

MSA SMarT News

The newsletter of the Sarah Matheson Trust

Contents

SMT Nurses	1
Carers	3
Letters	4
Tips & Hints	4
Research News	6
Fundraising	6



Fun
Walk
'Base
Camp'

Diary dates	8
-------------	---

Next issue:

Ask the Experts

We have a panel of willing experts, doctors and therapists who are ready to answer your questions.

Is there something you always forget to ask or something that you think other readers should know about? Is there something you are embarrassed about asking either because it's personal or you think it's a silly question? Here is your chance to get the answer.

Send your questions to Alison in writing or over the telephone and look out for the next SMarT News where we will print the answers.

The Sarah Matheson Trust provides a support and information service to people with Multiple System Atrophy (MSA) and other autonomic disorders, their families and carers, health care professionals and social care teams. We also sponsor and support research into MSA

From the Nurses

We are not sure exactly when it happened but something has changed at the Sarah Matheson Trust. We have progressed from being a support group and have become a 'family'. This change has occurred in several ways...

- Members are keeping in contact not only with the advice line but with each other, just like a family. The Contact Scheme is bigger than we ever imagined and judging from your responses it is essential to many of you in reducing the isolation experienced with having an unusual disorder.
- Through the Contact Scheme and Support Meetings SMT family members are willing to share experiences and feelings. This is not always easy. Most people coming to a Support Meeting for the first time are apprehensive. However the warmth and sense of fellowship that can be felt in these meetings enables people to relax. Difficult topics are sometimes raised but the discussions that follow are helpful and beneficial and show the sensitivity of those involved.
- During these meetings members are accepting of each other, they are not phased by walking sticks, wheelchairs or lightwriters and take care to look after each other just like a family. And, like all families, the SMT family are not afraid to laugh in the face of adversity. A sense of humour is not impaired by autonomic problems and this can be clearly seen in our support meetings.
- Members are committed to helping everyone within the SMT family. Fundraising, increasing awareness and taking part in research are all activities that benefit the whole family and as you read SMarT News you can see evidence of this.
- The SMT continues to grow as more people are diagnosed with autonomic conditions supported by an increasing number of health and social care professionals who register in order to provide the best care.

To maintain and develop our services as we grow we will have the additional input from a part-time administrative assistant in the SMT office from the New Year. This will allow us to maintain our calendar of support meetings and fundraising events. It will also enable us to offer help and encouragement to groups like the East Midlands Support Group which is now up and running (see page 5). Most importantly it will allow us to remain close with family members which for both of us is the most important (and the best) part of our jobs.

As 2003 draws to a close it's inevitable that Christmas seems to surround us. We thought that this would be a good time to inspire the artists among you to design next year's SMT Christmas card: see competition details (page 2). Staying on the subject of Christmas cards don't throw this year's cards away in January; send them to us. We can put them to good use.

Finally, thank you to the many contributions from members to this newsletter. We feel that this adds colour and originality to SMarT News and really makes it a family newsletter.

With this newsletter we send our warmest wishes for a Happy and Peaceful Christmas. We look forward to speaking with you and perhaps meeting you in 2004.

Catherine & Alison



Test Drive this Guide!

The new Gowrings Mobility Smooth Ride Guide, contains useful information about access to places of interest throughout the UK. The Guide is designed to help you plan days out. The guide has information on a variety of attractions, how to get there, what facilities are available and even local accommodation all with an emphasis on the needs of a variety of disabilities. It follows similar successful publications for the USA Eastern Seaboard and Australia & New Zealand.

We are looking for volunteers to test drive the Guide by visiting places mentioned in the book and letting us know if the guide got it right. We can send you details from any of 80 regional areas in England, Scotland, Wales and Northern Ireland. Let us know how useful the information is and we can then let everyone know through SMarT news.

