. Just keep some perspective and you'll be OK – remember you can't eat it, you can't drink it, and you can't have sex with it. So be cool, be happy and turn it off from time to time.

• The Features editor has got a new e-mail address. He used to have a nifty German one (he doesn't know why either) but has since been admitted to the UK. Contact him (and *Euro-Ataxia*) at:

m.morgan@mcr1.poptel.org.uk

• From Arnie Gruetzmacher of the National Ataxia Foundation in America we received information that the NAF internet address as provided in our last issue was not correct.

The new and correct NAF E-MAIL address is:

naf@mr.net

• NAF have also launched a new NEWSGROUP:

alt.support.ataxia

• However, Arnie has since added this appeal: 'We need your help. I understand that a newsgroup needs messages to get the group moving around the world – that is where you come in. If you find the group, please commit to a message a day for a week. I hope you continue on with us much longer, but that would sure help get us moving. Please give me any feedback if possible & thanks for your help.'

Some interesting World Wide Web sites to check. We got these mainly from UK magazines so they're English language and relate usually to UK matters:

• Disabilities Access – news, advice and text of Disability Now.

http://www.pavilion.co.uk/daccess

• Muscle Power on World Wide Web

Muscle Power is an organisation of people with Neuromuscular Impairments. However, the information contained on the service is largely relevant to all disabled people. Already, there is an online magazine, benefit information, letters page, small ads and lots of information about Muscle Power. The Web address is:

http://www.globalnet.co.uk/~pmatthews/DisabilityNet/MusclePower/MusclePower.html

E-mail: pmatthews@cix.compulink.co.uk (Paul Matthews, General Secretary, Muscle Power)

• ARIES is a non-profit association registered in Brussels, and set up by the European Foundation Centre, the European Citizens Action Service, and the European Committee of Worker Co-operatives. ARIES is supported financially by the European Commission Social Economy Unit, but this does not cover all costs, and ARIES makes a small charge for subscriptions of 50 ECU (equivalent to 42.50 pounds sterling or about 65 US dollars) per year. The service is aimed to meet the needs of the co-ops, mutual organisations and

non-profit organisations, as well as people in local and national government who work with these kinds of organisations, together with academics and other interested individuals.

ARIES is offering a one-month free trial to anyone who requests it. Delivery during the free trial is by mailing list. During the trial period, users receive all messages posted, but can later select the topics to which they would like to subscribe.

If you would like to receive ARIES information by being added to this mailing list for one month, please send your request by e-mail to:

aries-v.woodell@geo2.poptel.org.uk

quoting the e-mail address to which you would like to have the information delivered. Alternatively, you can give your postal address and they will send you some written information.

Vivian Woodell, ARIES, 1 Rue de la Concorde, B-1050 Brussels, Belgium

Tel: +32 2 513 7501/9502 or +44 1608 644022

• The Vereniging Spierziekten Nederland (Dutch Association for Neuromuscular Diseases), the parent organization of the Dutch FA-group, has recently established its own homepage on the Web. The site is still under construction and offers information in Dutch only, although some interesting links with other (international) sites are offered.

http://www.buscom.nl/vsn/vsn.htm

If you've been phoning – or faxing – the UK recently and can't get through don't panic... All UK phone codes have been slightly changed. You now have to add one more digit to the area code (The bit before your personal number). Appropriately enough this is a '1' and it's simply stuck on the front of the aforementioned code. For instance to ring Michael Morgan in Belfast, instead of dialling +44 232 611076 as you used, you now dial +44 1232 611076. And what's true of Belfast, Ireland, also applies to Guildford, England, home of FAG. Hence if you want to contact FAG Office don't ring +44 252 7028864 anymore but dial +44 1252 7028864 instead.

Not only in the UK telephone numbers have changed. In the Netherlands approximately 75% of all numbers have changed too, area codes as well as personal numbers. But the old numbers will be valid until 10 April 1996.

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