Special report:

Challenges and needs of people living with adrenomyeloneuropathy (AMN)

November 2019

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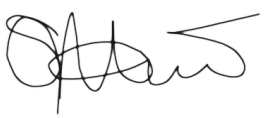
Executive summary

People with adrenomyeloneuropathy (AMN) are struggling to get an accurate diagnosis and have limited access to therapists to help with movement issues and everyday living. Symptoms of depression are common after receiving a diagnosis of AMN. Nearly all report balance and walking problems, and only a quarter feel their symptoms are well-controlled. Less than half have an annual brain scan. Most are worried about the impact of AMN on family life today and in the future, including the impact of cerebral involvement.

These are the key findings from a survey carried out by Alex, The Leukodystrophy Charity (formerly ALD Life), in partnership with Raremark, an international patient-powered network focused on rare diseases.

The survey also showed that people with AMN want to see greater AMN awareness amongst healthcare professionals, more information about clinical trials to take part in, and easier contact with other people with AMN. We hope you find this report helpful.

Pete Chan
Head of Research & Analysis



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What we did

We asked men over 18 in the UK, diagnosed with AMN, to complete a 15-minute online survey to improve our understanding of the condition. We asked participants about their symptoms and treatment, the health and support services they used and the impact of AMN on their daily lives and likely effects in the future. All the questions in the survey were approved by a leading AMN doctor, and all answers were anonymized.

Why do we need this survey?

We want to raise awareness about AMN, and show healthcare professionals and other health service providers the true impact of the disease on the everyday activities of people living with it. We believe that the patient voice is essential for successful planning and implementation of improvements in care for people with AMN now and in the future.

Who took part?

- 26 participants completed the survey
- Their average age was 45 years and their average age at diagnosis was 36.5 years
- Almost half (42%) have a family history of adrenoleukodystrophy (ALD) or AMN

Notes

1. Survey participants were drawn from the Alex, The Leukodystrophy Charity (formerly ALD Life) and Raremark communities, although individual participants may not have been members of both organizations when they completed their survey.
2. Specific questions may have been answered by fewer than 26 participants.

What were the results?

Symptoms

Balance problems, lack of coordination and weakness and stiffness in the legs were the most common first symptoms of AMN. Balance problems and difficulty walking affected nearly all respondents at the time of the survey, reflecting what is known about disease progression in people with AMN.

Symptom	% of participants reporting this as their first symptom(s)*	% of participants reporting this symptom(s) at time of survey*
Balance problems	50%	96%
Lack of coordination	50%	69%
Leg weakness and stiffness	46%	88%
Difficulty walking	38%	92%
Foot drop	38%	77%
Leg muscle spasms/cramps	38%	85%
Fatigue	35%	85%
Adrenal insufficiency (or Addison's disease)	31%	50%
Urinary incontinence	31%	73%
Bowel incontinence	27%	50%
Sexual dysfunction	23%	58%
Weight loss	19%	23%
Skin bronzing/darkening	15%	15%
Nausea	0%	15%

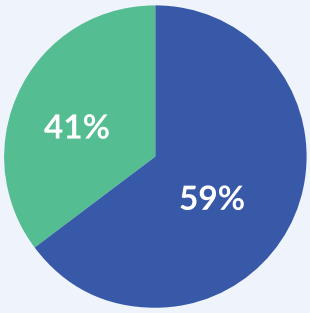
*Participants could select more than one symptom

Aside from the symptoms presented as options in the survey (ie: the symptoms listed in the table on the previous page), participants also told us they experienced other first symptoms, including memory problems, mood and personality changes (eg: anxiety, depression and irritability) and poor vision.

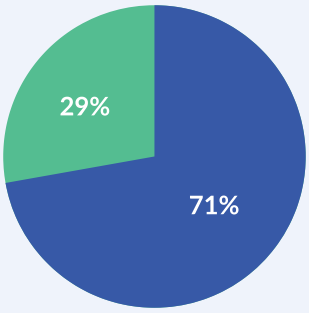
65%

experienced or were diagnosed with symptoms of depression after they were diagnosed with AMN

Of those 65%...



59% were seeing a psychotherapist or counsellor, or had done so in the past



71% were taking antidepressant medicine(s), or had done so in the past

Comments on how participants first described their AMN symptoms to a healthcare professional:

> My legs don't feel like mine. I walk very stiff and jerky. I struggle to run as my knees don't bend very well.