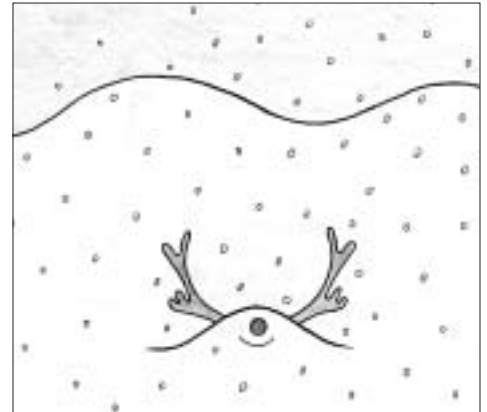
If you want to buy the guide, it is available at a discounted rate of £12.50 (normally £14.95) to readers of SMarT news. Order by ringing 0845 608 8050

Quality of Life questionnaire

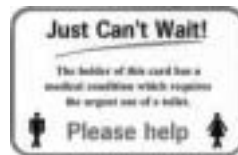
Apologies as the revised version of this questionnaire has taken longer than we thought to shorten and make user friendly without losing vital information. We will be posting them out to all members in the New Year and would be grateful if you could make some time to fill them in and return them.

SMT Christmas Card Competition

After the success of our Reindeer in the Snow Christmas card designed by Harry Fleming, son of one of our Trustees, we are now inviting you all to come up with something new for next year - we may even do more than one design. So all you artistic people out there please be inspired on our behalf. For the non-artistic amongst you, recruit your children, family or friends! Artwork in any medium - paper, photography or computer-designed - will receive equal attention. Original artwork can be returned if requested. All entries to be sent to Alison at the SMT office.



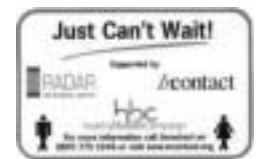
Just Can't Wait



RADAR (the campaigning and advisory body for disabled people) and Incontact (the national charity for people with bladder and bowel problems) have launched a credit card sized card, Just Can't Wait, which should help people gain speedy access to toilet facilities in shops and public places.

When shown to staff in shops, the holder should be given access to facilities there or directed to the nearest public toilets. John Lewis and the British Stores Association are supporting the initiative and it is hoped that news of the scheme will soon expand to improve toilet access when out and about.

Copies of the card are available from Incontact on 0870 770 3246 or info@incontact.org or from Alison.



Social Evening – 23rd February 2004

The Architectural Association in London, which had close connections with Sarah Matheson, will be the venue for a special gathering. Sir Roger Bannister, who is one of our Patrons, will welcome members and guests to an evening that will launch our new MSA folder, present the Trust's research work together with other achievements. Music and light refreshments will make this informal occasion an opportunity for members to meet many of the SMT team. If you would like more details, or to reserve a place, please contact Alison. Book early to ensure your place.

In Memory

Donations received with gratitude in memory of:

- | | | |
|-----------------------|----------------------|--------------------|
| • Florence Adams | • Paul Griggs | • Eric Loydall |
| • Rodney Bridges | • Camilla Grimsdell | • Patrick Markham |
| • Eileen Bromley | • Frances Hopwood | • Glenna Proudfoot |
| • Patricia Catchpole | • Roy Hunt | • Ian Russell |
| • John Forwood | • Joseph Jackson | • Martin Shaw |
| • Peter Foster | • Sara King | • Beryl Sonley |
| • Arthur Goldsborough | • Arnold Klausberger | • John Taft |
| • Paul Greensill | • Mario Leongson | • Douglas Willis |

Woman's best friend

Maureen Watkins appreciates her dog

Imagine my surprise when I was given an eight week old bundle of black fur, with adorable eyes and four big paws.

Our lives changed from that day. Sophie (named after an old jazz singer my husband likes), ruled and continues to rule the house! At first, she peed everywhere and didn't like being left alone. A bit like me! But we got through this difficult period; now, nine months on she is ready to begin her training to be an Assistance Dog. She will eventually be able to help me.

