

Thumbs UP

MND/ALS
Association of South Africa

The Newsletter of the
Motor Neurone Disease /
Amyotrophic Lateral Sclerosis
Association of South Africa

May / June 2016



Our two Consultants in Gauteng, Victoria Goodstein (left) and Karyn Casey, at the MND/ALS display at the 2016 NASA Congress (Neurological Association of South Africa) which they attended in March in the Drakensberg

The Mission Statement of the MND Assoc. of S.A. is
"To provide and promote the best possible support for people living with
Motor Neurone Disease, their families and carers and to raise public awareness."

MND/ALS of South Africa is a member of the International Alliance

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ACKNOWLEDGEMENTS

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Time passes so quickly - we are already half-way through the year! and we report on what has happened since our last communication and what is planned:

As you know, **21 June is MND Global Awareness Day**

And it was suggested that a 2-day Awareness Day Conference be held, and the Stellenbosch Medical School at Tygerberg Hospital has been secured as the venue. Such an event takes a lot of hard work: a lot of organizing, a lot of commitment and a lot of patience! Sheila Kendal, our Vice-Chair, has undertaken this huge task and we are all supporting her in this mission. The conference is aimed at medical professionals and the pharmaceutical fields which would have bearing on MND and the cost is R1,500 per delegate. Invited Speakers from various avenues have indicated their willingness to participate. Exhibitors are also being sought. If you know of anyone who is keen to partake, sponsor or exhibit, please advise Sheila. This Awareness Conference is a first for MNDA of SA and we trust it will be well received.

DONATIONS

We are very grateful to each and everyone for the donations received since our last Newsletter, whether its a regular monthly payment, was in lieu of a gift to someone, or in memory of a loved one, we thank you for the kind gesture. We would also like to, in particular, mention the following:

- Lake Farming Partnerships for their donation of R25,000 in memory of Jean Bricknell, a MND patient from Port Elizabeth who passed away in November 2012.
- Estate of the late Dehlia Rezek, who passed away in November 2013, for the bequest of R50,000. (We have in the past received donations from Burlington Stables in memory of Dehlia).

A BIG THANK YOU for these wonderful contributions the result of which enables us to continue the care and support of our patients

ROUND AND ABOUT

Port Elizabeth

Sheila Kendal, our Vice Chair and Senior Consultant, visited Port Elizabeth in February where she conducted a MND support group meeting and visited MND patients who were unable to get to the meeting.



Mandy Gurr Snyman, a MND patient for 18 years, (above left) had recently moved to a care facility and Sheila popped in to see how she was settling in.

Pretoria, Cape Town (& Oudtshoorn!)



Left picture: Francois Serfontein with his son Werner and Joost van der Westhuizen

Right picture: From left (standing) Werner Serfontein (Francois' son) and Linza Oosthuizen (Francois' partner/carer), with Francois and Joost.

Previously from Oudtshoorn, Francois Serfontein, now living in Parow, Cape, in December won the “SPAR Christmas Wish” after his son Werner nominated him.

They left for Pretoria on 16 April and met with Joost van der Westhuizen, where they chatted about rugby and the disease which they both have. Francois was entertained at Joost's Loftus Versfeld Rugby Box where they watched the Bulls vs Redds match.

Francois has had MND for over 30 years and Linza Oosthuizen cares for him in their Parow home.

Durban

Great news from KZN !

Kasturi Pillay, our Consultant for the area, had reason to celebrate – she sent us this photo and writes as follows:



“It was a double celebration on 6 February, we both turned 50 in January, and we got married at Ridge Function Hall in Shallcross. It was a beautiful and memorable day and sharing it with a few of my MND patients was a cherry on the top for the day. Just to have seen them all dressed up and looking happy, and coming in a wheelchair, just made my day more special.”

The bridal couple donated R800 (part of the monetary gifts they received), to MND to help towards the care and support of our patients.

