

Project to find test for epilepsy in newborns

Lyndsay Moss

A NEW £10 million research project in Scotland is to lead worldwide efforts to detect and treat epilepsy from birth.

Some children with the condition suffer severe brain damage before they are diagnosed.

The project hopes to develop tests which can be done as soon as a baby is born.

The research centre, to be based at Edinburgh University, has been made possible by a £1m pledge from the Muir Maxwell Trust. The charity, which works for those affected by paediatric epilepsy,

was established by Ann and Jonny Maxwell after their son, Muir, was diagnosed with the condition. Now 12, he needs 24-hour care after a series of seizures damaged his brain. He was six before Dravet syndrome, a severe form of epilepsy, was diagnosed.

Ann, from Eskbank, believes early diagnosis of children with epilepsy is key to improving their future lives.

"The earlier you can identify these conditions and prevent the first seizure happening, the more likely it is you can stop it evolving and give that child a chance at normal life."

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Late diagnosis: Ann Maxwell did not find out that her son, Muir, suffered from severe epilepsy until he was six. Photograph: Julie Bull

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Seize the day

Dramatic breakthroughs in genetic medicine have come too late for Muir Maxwell, who was born with severe epilepsy, but a research trust set up in his name is already producing earlier diagnoses and could ultimately lead to a breakthrough

WORDS RUTH WALKER PHOTOGRAPHS JULIE BULL

ANN MAXWELL admits she would have been just another corporate wife, enduring a social whirl of coffee mornings and cheese and wine parties, had fate not dealt her family a devastating blow. Twelve years ago her son Muir was born, to all intents and purposes a beautiful, happy baby boy. But a series of seizures throughout his early childhood ravaged his brain to such an extent that even now, almost a teenager, his communication skills are limited to those of a child aged around three and he will need 24-hour care for the rest of his life.

It wasn't until the age of six that he was eventually diagnosed with Dravet syndrome, a severe form of epilepsy. But because it is most active in the first five years of life, for Muir at least, the diagnosis came far too late. Since then, however, his mother has dedicated herself to ensuring other families receive the support and early diagnosis they were denied.

The Muir Maxwell Trust's initial ambitions were modest, but five years and £3.5 million in fundraising later (coffers boosted, in part, by Christopher Biggins' winnings from *I'm A Celebrity...*), the charity

is about to enter into a ground-breaking collaboration with Edinburgh University, which will see the establishment of a world-class chair in paediatric epilepsy.

"This project is in Scotland but it could have been any university," stresses Maxwell, "it just happened that Edinburgh was receptive and ready. It shone brightly and has a lot of the right expertise."

The charity intends to raise the initial £1m to kick-start the £10m project, and will be applying to international bodies for further funding. "It will very much be a centre of excellence in the world of paediatric epilepsy. This is about taking cutting-edge research that happens across the world, bringing that to Edinburgh and taking it to the next level."

When she was initially approached about the project she was reticent, but is now convinced the chair will have tangible, practical applications. "The onset of the condition happens before we can diagnose it," she says. "What we're hoping to achieve with the university is to get in so early that it doesn't evolve; you diagnose almost at birth. In an ideal world we'd be testing in the womb, in the way they do for Down's

syndrome. But certainly, we would hope they would start testing at birth – the heel prick is a great test not just for epilepsy but other genetic conditions. I would really hope – and the university has expressed interest – that the heel prick test is one we would start exploiting."

The collaboration will involve a building at Little France, which will sit alongside the new Sick Children's Hospital due to open there in 2013. "It is a Scottish project, but it will be working with Yorkhill, Dundee, Aberdeen, Great Ormond Street, Australia, France, the USA – it won't achieve its endgame if it doesn't because there will be other things happening in the world. Within five years we expect to have helped change the face of childhood epilepsy beyond what we see today."

Muir's breakthrough diagnosis came as a result of major discoveries in Australia in the field of genetic epilepsy. But because there was just one team analysing DNA samples for all kinds of genetic conditions from all over the world, the results took two and a half years to come back, by which stage much of the damage had already been done. Now, having funded a similar team at



Yorkhill in Glasgow, dedicated to children in the UK, families are guaranteed a result in 30 days.

Finally knowing what was wrong with Muir meant Ann and husband Johnnie, who works in private equity, and doctors could start getting his drug regime under control – a long process of trial and error, complicated further recently by the onset of puberty and raging hormones.