Soon after having her, I discovered various organisations that trained and supplied Assistance Dogs. Well, I thought Sophie was suitable and would respond to some training. DogAID came up trumps, as I found organisations like Support Dogs train their own puppies and expect you to "bond" with a fully trained dog, a procedure that can take several weeks, away from home in a residential place that suits them. Something I did not want; after all we had a puppy and I had no intention of staying away from home.

The appeal of DogAID is that a

trainer comes to your own home and trains your own dog. Obviously there are strict rules: they investigate the suitability and temperament of the dog, your vet has to give the dog a thorough health check and then they have to find a suitable trainer in your area.

At first, DogAID, which is based in Staffordshire, could not find a local trainer. But a local dog psychologist agreed to take us on.

We were lucky, so we could start. Sophie was eight months old and ready to learn. She passed the initial assessments and health check, so the training started in earnest!

The trainer comes about every ten days and puts us through our paces. Yes, there is much training for me and my husband as well as the dog! We then have "homework" to do before the next visit.

So far Sophie has taught herself to wait out of the way while I struggle down the stairs. She always sits patiently while



I get her food. She recognises the command 'touch' to nudge cupboard doors. This is a preparatory exercise to lead her into closing doors for me. She is learning to 'sit', 'lie down', 'leave it' and 'stay', and at the moment we are trying to discourage her from jumping up on visitors, something very hard for a nine month puppy. Visitors, despite a notice on our front door requesting callers to ignore

her, tend to want to make a fuss of her, stroke and talk to her, which does not help!

My husband has walking practice three times daily. We hope Sophie will eventually not be distracted by outside influences and be able to go out with me. Next week the trainer

has arranged for 'strangers' to come by, including two cyclists for her to cope with.

As I type this, Sophie is playing with my husband in the garden. Both are having a good romp. So you see, her spirit is not broken. When 'off duty' she is just this big sloppy Labrador. We think the world of her!

DogAID can be contacted at 25 Speechly Drive, Rugeley, Staffs WS15 2PT, www.dogaid.org.uk, or email joy@dogaid.org.uk

Patient and Public Involvement Forums These Forums will replace Community Health Councils in England and will be set up in every Primary Healthcare and NHS Trust. From September each Forum must have at least seven members, the majority of whom must be patients and carers. The Forums will consult patients and carers about local health services. If you have the desire to make a difference, the first step is to check out the Commission for Patient and Public Involvement website, www.cppih.org. This organisation is overseeing the development of these Forums.

Carers' Allowance An amendment from our eagle-eyed readers (thanks for the feedback) – the Carers' Allowance has no carer age limits and the earnings limit is now £77 per week after allowable expenses (e.g. childcare costs). Your savings are not taken into account for this benefit. For further information contact your local Social Services office, look on the website (www.dwp.gov.uk) or use the Benefit Enquiry Line (0800 882200).

Carers' Right Day – Friday 5th December Look out in your local newspapers for details of any events in your area to do with this Day. Events are being planned throughout the country to increase the uptake of benefits available for carers. For example, in Newcastle at Durant Hall refreshments, including soup and rolls, are available all day to carers who come in to find out more about the benefits and services available to them in Newcastle.

Carers' Line Due to funding difficulties, the availability of the Carers UK free national helpline (0808 808 7777) which offers advice on benefits, community care and other carers' rights has been reduced. The new hours are Wednesdays & Thursdays (10-12 midday, 2-4pm). An answerphone service will continue to offer a free Carers Pack for callers unable to get through to an adviser. Carers UK are seeking alternative funding, and hope this reduction in services will only be a temporary arrangement.

The Traffic Warden & his revenge!

I recently obtained a Blue Disabled Parking Permit, as walking has become much more difficult, but I can safely drive for the next few months at least. I therefore parked in a Disabled Bay and went shopping in the town centre.

On my return to the car I needed a few minutes rest before returning – so I switched the radio on, rolled down the roof of my Alpha Spider sports car, donned my sun glasses and considered my purchases.

There was a tapping on my car door and I looked up to see a Traffic Warden staring sternly at me.

