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PRACTICAL INFORMATION
FOR WOMEN WITH AMN
(ADRENOMYELONEUROPATHY)

ALDLIFE
helping to cope - helping to hope
A charity to promote awareness, research and prevention of adrenoleukodystrophy

PRACTICAL INFORMATION FOR WOMEN WITH AMN (ADRENOMYELONEUROPATHY)

Introduction

You have been diagnosed with AMN – adrenomyeloneuropathy – a rare inherited metabolic disorder that affects around one in 25,000. It is so rare that only around 30,000 men and women in the world have the disorder.

AMN in women usually starts causing problems around the age of 40, though there are exceptions. It is impossible to predict how mild or severe your AMN symptoms will become, or indeed if you will develop any. Although research into new treatments is ongoing, AMN is currently an incurable condition. However, there are many things you can do to help you and loved ones to live with this disorder.

ALD Life is Britain's leading charity dealing with your condition, run by people who have been in the situation you are facing now and who can help you.

> "IT WAS A RELIEF TO FIND A RELIABLE & TAILORED INFO CENTRE LIKE ALD LIFE."

We must stress there is no right or wrong way of dealing with AMN. In researching this booklet, we have spoken to many women who have AMN, and have put together the information they wished they had been given when they were diagnosed. The medical information, shown in shaded boxes, has been compiled from reputable sources and checked by medical experts in AMN.

We have a worldwide contact list of people who are willing to chat about all aspects of living with the disorder.

To get in touch please contact ALD Life on 020 7701 4388 or email info@aldlife.org
Or visit our website at **www.aldlife.org**

PRACTICAL INFORMATION FOR WOMEN WITH AMN (ADRENOMYELONEUROPATHY)

What is AMN?

Adrenomyeloneuropathy (AMN) is caused by mutations in the ABCD1 gene. This gene is also affected in ALD, a serious degenerative disease that mainly affects young boys. AMN most often appears in adult men. In women the effects of AMN are usually less severe than in men, and progress more slowly. ALD Life have produced a separate booklet for men diagnosed with AMN.

Symptoms of AMN can include stiffness, weakness and pain in the legs. This starts gradually and can progress over time. The medical term for this is 'progressive spastic paraparesis'. Damage to the nerves supplying the legs means unsteadiness and falls are common. The nerves to the bladder and bowel can also be affected in AMN.

Mobility can gradually deteriorate to the point where the sufferer develops significant problems. In rare cases women with AMN may become wheelchair bound.

Males with the ALD gene can suffer from adrenal failure, although not everyone is affected. Primary adrenal insufficiency, known also as Addison's disease, is caused when the adrenal glands do not produce enough of certain steroid hormones. Symptoms include chronic fatigue, muscle weakness and weight loss. It is treated by taking replacement hormones. It is extremely rare for females with AMN to suffer from adrenal failure, and routine testing is not required.

What causes AMN?

In AMN, there is damage to the axons of the nerve cells which control the muscles. Axons carry information around the nervous system in the form of electrical impulses. The nerves that run down your spinal cord to your arms and legs are made up of bundles of axons. The axons are coated with a sheath of a fatty substance called myelin, which allows them to carry information quickly and accurately.

In AMN these axons are damaged and can die back. Myelin is also lost. As a result, signals from the brain do not reach the muscles in the way they should. The longest axons, the ones to the legs, seem to be prone to damage in AMN. That is why with AMN you usually begin to notice problems with your lower limbs. The nerves to the bladder and bowel are also affected.

What drugs should I be taking?

There are currently no drugs that can repair nerves or stop axons from dying back. However, there are drugs available that can help alleviate some of the symptoms of AMN, such as stiffness. AMN sufferers report that these drugs seem to work for some people, but not for others.

You should discuss with your consultant the best treatments to help relieve your symptoms. You can also speak to other people with AMN via the ALD Life website, www.aldlife.org

What about Lorenzo's oil?

Lorenzo's oil is a blend of four parts glycerol trioelate oil and one part glycerol trierucate oil. It was developed to try and correct the biochemical abnormalities in the blood which are associated with both AMN and ALD: raised levels of very long chain fatty acids (VLCFA). It was thought that these raised fats in the blood might be causing the damage to the nervous system in ALD and AMN.

Unfortunately, although treatment with Lorenzo's oil, along with a special diet, does correct the blood abnormalities, it has little effect on the disease.

