



*the Hereditary  
Multiple Exostoses  
Support Group*

**Affiliated to Contact a Family**

The Committee Members:

- Christine Spencer – Chair
- Rosina Gordon – Vice Chair/Fundraiser
- Steve Harding –Treasurer
- Dawn Searle–National Contact
- Helen Small – Research Development
- Lynda Tams–Secretary
- Brian Gordon – Web Designer
- Judith Fletcher–Overseas Contact
- Pat Savage
- Stephen Watson

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**A Word From The Chair**

Hi everyone and welcome to our latest newsletter.

It was lovely to see some of you again at our Conference this year and even more rewarding to meet some of our new members for the first time. Although the attendance was lower this year we did receive some very positive feedback and suggestions for future content which is always helpful.

We do however wonder if these kind of conferences are exactly what our members want and to this end I enclose a questionnaire which I would be grateful if you could all complete and return to us by 31<sup>st</sup> January 2010. This will help us plan such events like this in the future. If you do have access to a computer this questionnaire can be completed online and returned to us if preferred.

We have had a couple of committee changes since our AGM.

Judith Fletcher has stepped down as Trustee but will still remain on the committee as Overseas Contact. Barbara Gibbs and her daughter Wendy Tollman have stepped down due to family circumstances.

I am sure I can speak for you all when I thank Barbara and Wendy for all their hard work and commitment over the years.

I am sure both Wendy and Barbara will

continue to help raise the profile of HME and will continue with her excellent record of fundraising challenges. We will be seeing them at future get togethers! Dawn Searle, our previous Fundraiser, is now our National Contact and normally is your first point of contact via our Helpline. Not forgetting of course Rosina Gordon who has now also taken on the role of Vice Chair.

We now have a new Patron of the Group, Jodie Prenger. For those who don't know Jodie was the winner of the BBC's 'I'd do Anything' show. A musical talent search for the role of Nancy in the West End's show Oliver! Thank you Jodie and welcome. I am looking forward to meeting Jodie soon.

Thank you for all your support and many thanks for those who have donated to our cause - this is always very much appreciated and will be put to good use.

The festive season is almost upon us. May I thank you all for your help and support this year. We wish you a Merry Christmas and a Happy New Year

*Christine Spencer*

**Changes To The Newsletter**

New Distribution method

It has become increasingly more expensive for us to fund the publication of the newsletter with costs of printers and postage going up. It has therefore been decided that from this newsletter onwards this will be published on the website for people to download as a PDF document, during the months of February and August. If you do not have access to the internet and/or would rather continue to have a copy posted out to you please let Christine Spencer know ASAP and we will make arrangements to have the newsletter posted out to you in future.

New Newsletter Editor needed

Wendy Tollman our previous newsletter editor has sadly decided to stand down due to family and other commitments and feels that she would not be able to give her best, so we are now currently newsletter editor less, if you have experience in this field and would like to give it a go, then please drop Christine Spencer an email letting her know your interest in the position:

[support@hmesg.org.uk](mailto:support@hmesg.org.uk)

A few words from our new Patron Jodie Prenger

*"When I heard of the excellent work carried out by The Hereditary Multiple Exostoses Support Group I was only too pleased to become it's Patron. Our aim as a Group is to adopt a positive approach to the support and care of families and individuals affected by HME. We endeavour both to educate and learn more about the genes that cause HME. Our overall aim is to make a difference to people affected by HME. It is the little extras that that the Support Group provide that mean so much and make such a difference to people with HME and their families. By helping me and the many volunteers who make up the HME Support Group,*



Picture of Jodie Prenger

*to continue raising funds, you can be assured that all the monies raised, will benefit and support the people nationally who are affected by HME.*

*Help us make a difference."  
Take Care*



## A Word From The Vice Chair

My name is Rosina aka Roz and for those of you that haven't met me already, I started out as the Fundraiser for HME SG and at our AGM in May, I took on the position of Vice Chair as well.