“We now have the right treatment for Muir and we have his seizures under control, but we also have the legacy of those ten years which ravaged his brain and he’s a teenager now and you can really see it,” says his mother. “In the beginning he just looked like a beautiful baby boy, then progressively you can see the damage it was doing, until you get this 13-year-old boy who is good-looking, fast-growing and gangly ... but damaged.”

She still sees the boy he might have become when she looks at his two brothers, Connor, 15, and Rory, ten. And he has good days and bad days. On the good, he’s happy and cooperative. But because his communication skills aren’t what they could be, he has to find other ways of making himself understood. On a recent walk along Portobello promenade, he didn’t want to leave until he had thrown twigs in the water. “He decided he wasn’t going back to the car and I couldn’t go on, I had a really sore ankle, so I was holding on to the back of his jacket. He is so strong, and he was screaming, ‘Hands. Off. Hands. Off’ at the top of his voice. When I wouldn’t give in he just turned round and went – whack! And I got full thrust of his hand on my face. It was so painful, my face was numb. I was mortified. People were walking past all the time thinking, ‘She has no control over that child.’ And I don’t when he’s like that.”

However, she expects to continue to see him develop in small ways. “He can count, but we never thought he’d be able to do that. He now recognises one or two letters, but if you’d asked me two or three years ago if I thought Muir would be able to write his own name I’d have said definitely not; now I think we will achieve that.

“Emotionally and hormonally he’s 13. He gets all the physical urges boys that age get,

he just doesn’t understand them,” she adds.

In the cruellest of twists, however, just as she was getting Muir’s condition under control, Maxwell herself was diagnosed with an incurable, extremely rare brain tumour. “Basically it’s bone cancer in the head. I’ve had two major surgeries that have removed the threat for the time being and as far as we know for the foreseeable future.”

Still, she’s not bitter. “I’ve never questioned the why of Muir; I’ve never questioned the why of me; I’ve just accepted both of those and I can’t explain why. I’m a firm believer in the fact that the course of your life is decided for you and I accept that and I think that Muir came to us for a reason and possibly it’s because of what we’ve done with the charity.

“It’s been my catharsis; my way of coping with it all. It’s the concept of a glass half-full or half-empty. I suppose had I not set up the charity I might have spent a lot of time thinking about what might have been and perhaps feeling a bit more resentful at the authorities, whereas to set up the charity provided me with a focus and allowed me to do something about it.”

Recently, however, she has taken a step

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back to dedicate more time to her “neglected” family; she is still the charity’s figurehead and speaks regularly at events but a hand-picked team now takes care of the day-to-day business.

And she admits the business of fundraising has never been tougher. “My supporters have largely been from the financial sector and the financial sector is, of course, having a really bad time. We’ve now broadened our outlook and are sending out applications to charitable foundations and that is where I believe our focus will have to be. I have to believe it will get better



because otherwise what was the point?" The DNA service at Yorkhill has now been in place for four years so it is still early days but, says Maxwell: "I'm sure that children will be much less damaged by their condition than they would have been had they not had their seizures under control."

One of the greatest coups has been seeing knowledge improve so much among paediatric neurologists that samples are now arriving from children just months old. "Only time will tell if they will be mainstream school candidates, but a lot of them are."

Her ultimate dream, she is now ready to admit, is to see the eradication of genetic epilepsy. "That has to be the dream. Last year a headline attached to an interview with me said, 'I want to find a cure for epilepsy' and I was a bit taken aback. But I've come a long way since then and I'm happy to stand by that statement - even though I don't believe I ever made it."

As for Muir's future: "When I received his first diagnosis, I was told he was likely to die in the second decade of his life. That absolutely broke my heart. Now I don't believe that. I think we've achieved far too much for that, and I think the doctor who told me that would agree. Also, we now know Muir to be an incredibly robust little boy. He's coped with a lot more than we expected him to."

"The future's not bleak; he loves life and he's the happiest child in the world, though he has his off days like anyone else. I would like to think he might live for a reasonably long time."

To find out more about the Muir Maxwell Trust or to make a donation see www.muirmaxwelltrust.com





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MAX FACTOR (Above) Muir Maxwell, now a teenager, with his mother Ann, who has spent the past 13 years fundraising; and (right) as a toddler

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