In young boys, treatment with Lorenzo's oil may be able to delay the onset of the brain disease ALD but, despite what is shown in the film, Lorenzo's Oil, there is no proof that it can affect the symptoms of ALD in any way. Clinical research has so far shown no evidence of benefits with Lorenzo's oil in AMN, and it is therefore not recommended as a treatment .

It must be emphasised however, that this is a very personal choice and some patients find benefit in keeping their diets low fat and using Lorenzo's Oil to assist in bringing down their VLCFA levels. Research is still ongoing into the consequences of raised VLCFAs on the general health of those with the ALD gene.

What tests should I have?

When you are diagnosed with AMN you will be seen regularly by a specialist who will monitor your condition and any deterioration. They may arrange electrical tests to look at the function of your nerves and spinal cord. Although you may have an MRI scan done as part of the initial tests to make the diagnosis, the brain is not affected in women with AMN and regular brain scans are not required. Your specialist will advise on what treatment options are available for your symptoms.

If this is the first instance of ALD in your family, it is imperative you have access to genetic counselling and arrangements are made to test your immediate family. See page 10 for further details of the genetic implications of your diagnosis.

PRACTICAL INFORMATION FOR WOMEN WITH AMN (ADRENOMYELONEUROPATHY)

Living with AMN

When you are first diagnosed with AMN, it's likely that you will have begun to have problems with walking and balance, fatigue & muscle spasm are also common. Perhaps you are experiencing bowel and urinary difficulties.

You may well have to live with these problems for many years. We have compiled a list of tips from AMN sufferers that may help you to manage living with this rare disorder.

Stay healthy

Stay as healthy as you can in every other way. Try not to get overweight, and eat a good, nutritious diet. That does not mean denying yourself treats, but do try to eat sensibly.

Exercise and flexibility

We would advise anyone with AMN to develop an exercise and stretching programme.

If you have never exercised before, it is not too late to start. The more you can keep your body in tip-top condition the better able you will be to cope with the problems this disease is going to cause you. As well as the physical benefits, exercise can also give you a psychological boost.

Your local gym or leisure centre should be able to advise on exercise programmes for the less mobile to strengthen core muscles. Water based exercise such as aquafit and swimming is reported to be especially effective. Sometimes your GP can refer you to these sessions.

Even everyday things like taking a brisk walk most days, climbing stairs and cycling will help you keep in shape.

Work on your flexibility. The biggest problem with AMN is often that your leg muscles become spastic, which means they are artificially stimulated by your nervous system into behaving as if they are under tension all the time. Physiotherapists call this excess tone. Every AMN patient will tell you that when they get up in the morning their legs are stiffer than they were the night before. This is because during the night your nervous system has been stimulating your leg muscles.

One solution to this is to develop a stretching programme, with advice from your physiotherapist, and carry it out regularly.

Another thing that AMN patients find useful and have recommended is a massage bed that combines cycloidal massage and infra-red treatment to help relieve the effects of spasticity www.cyclo-ssage.com. Walking awkwardly puts a lot of stress on your back. Pilates exercises centre your core muscles. When you have a disability like AMN, the more you can strengthen these core muscles the better.

Balance and foot problems

AMN is a condition that affects your balance and you may have difficulty with falling over.

One of the things that affects your balance is how your feet work. A problem with AMN is that you are constantly clawing with your toes to keep your balance. In addition, the nerves to the feet are damaged and you may not be fully aware of pain and high temperatures, putting you at risk of damaging your feet.

Most people with AMN have problems with their feet and should be referred to an orthotics department. The orthotist will make inserts for your shoes, moulded to the shape of your feet to assist with this.

It's important to remember that every little helps. If you can improve your mobility by five, or even three, per cent then it really helps.

"I FOUND YOGA HAS HELPED STABILISE MY CONDITION."

"IF I DON'T HAVE MY TOE PROPS THEN I DON'T WALK AS WELL."

"SOME DIETARY SUPPLEMENTS HAVE BEEN VERY HELPFUL FOR ME – IT'S USEFUL TO INVESTIGATE RELEVANT NON-TRADITIONAL APPROACHES TO MAINTAIN OPTIMUM HEALTH."