‘Do you know this is a Disabled Bay sir’ he said. ‘Yes’ I answered. ‘Well are you going to be long’ was the

reply. ‘Probably another ten minutes’ was my retort.

‘Do you know this is a Disabled Bay sir?’ To which I replied rather testily ‘yes’. ‘Well have you got a Disabled Permit?’ I removed from my glove compartment the permit I had just put away and immediately his demeanour changed and he became cheerful, chatty and good-natured. Unusual qualities in a Traffic Warden – I am sure you will agree.

However, three weeks later he got his revenge...

Armed with my success of the Disabled Bay I tried to park there a few weeks later and found the Disabled Bay filled with four cars two of which did

not display a Disabled Permit. But the same bay was divided into two and the latter half was reserved as a Loading Bay. Rather than park on the other side of the road on the double yellow lines, as permitted, I parked in the Disabled Bay, displayed my badge and went about my shopping.

On my return, half an hour later, I saw the ominous yellow envelope glued to my window – a Parking Ticket. My appeal to the Traffic Department was turned down as they rightly pointed out that a Disabled Parking Permit may not be used in a Loading Bay. News to me, and a warning, perhaps, to fellow members.

“Hedgehog”, Kent

Tips & hints

A few words of advice

Next to my telephone I have a tape recorder with the following words recorded by a well spoken friend:

“Before we start our conversation please listen to the following pre-recorded statement. You are speaking to I have a progressive neurological condition called Multiple System Atrophy (MSA). This is a rare movement disorder, which affects balance and speech. Therefore, while it is difficult for me to speak to you clearly, my thinking skills are unimpaired! Be patient and talk to me normally and I will make myself understood.”

For those people whose speech like mine is a little unclear this has been very helpful in removing any embarrassment or the person on the other end of the phone jumping to the conclusion that the person they are speaking to is slightly drunk! I find that people are very patient and sympathetic when they realise that they are dealing with genuine disablement.

I also produced a little pre-printed card to give to shop assistants, bank clerks, and garage mechanics prior to gaining their assistance. Again this has proved very helpful in gaining an appropriate level of service. The wording reads:

“I should like to introduce myself, my name is I have a progressive neurological condition called Multiple System Atrophy (MSA). This is a movement disorder, which affects balance and speech. Therefore, while it is difficult for me to speak to you clearly, my thinking skills are unimpaired! – Please talk to me normally and I will respond the best I can. I may need a chair to sit on as we speak.”

“Hedgehog”, Kent

A good turning tip

We really enjoyed the last edition of SMarT News. Alex did have difficulty getting on the dining chair to sit close to the table, until I found a turntable (suitable for TVs etc. up to 100kg weight) in our local £1 shop! It’s turned out (excuse the pun!) to be the most useful thing we’ve had, just for £1, as he just sits on it then swivels round on it to face the dining table!

Christine White, Swansea

It really works

The last SMarT News had a suggestion about using linseed to help constipation. I tried it without much enthusiasm but it really does work. Thank you for what is a great tip.

Stephen Smith, Stoke on Trent

Foundation of the East Midlands Support Group

Earlier this summer Elizabeth Brackenbury, sister of Sarah Matheson, invited all registered sufferers and their carers in the East Midlands to a superb luncheon at her home at Holme Pierrepont Hall in the beautiful outskirts of Nottingham. After a superb lunch of fresh salmon salad, Elizabeth outlined a little of the history of the Sarah Matheson Trust. Afterwards a healthy discussion with regard to our various symptoms and requirements took place. It was finally agreed that we should form what is now the East Midlands Support Group, the first ever, with the aim of promoting a wider public awareness of the condition and a forum where sufferers and carers can come together to discuss in confidence and friendship any fears or problems they may have.

On the 15th October the first official meeting was held, again at the residence of Elizabeth. We invited our very own (yes, you've guessed) Catherine. Catherine gave some further insight into the work of the Trust and what help she can offer on its behalf. Unfortunately our Alison was poorly and could not attend. Ah! As the discussion took place afterwards it was amazing to hear how different Health Authorities react to the condition. Again excellent refreshments were provided and a raffle was held which raised £19.50. The next meeting is planned for April 28th.