As some of you may know, Brian, the web designer for the Support Group is my husband. We live in Preston with our crazy dog and two cats. I am only 26 years old. Given that I am reasonably young, it seems strange to some people that I would dedicate my free time to a cause such as this but, I wouldn't have it any other way. With Brian, we set up a Beaver and Cub Scout Colony in the area - which was something that we had been trying to do for a number of months, however personal circumstances got in the way and in October 2008 we had our first meeting. In May, we held a fundraiser after the conference in Birmingham and I would like to take the time to thank the few people that were able to stay around and attend the event. We managed to raise just over £300 which is fantastic! It was an amazing evening and it was a shame that others couldn't enjoy the festivities which brings me on to my next topic!

Already I am kicking about a few ideas for other fundraising and social evenings that I would really like you all to attend if you can next year. Watch this space for further details or keep checking our

website! Other events for the next couple of years that I have in mind at present include a 5 a-side tournament, a talent competition and a Vegas Night so keep your eyes peeled ready for flyers on our website.

If you have any ideas or anything that you would like to share with me to do with the Fundraising then please feel free to contact me at

RosinaJGordon@googlemail.com

Thank you everyone for supporting the support group and if anyone would like to let me know details of any local fundraising that they may be doing and how much they have raised - no matter how big or small then please let Christine know so that she can update the other members.

*Roz*



Vice Chair Roz

## Preimplantation Genetic Diagnosis (PGD) for HME - Update May 2009

Preimplantation Genetic Diagnosis (PGD) is a treatment used to help people who do not want to pass on a genetic condition to their children. Basically, in-vitro fertilisation (IVF) is used to produce embryos in a laboratory. When the embryos are made up of only a few cells, usually eight, a cell is extracted and a DNA test is performed to check for the genetic condition of interest. Only embryos which do not have that condition are implanted into the mother.

The Human Fertilisation and Embryology Authority (HFEA) is the body which grants licenses for centres to perform PGD. The HFEA document download or view the pdf now lists clinics which have a license for PGD, but not all are using their licenses. Currently, PGD is available at several centres in London; the largest unit in the UK is at Guy's and St Thomas' Hospital in London. PGD is also available at Glasgow Royal Infirmary (only for translocations and sex-linked conditions) and Care Fertility in Nottingham. The Centre for Reproductive Medicine and Fertility in Sheffield and the Clarendon Wing, Leeds General Infirmary, both have licenses for PGD, but are not offering this treatment at the moment. Leeds and Sheffield both act as satellite units for Guy's; patients can have some of the preparatory treatment at these hospitals, but have to go to Guy's for the main PGD treatment. PGD can be obtained on the NHS at all these licensed centres, including at private clinics such as Care Fertility, Nottingham. However the funding for PGD is made by each NHS Trust, and very different decisions are made around the country; some Trusts fund treatments for two children, most only fund PGD if the couple do not have an unaffected child and some trusts refuse to fund PGD at all. The number of cycles funded also varies. There is a possibility in future that funding decisions could be taken at a regional basis; this would make the situation fairer.

These centres not only require a license to perform PGD, at the moment they also need to apply for a license for each genetic condition they treat. Once they obtain a license for a specific condition, it only applies to that clinic. The amended Human Fertilisation and Embryology (HFE) Act 2008 comes into effect on the 1st of October 2009. It is the first time that PGD is specifically regulated by law. The changes to procedures will make it easier for people to obtain PGD. From October all clinics licensed for PGD will automatically be able to treat patients for conditions licensed by the HFEA, even if it was another clinic which applied for the license. In the case of a condition which has not yet been licensed, clinics will no longer need to have a patient in mind when applying to the HFEA. After October 2009, once a license has been granted for a condition which is not currently licensed, that license can be used by all clinics licensed for PGD.

I have Hereditary Multiple Exostoses (HME) and, as people in HMESEG will be aware, I have a 50:50 chance of passing it on. For many years I worried about having children because of this risk. I wanted PGD, but I was told by my genetic counsellor that it was not available for HME. That was nine years ago and HME is still a condition for which PGD is not licensed in this country.