> Like dragging sand bags around on the ends of my legs.

Misdiagnosis

46% had been misdiagnosed in the past. Their doctors thought they had multiple sclerosis (MS), hereditary spastic paraplegia(HSP), motor neuron disease (MND), and/or mitochondrial cytopathies.

Comments on the role of healthcare providers:

> I am concerned that pressures on the NHS, especially shortage of trained medical staff is not conducive to the in-depth, systematic enquiry that is needed when a patient presents with vague-sounding early symptoms.

> Although I have a fantastic GP, I would love to see the hospital specialists adopt an integrated approach to managing this multi-system illness and improve communication amongst themselves.

> Now I have a diagnosis, the health care [team] have stepped aside. It would be nice to have more regular appointments rather than now just once a year. Seems I'm left to just deteriorate and deal with it myself.

Genetic counselling

More than half of participants (58%) have had genetic counselling. The benefits of genetic counselling include not only improved understanding of the genetic risks to individuals and their families but, in some cases, the possibility of new diagnoses.

Comments on genetic counselling:

> My mother got diagnosed with AMN and my daughters had their carrier status confirmed. Genetic counselling was offered to other family members too.

> Felt reassured that if we needed any more information from [the genetic counsellors] that we can call any time. Helped us really understand the genetic side of things, how it will affect our children.

Treatment

- Respondents took a range of medicines to manage their AMN; most commonly steroids
- Most needed walking aids and nearly one-third used a wheelchair
- Only 27% of respondents felt their symptoms were well-controlled
- Respondents most wanted to see improvements in:
  - Bowel and bladder incontinence because of the anxiety and embarrassment associated with these problems
  - Leg stiffness as this makes moving around and walking more difficult
  - Balance problems as this can cause falls

Access to specialists

- 55% of respondents did not have an annual MRI scan of their brain

- Nearly two-thirds of respondents were seeing an endocrinologist (a doctor who specializes in hormones) and around one-third were seeing a physiotherapist, occupational therapist or orthotist (a doctor who specializes in the mechanics of the body)
- 85% agreed or strongly agreed that they feel able to talk about their AMN to healthcare professionals

Family, social and working life

- 68% were worried about how AMN affected their family and only 34% felt able to talk about their condition with family and friends.
- 50% were able to socialize with family and friends, but only 10% were as active as they wanted to be.
- Around 60% of respondents agreed that they were able to work (full-time or part-time), although 70% felt their job options were limited by their AMN.

Future challenges, including potential for cerebral involvement

- 70% were worried about how their AMN would affect their family in the future, and 61% were pessimistic about their future.
- They were concerned about losing the ability to move and walk, being unable to work or progress in a career, and socialize with family.
- They were afraid of losing their independence and of uncertainty about their future.
- Cerebral involvement came across as an important theme; survey participants may have felt they were experiencing cerebral symptoms, or they were concerned about the prospect of developing cerebral involvement in future.

Comments on future challenges living with AMN:

> On a personal level, the biggest challenge would be the future loss of independence with regards to mobility, thereby affecting my ability to work full time.

> Losing the ability to drive, losing the ability to work, becoming dependent for my basic care needs and not being able to have sex.

> How quickly will I get worse? Will I die? What about my family? Will I see my three-week old baby grand-daughter get older? Will I be able to pay the bills?

> Still not knowing progression rates and what it will do to you physically and mentally

> No respite from the pain. Becoming a burden to my family as I'm unable to care to my needs

Comments on cerebral involvement now, or the fear of it in future:

> I have lost 3 uncles and a brother to this illness. I have cerebral involvement. Every day is a challenge.

> Legs getting worse and brain lesion getting active

> When I have to use a wheelchair full time. If disease goes to my brain

What does the AMN community need?

- The people who took part in the survey would like to see greater awareness of AMN amongst healthcare professionals and continued efforts to prioritize ALD for inclusion in neonatal screening.
- They wanted more information and advice for patients and families, including updates about potential treatments, and clinical trials for which they could be eligible.
- They asked for better contact with other people with AMN and their families – for mutual support.
- They want better communication and more joined-up working between local specialists so that requests for support are followed up and recommendations put into practice.

Was this report helpful?

Did you find the information in this report useful? Email info@alextlc.org or hello@raremark.com to tell us what you think. We'd love to hear from you.