All the best

Ian Jones, Nottingham
Secretary of the
East Midlands Support Group

If you have ideas for a similar group in your area we will be happy to provide support and practical help to get started. We'd love to see a network of similar groups develop across the UK.

Living with MSA

A personal view from Jane Honnor

Multiple System Atrophy (MSA) is a degenerative disease. It appears to affect people about 60 years of age. It is no respecter of sex and afflicts people from very varied backgrounds. No two people have exactly the same symptoms, but all are similar – tremor and loss of control of muscles (usually the limbs) with impaired co-ordination.

After the initial shock (why ME? Why NOT me?), I gradually began to accept it. As far as I was concerned, lack of balance was bothering me most. For no apparent reason I was falling and I was also covered in bruises because I also bumped into furniture. After 15 months of tests the eventual diagnosis horrified me – I had always been such an independent person – helping others, rather than being helped – and this disease seemed to have ‘thrown me on the scrap heap’. However I was determined not to let ‘it’ beat me – I have always been a fighter – and I felt that, as I accepted that ‘these things happen’, I might as well get on with my life and see what good came out of it.

I was amazed at how many good things I have found (and am still finding). Once I managed to have a positive attitude, my situation seemed better: friendship from members of the SMT Contact Scheme (emails and phone calls) and understanding visitors, and pleasure found from simple things such as sunlight and attractive surroundings.

One of the most devastating effects of the disease for me is the feeling of a sense of frustration. After many failures and considerable thought, I decided to try having more ‘achievable goals’ – instead of trying to cultivate the whole garden I would grow things in pots by the house, where they can be tended by anyone. I enjoy painting (it is after all a ‘sedentary’ hobby) – I would turn the underused bathroom (the shower which

my husband and I prefer is downstairs) into a studio and, if necessary, paint with two hands!

Gradually, as an animal with 4 legs gets used to the loss of 1, I began to adapt to my new situation. I slowed down – both in pace and speech; I learnt to turn by degrees instead of spinning round, I found that looking directly at whatever I was trying to do actually helped – having ‘touch-typed’ for over 20 years I found it beneficial to use a ‘key guard’ and look!

All my life I have found that ‘change’ is the hardest thing to accept but to adapt I have found it to be the most necessary. Once I began to change things again, I found I was quite obsessive! I ‘down-sized’ my possessions (both furniture and ornaments) and found I had more space, less bruises and broke less! I ‘pruned’ my wardrobe and found that a few garments with easy fastenings sufficed for my needs; I sorted out the kitchen to accommodate my reduced capabilities – giving away or dumping redundant tools, china and ingredients.

I then thought about what gives me pleasure, and decided it is being in attractive surroundings, which I have created out of ‘what I have got’ – I recently spent a year in a much smaller house and garden and found that there are benefits to be had from less spacious surroundings. I also enjoy the effect of light on different objects and enjoyed trying to capture it in paint; obviously I can't paint as I used to do, and was trying to find an area to use where things would ‘stay’ where I left them. I suddenly realised that both my husband and I prefer showers (in our house downstairs) and that the bathroom is underused – it makes an excellent studio!

I could eulogise forever about the beneficial effect of change – as I said – once started it can become addictive!

Sarah Matheson Trust Research Fellow

Research update – current and proposed studies Hello again, here's an update on my research. My CO2 study is now complete and we are just awaiting some of the blood and saliva results so that we can start a full analysis of all the results. As I mentioned last time, my own feelings are that the Laser Doppler results from this study will prove to be the most exciting as they seemed to show a clear difference between the MSA and pure autonomic failure (PAF) patients. This is a key concern of my research, as it is not always possible to correctly distinguish these two medical conditions in the early onset with existing tests.