We wish them a long and very happy future together.

To the world you may be one person, but to one person you may be the world

RESEARCH



(Article in Spring 2016 issue of Thumb Print – MND Association, UK)

For the first time the MND Association (UK) is funding four non-clinical fellowships and two of them aim to identify possible genetic links in people living with MND.

Dr Ashley Jones works at King’s College, London. His fellowship builds on similar research he carried out funded by the ALS Association (USA) and the MND Association.

Dr Russell McLaughlin from Trinity College Dublin, has just finished a Post-Doctoral fellowship with the ALS Association.

Funding of their work emphasizes that developing researchers is an international effort, and that collaboration is key to keeping talented individuals working in MND research.

Their mission -

For approximately 5-10% of people living with MND, the cause of the disease is primarily due to a mistake within the genes. However, for the majority of cases of MND, genes are thought to play a more subtle role. However, it is becoming increasingly difficult to find these genes.

The current method of identifying disease-causing genes, called whole genome sequencing, involves comparing people’s genetic codes to identify similarities or differences. This has pinpointed some of the genes associated with MND.

This project may answer questions over if populations with a certain ancestry seem to be at greater risk of developing ALS than others.

Rare genes that might be responsible are harder to find, as these genes are almost invisible to traditional gene hunting methods. On average, every person has between 40 and 100 rare gene variants (versions of a gene). Because of their rarity, it is hard to work out which, if any, of these variants may be associated with disease. You could compare MND gene hunting to panning for gold. It is easy to spot the large gold nuggets by eye, representing the MND genes found by current methods, but you need to use a different technique to separate the small flecks of gold, the rare disease-causing gene variants, from the gravel and dirt, or the ‘normal’ rare gene variants.

Why do MND genetic research ?

There is now evidence that there is a significant genetic aspect to all MND, regardless of family history. Studying genes can help us understand their influence in the cause, onset, and the speed of progression of MND. It can also help researchers develop models for studying MND and ultimately in developing new treatments.

Project MinE

Project MinE is a global project hunting for MIND-causing genes. The UK arm of this project is using samples from the MND DNA Bank, and is one of the main research areas that received monies raised through the Ice Bucket Challenge.

So far 768 DNA Bank samples from people with MND, and samples from healthy people for comparison, have been sequenced, and a further 672 have been sent for analysis this year.

Russell's objective:

To identify the genetic causes of MND in specific populations

Russell will study the impact of ancestry on the development of MND.

Sometimes looking deeper into a family tree throws out unexpected relations, such as Benedict Cumberbatch and King Richard III being very distant relatives.

Russell will sequence the genetic code of over 1,000 Irish individuals, 700 of whom have ALS (amyotrophic lateral sclerosis, a form of MND). Using these, Russell will construct large family trees. It is likely that these will link ALS patients previously assumed to be unrelated. These 'superfamilies' will give researchers a greater chance of identifying rarer gene variants linked to ALS development.

Talking about this research, Russell said : "This project will help to answer questions about whether populations with certain ancestries might be at greater risk of developing ALS than others. It could also help explain if the geographical differences in risk of ALS seen throughout Ireland are linked to genetics, or down to local environmental effects. The sequencing of data from this study will also contribute towards Project MinE and will be used to discover genes that cause ALS".

Ashley's objective:

To prioritise rare genes to investigate further

Ashley will begin by creating a list of candidate MND-causing rare genes using information on how motor neurones develop, together with data from Project MinE. This will guide him towards rare genes to prioritise for further investigation, by studying post-mortem brain tissue and DNA sequencing.

Ashley explained : "My research will examine how the genome (the body's genetic instructions) interacts with itself in the motor cortex of the brain, and how this interaction contributes to MND. Identifying these interactions will give us promising candidates for gen-based therapies."

"Our invaluable partnership will defeat MND"

Not all people with MND show the same symptoms in a physical examination, and the rates of disease progression may also vary substantially.