I am a member of the Human Genetics Commission (HGC) consultative panel. The commission advises the government on issues related to ethics and genetics. The consultative panel is made up of people who either have a genetic condition themselves, or they are carers of someone with a genetic condition. The consultative panel meet once a year; I have been a member since 2007. My husband and I run an educational software company called Illumination Educational Software. We have developed an online site on genetics called Gene Journey. Attending the HGC consultative panel has helped us keep up to date with issues in this field.

In the meeting in May 2009, Preimplantation Genetic Diagnosis was on the agenda for the consultative panel. Dr Chris O'Toole and Danny Edwards from the HFEA attended the meeting. Under the new Act the HFEA will be responsible for deciding the seriousness of each genetic condition in general. It needs to be satisfied that there is 'a significant risk that a person with the abnormality will have, or develop, a serious physical or mental disability, a serious illness or any other serious medical condition.' The HFEA is currently considering consulting with patient groups in order to help them decide whether a condition is serious enough to justify PGD, in general. It will remain the responsibility of the clinics to decide whether PGD is suitable for individual cases. There was a strong consensus of the consultative panel that the HFEA should consult patient groups. If this comes about then HMESEG may be consulted, perhaps through its affiliation to GIG (Genetic Interest Group).

I met Natalie Glascott-Tull six years ago at the first HMESEG meeting I attended. I interviewed her for Gene Journey; she is currently having PGD for HME. If you would like to see the video of Natalie, email me at: [tina@illumination-ed.co.uk](mailto:tina@illumination-ed.co.uk) and I will be happy to arrange free access to Gene Journey for you. Natalie was told by the PGD clinic she attended in the UK that a license was not available for HME. What they didn't make clear was that they could have applied for a license to treat her, but, for whatever reason, they chose not to. She has been funded on the NHS to have PGD in Belgium, but not in the UK. If

anyone in HMEG is considering PGD, I would be interested to hear from you. If you do attend a clinic and find they do not apply for a license, try and find out the reasons for their decision. The good news is that, under the new Act, if you do manage to get a license for PGD for HME, the next time someone with HME wants to have PGD, the license will already be available at all UK clinics licensed for PGD. Please email me if you have any other queries.

Tina Read

(this document was kindly submitted by Tina Read and is also available on the website under Research)

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## Birmingham Conference 2009

We had several speakers attend this year's conference, one of them was delivered by Mr A.D.L. Baker *MRCs* an Orthopaedic Registrar (Orthopaedic surgeon in training) working with Mr D.E. Porter *FRCS* (Consultant Orthopaedic surgeon) who runs a specialist HME clinic in Edinburgh. Mr Baker has an interest in both spine surgery and HME. He gave us a talk on Exostoses around the spine, his talk is documented below.

As well as Mr Baker we were pleased to have with us Dr Stephen Minger who is the Director of the Stem Cell Biology Laboratory and a Senior Lecturer in the new Wolfson Centre for Age Related Diseases at King's College London. He gave us a very insightful and fun talk on stem cell research and the advances that they are making in this kind of research.

### Exostoses around the Spine

#### Introduction:

The following talk includes our local experience in Edinburgh of exostoses around the spine, and a review of all the available literature. It should be regarded as my initial thoughts and exploration of evidence rather than established practice.

Complications associated with exostoses around the spine can be severe. However, it is worth pointing out at this stage that these potential complications are rare and in most cases treatable. Also that, if an exostosis is discovered around the spine it may be that no treatment is required.

Pictures showing an osteochondroma (exostosis) that had been removed at surgery, and how it appeared prior to removal both on x-ray and a photograph of the affected limb were shown.

Further examples causing problems within limb length were also shown. X-rays were presented together with MRI scans showing a cartilage covered osteochondroma (exostosis) of the spinous process of the fourth lumbar vertebrae. The possibility of a similar lesion causing spinal deformity was discussed.

#### How rare is rare?

From the literature we can estimate that approximately 4 to seven percent of all excised spinal tumours turn out to be osteochondromas. However, there are only 200 reported cases in the literature. The most common age to develop the spinal osteochondroma is in the third decade of life. They can either be solitary or part of a Hereditary Multiple Exostosis (HME). Some literature suggests that spinal exostoses may be more common in the inpatients with HME perhaps as many as 9%.

