



helping to cope - helping to hope



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PRACTICAL INFORMATION
FOR MEN WITH AMN
(ADRENOMYELONEUROPATHY)



A charity to promote awareness, research and prevention of adrenoleukodystrophy



PRACTICAL INFORMATION FOR MEN WITH AMN (ADRENO MYELONEUROPATHY)

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.

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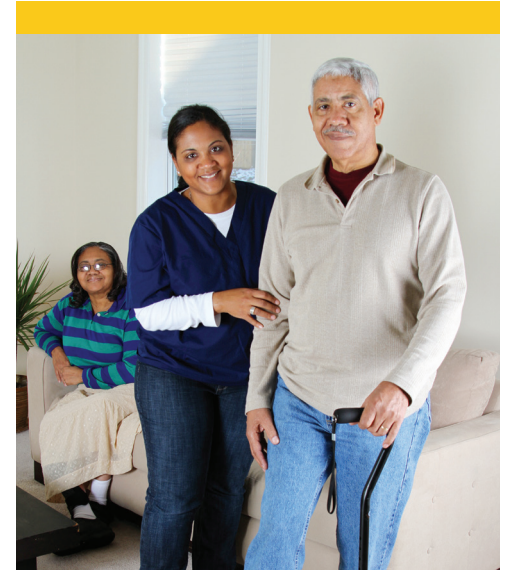
IT WAS A RELIEF TO FIND
A RELIABLE & TAILORED
INFO CENTRE LIKE
ALD LIFE.

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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.

We must stress there is no right or wrong way of dealing with AMN. In researching this booklet, we have spoken to many men 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 MEN 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. Women can also get AMN, but the condition is usually less severe than in men. ALD Life have produced a separate booklet for women 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, bowel and sexual organs can also be affected in AMN.

Mobility can gradually deteriorate to the point where the sufferer eventually loses the ability to walk and becomes wheelchair bound. But this does not necessarily happen to all sufferers.

About 20 % of men with AMN eventually develop some damage to nerves in the brain, which may result in dementia-like symptoms. Occasionally, some men affected in this way go on to experience severe physical and mental deterioration, similar to that seen in boys with ALD.

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, bowel and sexual organs are also affected.

Addison's Disease or adrenal insufficiency

Males with the ALD gene can suffer from adrenal insufficiency, 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. People with adrenal insufficiency can become extremely unwell very quickly if they get a viral or bacterial infection. If you are going to

develop adrenal insufficiency, this will usually happen before your legs are significantly affected. It is unusual for men with AMN to develop adrenal failure once they have a spastic paraparesis.

Once you have been diagnosed with AMN make sure that you are checked for adrenal insufficiency as soon as reasonably possible. In ALD and AMN, the adrenal gland, which produces an essential hormone called cortisol, can stop working. The consultant who made the AMN diagnosis should refer you to an endocrinologist (a doctor specialising in hormones), who will be responsible for making sure adrenal tests are carried out.

Adrenal insufficiency can be treated easily with steroid tablets (hydrocortisone with or without fludrocortisone) to replace the hormones the adrenal gland usually produces. If untreated, adrenal insufficiency can lead to a life-threatening condition called 'adrenal crisis'.

A common sign of adrenal insufficiency is being bronzed, i.e. becoming progressively tanned even in winter, sometimes with brown creases on the palms of the hands (rather than red ones).

Other signs include becoming lethargic and having difficulty recovering from infectious illnesses. For example, a minor viral infection may result in severe repeated vomiting which can lead to dehydration and potentially dangerous changes in blood salts. This can also

happen when males with adrenal insufficiency are very stressed, such as by unusually heavy exercise.

Training will also be given to your partner, friend or relative to give an emergency cortisol injection in case you are not retaining your hydrocortisone due to vomiting or diarrhoea.

The endocrinologist should also give you a letter stating that if you have to go to Accident and Emergency you are to be seen immediately because you suffer from adrenal insufficiency. The endocrinologist will also inform the ambulance service that you have adrenal insufficiency so they carry the correct medication for you if you need to call 999. In some areas the procedure can be different so please ensure you check the correct procedure for your area.

All with adrenal insufficiency should have a medical identity bracelet or necklace stating their medical condition that they wear all the time. That way, if something happens and you are alone, medical authorities will be alerted to your condition and be able to treat you properly. There are many places now that supply these – just search online for medical ID jewellery for a list of suppliers.

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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 trioleate 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 may be offered annual MRI scans to check that there is no evidence of disease in your brain. It is estimated that up to 20-30 % of men with AMN may go on to develop evidence of brain involvement, similar to that seen in boys with ALD, as they get older. This is not usually treatable, although bone marrow transplantation, which is a very major procedure, has been tried in a few cases as a way of preventing further deterioration.