Walking aids

Some AMN patients use FES (functional electrical stimulation). Basically it is a piece of equipment that straps to the leg and gives out a stimulating electrical current, which helps your walking by helping your foot to lift. FES is now available from a number of NHS clinics. Ask your doctor about getting referred for assessment.

A new walking aid called Musmate has also been developed, which some AMN sufferers find more useful than FES. More details at www.musmate.co.uk.

Some people with mild symptoms find simple aids such as a walking stick sufficient to begin with, but may need further intervention later on.

Bladder and bowel problems

Incontinence is a problem for anyone who has a neurological condition affecting the spinal cord and pelvic nerves, such as AMN. Unfortunately, bowel and urinary problems can affect AMN sufferers. Urinary urgency (the need to rush off to the loo straight away) is common, and sometimes people do not make it in time. Constipation is very common.

These symptoms tend to get worse as time goes by. It is one of those sensitive areas that people have difficulty talking about, but treatments are available. It is important to discuss these issues with your doctor, who can refer you to a urologist.

There are around 7,000 locked public toilets throughout the UK for people with disabilities, bowel and urinary problems. For a small fee you can purchase a key that allows access to these toilets that have been locked to prevent vandalism and misuse. www.radarkeys.org

ALD Life can also provide you with a card explaining you have a condition causing urgent need to use a toilet. Most shops etc. will allow you to use their facilities upon production of this card.

What about my children?

If you have been diagnosed with AMN then you have the AMN/ALD gene. There is a 50-50 chance of passing the gene on to any children you have.

If you already have children, we recommend they should be tested immediately to see if they have the ALD gene. Your brothers and sisters should consider testing too. Testing is especially important if you have young sons or male grandchildren, because if the gene is discovered early, before symptoms appear, there is a chance of preventing ALD from developing. ALD is a terminal disease. Boys with the gene should be considered for a preventative programme, which is explained in another leaflet - Boys with Asymptomatic ALD.

There is a 1 in 2 (50%) chance any daughters you have are carriers of ALD. If they are, they have a 50% chance of passing the gene on.

The idea of genetic testing for your children can be frightening. Obviously, if they are adults, they will need to make their own decision. A genetic counsellor can talk you or your children through the implications of testing, and help you make an informed choice. Your GP or specialist can put you in touch with one.

Both men and women who have the ALD gene need to consider their options carefully when thinking of having children. There are ways to have children who are free from the gene. This is discussed in our leaflet for female carriers of ALD.

“DOCTERS HAVE NO IDEA HOW TO FIX US...YET.”

“PEOPLE NEED TO BE OPEN-MINDED ABOUT NEW DEVELOPMENTS AND YOU SHOULD TRY MANY THINGS TO SEE IF THEY HELP.”

Explaining the genetics of ALD

ALD is an X-linked disorder, which means that the genetic abnormality involves the X-chromosome.

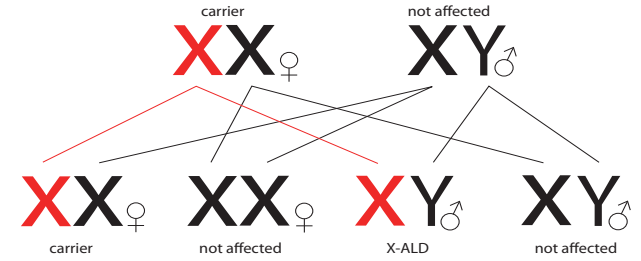
Women have two X chromosomes. In women who carry the ALD gene mutation on one X-chromosome, the full-blown disease does not appear because there is a normal copy of the gene (Figure 1) on the other X-chromosome.

Men have one X-chromosome and one Y-chromosome (Figure 2). In men who have ALD on an X-chromosome there is no other X-chromosome for protection; therefore the male can develop symptomatic ALD.

For reasons we don't understand, not all males who have the gene are affected in the same way. Some develop cerebral ALD as boys, others have no symptoms for many years and then develop AMN as adults. Some males develop Addison's disease (adrenal insufficiency) rather than ALD or AMN. Most women with the gene will also develop some, usually mild, AMN symptoms later in life.

