Calls to screen all Scots babies after deaths due to ‘Lorenzo’s Oil’ condition

EXCLUSIVE

Charities and those living with rare conditions plead for NHS testing for rare disorders that could save hundreds of lives

By Martin Williams

DEMANDS are being made to help save hundreds of lives lost each year to rare diseases – by having better access to newborn screening following the deaths of boys suffering from a rare nervous disorder made famous through the film Lorenzo’s Oil.

One rare diseases charity supported by a Scots grandad who is living with an incurable life-threatening genetic disorder says that changes should be made to save lives and relieve unnecessary suffering.

An analysis by Genetic Alliance UK says the NHS is lagging behind other countries and currently gives newborns a blood test to screen just nine conditions – while more than 20 European countries are looking out for more with the majority seeking to detect 20 or more illnesses. The US screens for more than 50.

Rare diseases are collectively common and it is estimated that around 412,080 people in Scotland have a rare disease. They can be life-limiting and life-threatening, and disproportionately affect children. Some 75% of rare diseases affect children and more than 30% of children with a rare disease die before their fifth birthday.

Alex, the Leukodystrophy Charity (Alex TLC) has tried in vain to have adrenoleukodystrophy (ALD) added to the UK newborn screening programme.

Since an attempt was made through the UK National Screening Committee (UKNSC) in 2017, the charity says it knows of 29 boys who have been diagnosed with untreatable childhood...
cerebral ALD and nine have died.

**ALD symptoms**

ALD is a degenerative neurological disorder that results in chronic and progressive symptoms such as loss of vision, progressive dementia, learning disabilities, dysphagia (difficulty swallowing), seizures, deafness, lack of co-ordination and balance, fatigue, intermittent vomiting, weight loss, lack of appetite, nausea, darkening of the skin, muscle weakness, low blood sugar, and headaches in the morning.

It is a genetic condition that may be inherited from one or both parents. It most severely affects males when it can either emerge during childhood or adulthood.

Women who are carriers for ALD develop a milder form of the disease during adulthood. The three main types of ALD cause different severity in symptoms. In childhood cerebral ALD (CALD), symptoms start appearing from ages four to 10 and progress rapidly. Prognosis for these patients is poor, with death occurring within five to 10 years of diagnosis if left untreated.

The condition was featured in the 1992 movie Lorenzo’s Oil starring Nick Nolte and Susan Sarandon, which garnered two Oscar nominations.

The charity says it is “logical” that newborn screening could be preventative and that the system should be more open to more screening of rare diseases.

Its campaign is being supported by Michael Conway, a 56-year-old BAE Systems estimator from Renfrew who says his life would have been very different had his family had access to early screening.

There is further hope for ALD sufferers with stem-cell treatment that can help if the condition is caught early enough.

He said: “Newborn screening does not happen in Scotland or the UK. They wait until you get the symptoms or if it is in your family then they test you.

“If I was screened, looked after early, received stem-cell replacement (if required), my late onset ALD at 49 years old might never have occurred.

“I can only guess if screening did happen they would’ve taken the necessary action to make things easier for me when I was newly born.”

**Expert advice**

SCOTTISH ministers set screening policy for Scotland in light of expert advice from the UK National Screening Committee and the Scottish Screening Committee.

It is the UKNSC that sets the criteria for screening programmes.

Its guidance says that for a condition to be included in the screening programme it “should be an important health problem as judged by its frequency and/or severity”.

There is concern that this discriminates against rarer diseases such as ALD.

Louise Fish, chief executive of Genetic Alliance UK, stated: “We want the UK to make the most of opportunities to improve the lives of babies and their families where screening might be possible for a specific rare condition. The newborn bloodspot ‘heel-prick’ test given to every newborn baby in the UK screens for a maximum of nine conditions but there are more than 20 European countries screening for more than the UK, with most of those screening for 20 conditions or more.

“A great example is spinal muscular atrophy where babies can now be given life-saving and life-changing medication before symptoms appear and irreversible motor neuron damage is done. But we’ve been slower than other countries to start the process to build a screening programme for SMA, and this urgently needs to change.”

Mr Conway, who celebrated the birth of a third grandchild in September and has a grown-up daughter Rachel and son Matthew, says that his diagnosis means members of his family are screened at an early age and are being continuously monitored by their neurologist as there is a 50:50 chance they will be severely affected.

He became unwell in 2010 when he started feeling tingling and pain in his right arm and calf, and his balance faltered. He had some five years of testing before he found out he had ALD and said he was originally misdiagnosed with multiple sclerosis on Christmas Eve 2015.

He said: “I had late-onset ALD and wasn’t diagnosed until I was 49.

“By that time, my daughter was about 25. Would I have made different reproductive choices had I known earlier? That is a hard and probably pointless question for me to answer.