From the database of patients with HME further information was gathered. The database is made up of 172 individuals from 72 families all suffering from a HME. In total there were 278 operations performed the removal of bony lumps and 5760 palpable exostoses. Only five palpable spinal exostoses were seen and 10 individuals had had exostoses previously removed from around the spine. The mean age in the database for removal of an exostosis was 35 and there was a male to female ratio of two to one.

#### Where do they occur?

From a review of the literature it appears that both in hereditary multiple exostosis and in sporadic cases exostoses were found more commonly towards the top of the spine and towards the back.

88% of the exostoses originated from the posterior elements. In the HME group 57% were found within the cervical spine

**Exostoses around the Spine (cont)**

(neck), 36% within the thoracic spine (chest) and only 2% within the lumbar spine. 19% were identified in the posterior arch of C2 (the second cervical vertebra).

Looking at spinal exostoses as a whole 54% were solitary and 43% were identified in patients with hereditary multiple exostosis.

**Dangers specific to the spine:**

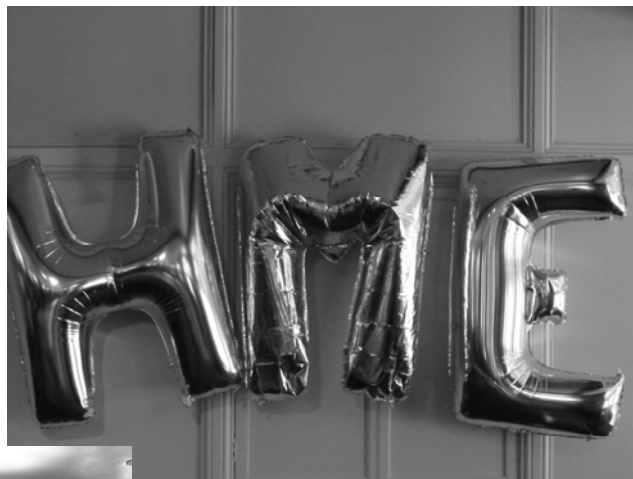
Exostoses elsewhere can cause abnormalities in growth and painful lumps. However, in the spine there is an additional risk that they can press on neurological structures leading to severe complications. If the exostosis compresses a nerve root that has exited the spinal canal it may cause problems with the function of that nerve leading to pain, loss of sensation and perhaps motor dysfunction in the distribution of the nerve root affected. Worse still if the exostosis invades into the spinal canal and presses on the spinal cord there is the theoretical risk of causing paraplegia.

**Presenting symptoms:**

So how can we know if somebody has a spinal exostosis that requires treatment? A review of the literature reveals that the most common presenting symptom for an exostosis that subsequently required removal was that of a neurological deficit (71%). Other presenting symptoms including a mass of being felt (17%), pain (5%), and a group of other symptoms (7%). Other more rare presenting symptoms included scoliosis, dysphagia, stroke, and an incidental finding of a scan. In patients with HME 90% of the presenting symptoms were neurological.

Pictures were shown of an MRI scan together with a CT scan and myelogram showing how an exostosis might compress the spinal cord or nerve root potentially causing neurological problems

We thought that anyone suffering from an HME might want to be aware of some things to look out for in case they develop an exostosis around the spine.



The conference gets underway

HMESG Conference 2009



Mr Baker begins his talk

Telephone: 01438 861866 email: [support@hmesg.org.uk](mailto:support@hmesg.org.uk) web: [www.hmesg.org.uk](http://www.hmesg.org.uk)

## Fundraising Page

### Beach Party Fundraiser!

Well the grass skirts were certainly swaying to the beat of the music in Gateshead back in August at our fundraiser. No matter whether you were one of the few members of the group that came or just there for a good night out, everyone that came really enjoyed themselves.

It was a fantastic evening with all kinds of entertainment. We had the Auction, the Silent Auction, a raffle, performances and general fun and games that got everyone on their feet.