Figure 1:

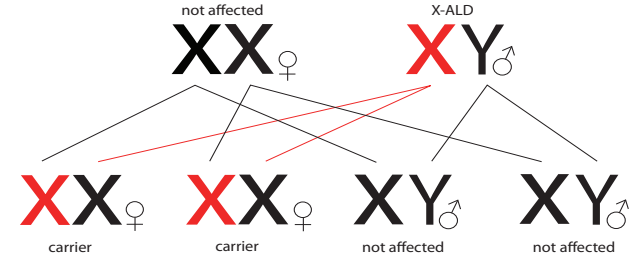
If a woman is a carrier for ALD she has the following possible outcomes with each newborn: With a daughter, there is a 50 % chance (1 in 2) that the daughter is a carrier of ALD and a 50 % chance the child is unaffected. Where the child is a boy there is also a 50 % (1 in 2) chance the son will have ALD and a 50 % chance he will be unaffected.



X **X-ALD X-chromosome**
X **normal X-chromosome**
Y **Y-chromosome**
♀ **Female**
♂ **Male**

Figure 2:

If an affected man has children, then all of his sons will be entirely normal (because sons get the father's Y-chromosome). But all of his daughters will be carriers (because he passes his only X-chromosome to his daughter).



X **X-ALD X-chromosome**
X **normal X-chromosome**
Y **Y-chromosome**
♀ **Female**
♂ **Male**

Can I get any financial help?

Anyone with AMN should apply for the Personal Independence Payment (PIP) if they are having mobility problems or need help with personal care. www.gov.uk/pip PIP gives a monthly tax-free income, which will help you with items that you need to make life easier, like a car through the Motability scheme. www.motability.co.uk.

If you need to stop working because of your AMN you should consult the government website at www.gov.uk/financial-help-disabled or the independent website www.entitledto.co.uk for information on what you are entitled to.

How can I get more information?

You can get in touch with others in a similar situation through ALD Life, who can also provide practical information, support and advice. ALD Life also runs an annual event which brings together families and individuals suffering from all aspects of carrying the ALD gene to learn about innovations in treatment and research and share experiences.

Listed below are just some of the useful organisations that are there to help you:

INFORMATION, SUPPORT AND ADVICE

ALD Life

ALD Life was founded by Sara Hunt after both her sons were diagnosed with ALD. She has first hand experience of most aspects of dealing with the disorder: her elder son, Alex, had symptomatic ALD and was diagnosed at age 7 in 2001 and passed away in 2012. Her younger son, Ayden, had a successful bone marrow transplant in 2008. The charity provides practical, emotional and financial support for all those affected by ALD and AMN.

www.aldlife.org
Tel: 020 7701 4388

Email: info@aldlife.org

Carers UK

Carers UK campaigns to make sure carers receive the practical, financial and emotional support they need.

www.carersuk.org

Citizen's Advice Bureau

Free information and advice on legal and money problems and can help you if you experience problems with DSS. Your local branch can be found on the national website.

www.adviceguide.org.uk

Directgov

Government website for information about benefits and entitlements.

www.direct.gov.uk

Personal Indepence Payment helpline

Tel: 0345 850 3322

Attendance Allowance helpline

Tel: 0345 605 6055

"ONCE YOU FIGURE OUT YOUR BODY ON AMN AND GET INTO A ROUTINE THERE ARE MANY WORSE THINGS OUT THERE."

PRACTICAL INFORMATION FOR WOMEN WITH AMN (ADRENOMYELONEUROPATHY)

Directory of Grants for Individuals in Need

Directory of grants for Individuals in Need available from the reference section of your local library or online.

www.grantsforindividuals.org.uk

Disability Rights UK

Provides advice and support for disabled people on a wide range of issues from claiming the right benefits to employment and independent living.

Ground Floor
CAN Mezzanine
49-51 East Rd
London
N1 6AH

www.disabilityrightsuk.org
Tel: 020 7250 8181
Email: enquiries@disabilityrightsuk.org

Disabled Living Foundation

Disability Living Foundation is a national charity providing independent advice on mobility aids, disability aids, daily living equipment.

www.dlf.org.uk

Motability Scheme

The Motability charity helps provide cars, wheelchairs and powered scooters for disabled people in the UK.

www.motability.co.uk

The Multiple Sclerosis Society

operates a number of therapy centres around Britain, which can be used by AMN sufferers. For details of therapy centres in you area log on to.

www.mssociety.org.uk

INFORMATION SOURCES

The medical information in this leaflet, shaded in pink, has been compiled from the following references, and reviewed by an expert doctor.

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All medical and benefits information is correct at time of going to press January 2015.