HINTS AND TIPS AUGUST 2012 from Roland Lewis Editor 01923 720198

The social and educational effects of caring for a parent with MND

Olly Clabburn
Last year, we helped Olly Clabburn to advertise an opportunity to take part in his dissertation research project on our website and in our monthly membership magazine Thumb Print.

To give you some feedback on what drove him to study the social and educational effects of caring for a parent with MND and his key findings, he's written us a guest blog:

When I was seven years old my Dad was diagnosed with Motor Neurone Disease. At the time, I found it hard to understand why he had to stop doing the normal 'Daddy things'. He stopped going to work, began speaking in a slow and strange way, and then had to give up his car which was extremely hard for him.
Gradually over the three years in which we looked after him at home, myself and my family became full-time carers for him. This involved helping him when he fell over, getting him drinks, food, toileting amongst a plethora of other things. Yet my friends at primary school seemed to be having a very different life at home while I was doing a variety of things for my Dad which I believed to be 'normal'.
As my Dad began to deteriorate more, caring at home became more and more challenging. Consequently, he moved into the Hospice for the final few years where he could get the specialised help that was now required. Although the Hospice staff were amazing for my Dad and family, the inevitable happened in April 2004 when he passed away after a long battle with MND.
Years passed and after studying Psychology at Sixth-form, I developed a keen interest to how we work as people and why we 'do' certain things. I then enrolled in Lancaster University to study Psychology in Education for which I conducted a dissertation research project. Fuelled by my experiences, I decided to further investigate young-carers and their experiences caring for a parent with MND. My project subsequently was titled 'the social and educational effects of caring for a parent with Motor Neurone Disease'.
Upon deciding to research this specific area, I knew recruiting participants would be challenging with MND being so rare and not generally considered to impact upon young-people's lives. I therefore established communications with the MND Association in the hope for some advice or guidance. Ultimately, the association made my research possible as without their assistance, I simply would have not been raise awareness of my study and conduct the research.
The MND Association allowed me to publish a letter in the Summer 2011 edition of Thumbprint outlining my study and need for young people who were once young carers. The Association later added a webpage under the research section of the Association's website which also assisted with recruitment.

As a result of the magazine and webpage, I managed to recruit and interview 7 participants who had once, or currently were, caring for a parent with MND.

Upon writing up my research, there were 6 clear themes raised by the participants which were considered to be the main social and educational effects of caring for a parent with MND.

1) DIAGNOSIS: Many of the young carers felt somewhat confused and unaware about MND and what their parent being diagnosed actually meant. Consequently, it often came as quite a surprise when a parent's care needs increased. It was also found that the terminal nature of MND was often hidden from the young-people in an attempt to shelter them.

2) YOUNG CARER DUTIES: One of the key duties a young person adopted after the diagnosis, was increased responsibility for household chores enabling their healthy parent to spend more time with the MND patient. It was also noted that the young people tended to adopt a more 'social care' role, meaning they would often sit with their parent and keep them company rather doing the more intimate caring tasks.

3) RESPONSIBILITIES: Older siblings tended to adopt a more parental role for younger siblings by helping out with school runs, help with homework or carrying out more caring tasks for the ill parent to shelter their younger sibling. It was also noted that all participants had a greater appreciation for their healthy parent and a closer relationship as a result of MND.

4) EDUCATION: All participants emphasised the importance of education (school/college/university) providing a period of escapism. This meant that for the time in which they were in the educational setting, they could temporarily forget about life at home and be 'normal'. Interestingly, it was also noted that having a parent with MND brought some educational benefits. For example, their parent being permanently at home provided an opportunity to help with homework. It was also commonly acknowledged that the disease/bereavement fuelled a great deal of motivation for the young person to achieve educational success.

5) SOCIAL: It was noted that peers and friends provided another extremely important method of escapism. Participants found that they could gain advice or simply ease the burden by discussing life at home. It was additionally noted that peers may introduce the young carer to new hobbies and interests which also allowed the individual to escape or channel emotions. However, it was also outlined that guilt was also a common feeling when with peers and not at home with their parent.

6) POSITIVE ASPECTS: Overall the participants in the research outlined a variety of positive aspects that they have drawn from the experience. Most notably, a feeling of maturity compared to peers, the ability to accurately empathise with others, closer relationship with family members and increased motivation leading to educational success.

Finally, it was noted that a diagnosis of MND is inevitably traumatic and creates many negative outcomes for all involved. The research however aimed to reinforce the idea of optimism thus coinciding with the '**MND Month for Optimism'** campaign.

Young carers will spend much of their time caring for their terminally ill parent and later suffer bereavement. Nevertheless, the research highlighted the positive benefits that individuals can gain from this known negative experience.

Young carers will spend much of their time caring for their terminally ill parent and later suffer bereavement. Nevertheless, the research highlighted the positive benefits that individuals can gain from this known negative experience.

Kelly Edwards

Profilin 1 identified as a cause of inherited MND

by Kelly Edwards

An MND causing gene called profilin 1 has been identified as the cause of about two percent of cases of inherited MND. This finding provides new insights into the causes of MND and suggests a potential role of the cellular scaffolding in MND. The finding was published in the 15 July edition of the prestigious journal Nature. This international collaborative study was led by Dr John Landers, at the University of Massachusetts Medical School, USA.