On a similar point, my new study of

skin blood flow in MSA and PAF is now well underway. There have been encouraging results, with the early findings matching our predictions of the different responses to simple stimuli such as cold, exercise and mental arithmetic in MSA and PAF. Interestingly mental arithmetic still seems to be able to produce a narrowing of the small blood vessels just under the skin (as seen in healthy people). This implies that there are still some preserved pathways allowing the stress of mental arithmetic to activate the sympathetic part of the autonomic nervous system in MSA. However, in work I have just presented at the 2003 annual Clinical Autonomic Research Society (CARS) meeting in London, I have shown that mental arithmetic is no longer able to increase blood pressure in MSA as it would in healthy people.

These findings would be in keeping with a dual model of blood vessel control in MSA, with the blood pressure control pathways being affected in MSA but with relative preservation of the pathways controlling the small skin blood vessels.

As a further part of the work I presented at CARS, I was able to demonstrate that, unlike mental arithmetic, cold stimuli were still able to significantly raise blood pressure in MSA (healthy controls show an increase in Blood pressure with cold) but not in PAF. This would be compatible with the known preservation of the more peripheral autonomic nerves in MSA but not in PAF as cold stimuli is thought to act more superficially. It would indeed be expected that there would be some preservation of response in MSA where the nerve damage is more central.

Fundraising

London Marathon 2004 The Sarah Matheson Trust will have 6 Golden Bond places available in 2004. All interested parties are asked to apply independently for a position through the ballot and those who are unsuccessful will then be considered for one of our reserved places. The closing date for Golden Bond entries in 30th January 2004. Decisions for SMT places will be made in the first two weeks of January. Bond places are only offered to runners with a link to the Trust through family or friends. Any interested parties should contact Amanda Bradburn on 020 7659 9790 or by email, amanda@aggmore.co.uk message service.

Fun Walk – Sunday 21st

September 2003 What wonderful weather for this SMT “family” gathering. Over seventy people and two dogs (Poppy and Galahad) turned up to support the event. Forty-five intrepid walkers registered for the 5 mile circuit and set off with hounds and children in tow across Hampton Court Park and back along the River

Thames. A further twelve people opted for



Walkers and supporters at the Fun Walk 2003

the more leisurely Treasure Hunt through the Wilderness Garden with clues set by Catherine and Alison.

Around noon we gathered at our “base camp” for a communal picnic.

It was a lively event with much chatter and exchange of news between old friends and new. The walkers were entered into a Champagne Draw – the lucky winner being Mrs Murray. Two teams, the Hemel Hoppers and the Yorkshire Puddings, tied for first place, solving the clues and collecting the booty on the Treasure Hunt. The prize was won on the tie-break, by the ‘Hemel Hoppers’, Linda and Kieran Stone. We will find more cryptic clues for next year!

We are pleased that the Fun Walk is proving to be an annual SMT “family” event that reflects the growth of our family and the continuing support we receive from



our varied membership – now standing at over 1,600 people. We had a most enjoyable day with much socialising and a considerable amount of fundraising, over £2,000 thus far. Many thanks to everyone who supported us.

And our thanks also to Pat Sweeny and her colleagues at Hampton Court Palace for their gracious support of this event.

Pyrenees to Paris – July 2003 Just to keep his hand in (or in this case wheels) Peter Murray, one of our Trustees, completed the Pyrénées to Paris stage of the Tour de France cycle route. A remarkable achievement, that proved a lot hotter than expected, raised vital funds for the SMT (in excess of £20,000) and also for the



The other major project I am engaged in at present is to examine blood pressure overshoot, which appears to occur in PAF but not MSA, after tilting the patient upright and then returning to the horizontal. More details of this study were covered in the last newsletter. Finally, we are waiting ethical approval of a study looking at an alternative medication, which could be used for controlling standing blood pressure in MSA and PAF. I hope to have more news about this study in the next newsletter.