It is not always clear what changes on MRI scans mean, and as there is no treatment available, regular MRI scans are not recommended by all doctors. In any case, they are not something that all people with AMN would want to have. In consultation with your doctors, you must make that choice for yourself.

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. Perhaps you are experiencing bowel and urinary difficulties.

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

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 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.

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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.

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.

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

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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.

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I FOUND YOGA HAS HELPED
STABILISE MY CONDITION.

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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.

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 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 if necessary.

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.

Erectile problems

Erectile problems (impotence) can happen in AMN due to damage to the nerves. A lot of men worry that developing AMN will impair sexual function. However, it's not inevitable, and the chances are you won't have any problems sexually.

If you do find this is a problem, don't suffer in silence. A number of different treatments are available; ask your GP for details. GPs are used to treating erectile problems, so there is no need to be embarrassed.

What about my children?

If you are a male with the ALD/AMN gene, you will always pass it on to any daughters you have – see the section on genetics on p.10. Upon diagnosis you should be offered genetic counselling and have your immediate family, siblings and any daughters tested immediately.

Females with the gene tend not to have such serious symptoms as males. However, they can pass it to their sons, who may be seriously affected in childhood or as adults. So if you have daughters it's very important that they get tested to confirm if they have the gene, particularly if they have young male children. There is a chance ALD in boys can be prevented if it's caught before symptoms appear.

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, and this is discussed in our leaflet for female carriers of ALD.

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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 (see 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.

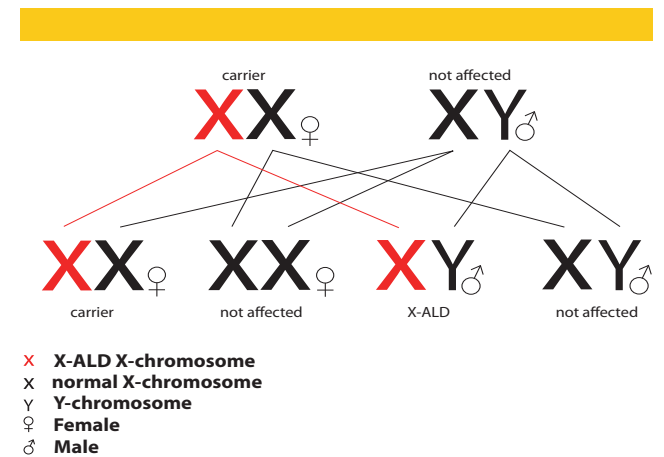
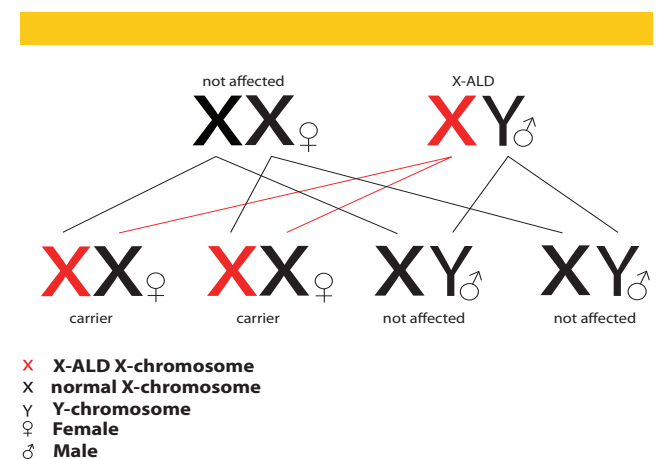


Figure 2:

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).



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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.directgov.uk. 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.

For adult males with adrenal insufficiency, free prescriptions are available even if you are working. According to the NHS website, Medical Exemption (MedEx) certificates are issued on application to people who have a form of hypoadrenalism (for example Addison's disease) for which specific substitution therapy is needed. To apply ask for an application form FP92A, available from your doctor's surgery. You need to fill in parts 1 and 2 and your doctor (or an authorised member of the practice staff) will sign to confirm the information you've given is correct. The surgery will arrange to send completed application forms and applications are normally processed within three working days.

If you need to stop working because of your AMN you should consult the government website 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



“ 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.

”

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Carers UK

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

www.carersuk.org

Citizens Advice Bureau

Citizens Advice Bureau have free information and advice on legal and money problems and can help you if you experience problems with benefits or housing. 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 Independence Payment helpline

Tel: 0345 850 3322

Attendance Allowance helpline

Tel: 0345 605 6055

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

Multiple Sclerosis Society

Multiple Sclerosis Society operate a number of therapy centres around Britain, which can be used by AMN sufferers. For details of therapy centres in your area log on to their website.

www.mssociety.org.uk

INFORMATION SOURCES

The medical information in this leaflet, shaded in purple, 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.