Using cutting-edge genetic technology, Dr Landers and colleagues first identified genetic mistakes in the profilin 1 gene in two families with inherited MND. To verify these findings, they went on to identify five additional families that also have mistakes in the profilin 1 gene. They did this by examining the genetic spelling of this gene in 272 further people with inherited MND (with no known genetic cause). This means that the genetic mistake could account for approximately two percent of cases of inherited MND.

Four different genetic mistakes were identified in the profilin 1 gene in the seven identified MND families. Three of these genetic mistakes were not found in any healthy controls, which mean that these mistakes are most likely a direct cause of MND. The forth genetic mistake was identified in a small number of healthy control samples, which could mean that this mistake could be a less significant cause of MND.

What does profilin do?

Profilin plays a vital role in maintaining and shaping the cells scaffolding – the cytoskeleton.

The cytoskeleton can be thought of as being made up of stacks of Lego bricks, called filaments. To maintain the shape of the cell, these bricks push against the cell membrane. To stretch and move the cell, more bricks (called actin) are added to the outermost end of the filament, which forces the membrane to extend. Toward the innermost end of the filament, the actin units separate, similar to pulling off individual bricks from the bottom of a stack, where they're then collected and attached to profilin. Profilin then recharges and recycles the actin units, so that they're ready to be added to the top of the filament again.

What did the research group find?

Through this study, the research group identified that the ability of profilin to attach to actin is affected by the genetic mistakes, making it 'clumsy'. They also identified that the mistakes affect the ability of the cells to grow, which could be an attributing factor to how these mistakes can cause MND.

In this study, the researchers also confirmed that profilin is normally found throughout the 'factory floor' of the cell, the cytoplasm. However, when profilin is faulty the research group identified that it often assembles into clumps of protein marked for destruction – a hallmark of MND.

Interestingly, they also identified that when profilin is faulty, TDP-43 also clumps together. This suggests that faulty profilin may also cause MND through its effect on TDP-43. It's also worth noting that when TDP-43 is faulty, profilin is not found within the clumps of faulty TDP-43 suggesting that profilin has an effect on TDP-43 and not vice-versa.

What does this mean for people with MND?

Profilin 1 is the twelfth MND causing gene to be identified in MND, which means that we are one step closer to knowing all of the genetic causes of MND. Learning more about how genetic mistakes can cause the rare inherited form of MND (5-10% of cases) helps us to learn more about all forms of MND as the more common sporadic form is clinically

indistinguishable to the inherited form.
As this genetic mistake is thought to only be attributed to a small number of families with MND, it is currently unknown if a genetic test will be developed for inherited MND. If you have inherited MND and want to find out more information about genetic testing, please speak with your doctor or neurologist.

What does this mean for the future of MND research?

These findings will need to be verified in larger numbers in different populations to determine a more accurate figure for how many families are affected by mistakes in the profilin 1 gene. More work will also need to be done to determine how the cytoskeleton is affected in MND and whether it can provide any therapeutic targets to treat MND in the future.
In summary, problems with the cytoskeleton have long been thought to be involved with MND, but having a direct genetic cause of MND strongly associated with the cytoskeleton will most likely reignite this avenue of research in the coming years.

Researcher Dr Martin Turner awarded ENCALS Young Investigator Award

Motor Neurone Disease (MND) Association funded researcher Dr Martin Turner has been awarded the prestigious European Network for the Cure of ALS (ENCALS) Young Investigator Award.The award was presented on 27 May 2012 at the ENCALS 2012 meeting in Dublin.

The award recognises the most promising researcher under the age of 40 who, in the opinion of the ENCALS panel, has generated research that is the most outstanding or innovative in furthering the understanding of MND.

Prof Ammar Al-Chalabi, Chair of the ENCALS Award Committee, said: “This award is prestigious because the competition is open to anyone, so it is a truly international award, and the winner is therefore among the most outstanding young researchers in the world in MND.”

“The ENCALS award panel were particularly impressed by Dr Turner’s MRI work, biomarkers study and creativity in exploring new ideas about MND.”

Dr Turner was awarded with the MRC/ MND Association Lady Edith Wolfson Clinical Research Fellowship in 2008 for his study to identify biomarkers in MND. Since then, Dr Turner has already published two findings from his five-year disease marker study in the prestigious journals Neurology and Brain. Using advanced brain scanning technology, his study has identified a common pattern of nerve damage in the brains of MND patients. This holds the promise of a much-needed disease marker.

Dr Martin Turner commented that, “The ENCALS award marks a major highlight in my career. I am passionate about MND, and feel privileged to help care for those living with the most challenging of diseases. To be recognised as having made a useful contribution to research as well, by international leaders in the field, means an enormous amount.This award will strengthen my bid to become an independent research leader, and develop the next generation of neuroscientists.”

The MND Association's Director of Research Development Dr Brian Dickie commented, "We're delighted that one of our Lady Edith Wolfson Fellows has won this prestigious international award."

"The Fellowships were created to attract and retain the brightest and the best young clinicians to MND research and it is a fitting tribute to the knowledge, expertise and dedication that Dr Turner brings to this important field of MND research.