Fragile X There is some interesting work emerging from the USA regarding a possible genetic link with MSA. At present there is no known genetic link with MSA, and MSA does not run in families. Indeed, the current guidelines for diagnosing MSA actually suggest

that a diagnosis of MSA should not be made if a patient presents with symptoms that have a hereditary element. The USA research concerns a different medical condition called Fragile X syndrome (FXS). Patients affected with Fragile X develop symptoms which are very different to those seen in MSA. They present as youngsters, usually with mental retardation and other clinical features such as long faces, and large jaws. About 1:4,000 males and 1:8,000 females are affected by FXS. It is a genetically inherited condition, but it is not usually passed on by affected individuals (this is thought to be mainly due to the fact that the often marked mental retardation makes it very difficult for patients to form relationships and have children). So how come FXS is still with us? Well, the answer lies with the rather complex

genetics of the condition. To try and explain this I will first try and provide some background knowledge.

Each cell in our body contains long strands of DNA which provide the blue print to make all the molecules we need. Each molecule that cells need is coded for by a stretch of the DNA called a gene. When a particular molecule is needed by the cell then the code is "read" by a special molecule called RNA, which then in turn is used as a template to turn the code into the molecule required. Many known genetic disorders are caused by a specific defect in the DNA code of the gene which is can then be passed on to children if they inherit the faulty gene. FXS is an example of a disease caused by a "trinucleotide repeats". This is a more complex situation. Consider the gene
(Continued overleaf)

development fund at the National Hospital for Neurology in London. Peter's only real complaint was that there was a distinct lack of complex carbohydrate available in the French restaurants along the route, a vital source of energy for such athletic pursuits! ("Frites" with everything, so very English!) Peter cycled 1100km, over 10 days. That makes around 70 miles a day! He regretted that cycling solo through the wine regions did not leave enough opportunity for sampling the assorted regional vintages along the way, and succumbing to any temptation to make up for this oversight with his evening meal would make the next day more effortful all round. Peter was glad when he made a triumphant entry into Paris to complete the ride and was

treated to a well deserved champagne reception! And for his next feat?



Vistas and vintages along the miles

Line Dance Our first fundraising Line Dance was held at Dawlish Leisure Centre on the 22nd November. The event was organised by Janet Fear and her friends, and included a Christmas Raffle. It was a lively event!

Christmas Cards We have now sold out of our Reindeer in the Snow cards, thank you to everyone who ordered them. There will be new cards available next year. A very big thank you, too, to Doreen Dowling who has been very busy supplying us with beautiful handmade Christmas cards. These have been selling like hot cakes at support meetings and the Fun Walk and have raised over £200.

Great North Run The Trust's own running man, Alan Commette, was running again this year proudly displaying the SMT colours. This year his fundraising efforts were shared with another charity. Alan was delighted to complete in 2hrs 20 minutes – as he said himself, only twice the time recorded by Paula Radcliffe!

New York Marathon Our first New York Marathon runner was Michael McMahon, running to raise funds in memory of his father in law, Patrick Brolly. We hope that the sleepless nights from his expected new arrival did nothing to detract from his

Fundraising

marathon attempt. We will update you in the next edition.

Berlin Half Marathon On a cold Sunday in April, Melanie Catling bravely completed the Berlin Half Marathon in just under 2 hours. Mel was nobly supported in freezing conditions (including a snow storm) by her partner Richard. She highly recommends the event for you runners out there as the course passes many of Berlin's landmarks (Checkpoint Charlie, Brandenburg Gate). Mel's father Raymond has MSA and the whole family are involved with supporting him and the work of the Trust. Mel's efforts in this event have raised over £200 for the SMT.

Other fundraising events These include donations in lieu of gifts at Peter Thirsk's 60th Birthday Barbecue in memory of his brother Jim. A coffee morning organised by Janet Fear in Devon raised £250. And we must not forget David Barr, the son of two of our Scottish members Leslie and Marion, who raised £200 for SMT in the London Marathon this year. Well done all, and thank you.