“Had I been tested ALD-positive on the day of my birth on December 29, 1966, and they took immediate steps, maybe I would not be living the poor life quality I am now.

“I have chronic fatigue every day. On waking every morning, I feel as if I have been through 10 rounds with a champion boxer. I have not had a good night’s sleep for over eight years.

“My balance is severely affected and I have to walk with elbow-crutches or I will fall on my face. There are also concentration and speed of processing issues which affect my life, in and out of work.

“Had I received newborn screening, early steps could have been taken to treat me straight away and steps taken to relieve the situation.

“I know from my cousin’s little boy that help and monitoring started the minute he was tested at a very young age. He is about seven or eight now and is being looked after by neurological services. They are trying to do what they can in lieu of newborn screening while he is still young as this disease kills young boys.”

His father Alexander passed away in 2002 after battling motor neurone disease (MND).

Mr Conway, who said BAE Systems has “taken great care of me” by providing a new heritage role in its UK shipbuilding sector, added: “I agree that detecting and treating as early as possible can make a huge difference and save lives.

“I’ve made friends with an Irish family who’ve been severely affected by ALD. Their children are adults now, but the oldest boy wasn’t screened at birth and he has been forced to live a life in a vegetated state, is blind, can’t walk and is really suffering.

“The next boy then got tested immediately and has received stem-cell replacement at Great Ormond Street Hospital and is living a normal healthy life. Their mother is in a wheelchair and has a hard life.

“But I have quite a positive attitude to my disease and treat every day as if it’s
Evidence concerns

SARA Hunt, chief executive of Alex TLC said there is screening in the US, Italy, Netherlands and Taiwan but says the UK won’t yet accept the evidence it’s beneficial.

The rejected application for newborn screening was reviewed in 2021 and rejected again – it is up for re-examination again in 2024/25.

She says that people should not be only capable of accessing life-saving treatments if they are “lucky enough” to have been identified early.

“If you don’t treat males early enough [with ALD] they will die – newborn screening is the only way to detect early unless you’re ‘lucky’ enough to have someone in your family already diagnosed,” she said.

Her older son, Alex, was diagnosed at age seven, lost all function by eight, and died at the age of 19 after 12 long years of what she calls “catastrophic neurodegeneration and complex disability”. Due to his diagnosis, his younger brother, Ayden, was also diagnosed, aged one at the time.

As a result he was monitored closely and had a successful bone marrow transplant at the age of eight. She says he is now in his 20s and living a normal life, relatively untouched save psychological scars.

“Once a boy/man develops symptoms of cerebral ALD that can be seen it is too late for treatment and they will deteriorate to a state of complete dependency: blind, tube fed, unable to move or communicate,” she said.

“The only treatment is a bone marrow transplant which is only successful if done before you can physically see symptoms. Males at risk are screened by MRI scan which will identify ALD progression before symptoms can be seen,” she said.

A Scottish Government spokesman said: “We recognise the profound impact an adrenoleukodystrophy diagnosis can have on both children and their families, and understand the urgent desire to receive treatment as early as possible.

“All UK nations receive independent advice from the UK National Screening Committee which evaluates the benefits and harms of all new screening programmes.

“When it last considered screening for ALD in 2021, it concluded that there were significant limitations around evidence for a number of issues including the impact of earlier diagnosis and the accuracy of screening tests. It therefore continues not to recommend newborn screening for the condition.

“It will look at any new evidence as part of regular reviews of its decisions. If it were to recommend a screening programme, we would seek advice from Scottish screening organisations on how it could best be rolled out in Scotland.”

Consequences

A DEPARTMENT of Health and Social Care spokesman said: “Although rare, adrenoleukodystrophy (ALD) is a devastating genetic disorder, and our sympathies go out to anyone having to deal with the consequences of this condition.

“The department is guided by the UK National Screening Committee, an independent scientific advisory committee, which reviewed the evidence for ALD in 2021 and made the recommendation not to introduce routine screening. It made this decision based on there being limited evidence on the outcome on treatment and accuracy of screening tests.

“The committee will review the evidence for this condition again. In the meantime, should any new published peer-reviewed evidence suggest a change to the committee’s recommendation then we would encourage this to be submitted via the committee’s annual call process as an early update.”

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Above,
Michael Conway, a 56-year-old BAE Systems estimator from Renfrew says his life would have been very different had his family had access to early screening for ALD

Inset, the movie Lorenzo’s Oil
The NHS is lagging behind other countries and currently gives newborns a blood test to screen just nine conditions. Picture posed by model. Picture: Getty
Below,
Louise Fish,
chief executive of Genetic Alliance UK wants the UK to screen babies for more rare conditions