However the reasons for this are poorly understood. The ability to measure disease activity and progression in MND requires the identification of biological markers – these are called biomarkers.

The Oxford Study for Biomarkers in MNF (BioMOx, www.biomox.net) began in 2009, funded through the UK MND Association's Lady Edith Wolfson Fellowship Scheme in conjunction with the Medical Research Council (MRC).

In non-therapeutic research like BioMOx, there is no direct benefit to the participants, because no new treatment is being offered. However, the enormous enthusiasm of patients, their friends, and family to actively contribute to the common goal of a world free from MND has been very striking to all the researchers working on this study.

Part one of the study involved over 70 participants with MND who underwent tests every six months for up to two years, along with a similar number of healthy individuals who participated on one occasion for comparison.

Through collaboration with Dr Andrea Malaspina's biomarker programme at Queen Mary's University London, BioMOx helped to confirm neurofilaments, which are breakdown products of nerve cells, as a leading biomarker for MND. The levels of neurofilaments in spinal fluid and blood show promise not only as attest to aid diagnosis, but also as a tool to monitor future treatment effects.

Biomarker development is not the only research area which depends on the involvement of people affected by MND. Patients donate skin samples to help with stem cell research, and give information about lifestyle and environmental factors which may affect the development of MND.

To all those who gave their time and effort so enthusiastically, and to those thinking of doing so in the future, thank you on behalf of the MND researchers across the globe who are all using the information gleaned to further the development of tests and treatments for MND. It is through invaluable research partnerships between scientists and those affected by MND that will defeat this disease.

Prof Martin Turner, Lady Edith Wolfson Fellow

This narrative was sent to us by Karyn Casey, one of our Consultants in Gauteng:

Some-of-the -time in Darrenwood

I miss not biting into a grape,
all the way from Fairest Cape
and it's truly not much fun
not to taste a hot cross bun.
at Easter time.... in Darrenwood.

Easter eggs and chocs are out of my reach
And also my favourite yellow cling peach.
Oh for the lick of an Ice Cream cone
Or an English strawberry-creamed scone.
At high-tea time.....in Darrenwood.

Food's chopped and mushed all together.
Soup is in, no matter the weather.
So bite your teeth and not the Bullet,
With all good things you sure can full-it.
at supper time... in Darrenwood.

It's no good to cry and moan
Just because you can't use the phone.
Just type a long and cheery letter,
A cup of tea will make it better.
At even tide ...in Darrenwood.

Put on the mask and take a pill,
Put on 7 0 2 if you will.
Pray that you will see the morn
Twittering birds stir the dawn
At sunrise time..... in Darrenwood.

It's sad I can no longer sing
In church when loud Hosannahs ring.
But Beethoven in deafness stirring music wrote,
I sing joyously in my heart not from the throat
It's praise time in Darrenwood

Each has her own cross to bear.
Is the future faced with fear?
No, in God's hands we are safely placed,
No matter the difficulties to be faced.
Till the end-time.....in Darrenwood.

Condolences to Family and Friends of:



Elizabeth van Rensburg (11/10), Christiaan van Zyl (24/11), Jacky van der Walt (1/12),
Johannes Pienaar (10/3), Aysha Salasa (13/3), Helm van Zyl (20/3),
William Mlungwana (27/3), Isabella van Rooyen (9/4), Raymond Trollip (7/4),
Marie Bezuidenhout (2/5), Mercia Marx (4/5), Barbara Lee (9/5), Beryl Goldman

**We welcome your ideas – *THUMBS UP* is
your voice –
So if you would like to Contribute to the next
issue – Please write to us !**

HELPING THROUGH YOUR WILL

*Your Will can be a convenient vehicle for
making a charitable gift of a lasting value.
Please consider MND Association as a living
memorial for a loved one.
Many people support the work of the MND
Association of South Africa
through bequests from their Estates.*

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