Sarah Matheson Trust for
Multiple System Atrophy

Information, Support, Education & Research in Multiple System Atrophy and other autonomic disorders.

Providing services to people with MSA, families, carers and professionals.

- Information leaflets and newsletters
- Specialist nurses
- Telephone advice line
- Regional support meetings
- Training and education sessions
- MSA research
- Communication Aid Loans

Patrons:

Sir Roger Bannister CBE FRCP
Professor CJ Mathias DPhil DSc FRCP

Trustees:

Mrs Robin Brackenbury
Michael Cook AO
Valentine Fleming
Christopher Marsden
Hugh Matheson
Peter Murray
Eileen Lady Strathnaver OBE
Lady Harriot Tennant

All correspondence and enquiries to:

Alison Aberly
Sarah Matheson Trust
Pickering Unit
St Mary's Hospital
Praed Street
London W2 1NY
020 7886 1520
020 7886 1540 (fax)

www.msaweb.co.uk

The Trust is financed entirely by voluntary donations.

Registered Charity Number 1062308

central to FXS. Normally this gene will produce a molecule called FMRP. This molecule is thought to be important in the development and upkeep of nerves—specifically in the synapses, which are where the nerves are able to pass on whatever signal they happen to be carrying. Even healthy people have a number of minor defects in the gene coding for FMRP (specifically these take the form of a pattern of 3 different DNA building blocks repeated about 20 times in a row, called repeat trinucleotide sequences). These defects do not interfere with the process of making FMRP unless the number of repeats increase to say 200 or more at which point the gene is unable to create the FMRP and the disease FXS will develop in that individual.

Although it is not known why the small number of repeats can suddenly increase to the point of causing FXS is not known, it is known that this usually happens in more than one step. The classic picture would be that a man would be born with a slight increase in the number of trinucleotide repeats in the FXS gene (about 60 or so). If he has a daughter then she would inherit even more repeats (but being female would be somewhat protected against developing FXS). If this daughter then had a son, he would inherit even more of the repeat trinucleotide repeats (say over 200), meaning the FXS gene would no longer work and that this son would develop FXS disease.

The possible MSA connection developed as follows. It was noted that some male children with FXS had grandfathers who were starting to develop either tremor or ataxia late on in life (called Fragile X Associated Tremor/Ataxia Syndrome, or FXTAS). It was then realised that having even 60 or

so repeats could cause symptoms, although these were clearly different to FXS. It seems that although these people could still produce the FRMP, this was no longer working properly because of the increase in trinucleotide repeats. What was interesting was that many of these grandfathers had a fairly characteristic MRI finding (hyperintense rim of middle cerebellar peduncle). This finding is also found in about 40% of MSA subjects (and seen in some other conditions). Fascinatingly in one small study in America, of 19 MSA patients 40% were found to have a slightly increased number of trinucleotide repeats in the FXS gene, and these also had the characteristic MRI findings.

It would be very tempting to speculate that there could be a sub-population of about 40% of MSA subjects who had this increase in trinucleotide repeats as an underlying aetiology. Certainly the unusual genetics involved might explain the absence of evidence for MSA “running in families”. However, much more work needs to be done before such a link could be proved or disproved. Although autonomic failure might be a factor in some of the FXTAS, tremor is not as common as a presenting complaint in MSA. Furthermore, there is no specific treatment as yet for FXTAS and so screening for this condition would not yet be easy to justify. However, if these early results were built upon by larger studies, they would create a real new frontier of research into MSA which is still considered to be a disease without a significant genetic link.

*Dr Tim Young,
Neurovascular Medicine (Pickering Unit),
Imperial College London,
St Mary's Hospital*

Diary Dates:

Awareness days

Middlesbrough January 22nd, The Disablement Services Centre, James Cook University Hospital

Plymouth February 2nd, Derrisford Hospital

Liverpool February 12th, Glaxo Neurological Centre

Social evening

London February 23rd, Architects' Association

All our members will automatically be notified of any events held in their area, an invitation will be forthcoming once the date and details are fixed