Cocktails were flowing over on the Tiki Table which certainly added to the merriment of the limbo game - which we had to run 2 rounds of just because it was so successful and that combined with some hula hoop competitions and the standard "whose birthday/anniversary is it this month" really set off the party off.

Dancetroop Evolution donated their time to do two 12 minute sets of incredible dance moves, flips and tumbles which went down a treat and was so successful that they too ran around with buckets to help raise some extra money - after raising an extra £50 approx they offered to do one final dance set for us and then joined in the rest of the disco for the evening.

The Auction included some generously donated Karate Items - Gi's and lessons, signed items that had been donated from Bono and the rest of the U2 band that were donated especially for the fundraising events that we run and not to mention some football memorabilia as well.

The Silent Auction was also very successful with a variety of items to being up for grabs at bargain prices including some more football goodies and signed merchandise from Katherine Jenkins and the Hollyoaks cast.

The raffle went down an absolute storm with stacks of prizes being given out - a very special thanks goes to the younger members of the support group especially Ryan Spencer and his karate friends from Low Fell Shotokan Karate Club who attended and for helping out with the selling of the tickets throughout the evening.

Rob Searle took a few minutes out of his evening before the Auction to give a quick talk about HME and how the condition affects him - thank you very much for the words you spoke and for really personalising the reason why we were all there and what we were raising money for.

Another very special thank you goes to the amazing Roxy Searle who once again was able to wow the audience with her stunning vocal ability and overall performance however, we cannot forget the wonderful Harry that was also performing with Roxy both singing and playing the guitar - both of them even raising a little money to come on and do additional sets for us.

With all that in mind and the huge amount of hard work from Christine, Dawn and myself, that went into the evening it was safe to say that everyone who attended really enjoyed themselves and helped to raise over £2200 so it was really worthwhile.

In the future our hope is to run some more large scale fundraising events and social gatherings to help raise much needed funds to support the conferences and general running costs that the group does incur. If anyone is holding any events locally that they would like to send us info of, please feel free to contact so we can help promote your event and pass the details round. Thank you again everyone for your help and support.

*Roz*

## Fundraising, Pens, Rulers & Trolley Coins

We still have a large number of pens, trolley coins and rulers! Don't forget these are perfect for promoting the group and giving out to people. Here are some ideas on what you could do with them;

- Purchase 10/20 pens and then ask friends and family to buy them from you.
- Trolley coins always come in handy -

why don't you purchase a few and give them to your friends/relatives that have not got one?

- The rulers are amazing and with a calculator on them too so they are excellent for saving space in a bag/pencil case.

If you would be interested in purchasing any of the items that I

mentioned above then please let me know ASAP and I can let you know the details of the postage costs.

[RosinaJGordon@googlemail.com](mailto:RosinaJGordon@googlemail.com)

We have had so many donations to the support group for the research fund and in general, we would like to say a **Big Thank You** to all those that have and are still carrying on a great deal of work to raise funds for the group. There have been so many kind donations since our last newsletter. Too many to list here today.

A thank you goes out to Mrs Sue Penniall (a friend of Wendy Tollmans) and Reynolds Porter Chamberlain LLP the company Sue works for. They ask its staff to nominate charities which they are involved with based on their experiences and from the applications a number of charities are selected to receive a donation. Sue very kindly nominated us (she has done some personal fund

raising for us before) and spent a lot of time and effort preparing a very good nomination. I should say at this point that I was totally unaware of any of this - she kept it as a complete surprise - but her husband subsequently told me how much time and work she put into it as the company is a large law firm and competition for donations is huge. It is even more kind given the fact that, although a friend, she is not a close friend and the fact that she was prepared to go to this much effort for us is extremely touching. So once again thank you Sue and Reynolds Porter Chamberlain LLP.

A thank you goes to the Anderson family for raising £40 with their collection box.

And a final thank you goes out to our very own Chairperson Christine Spencer and her son Ryan, who both took part in the Boxing Day Dip, and raised £776.80 and not even a cold in sight.



## New Conference organiser needed



We need you to come forward and help the group, are you a lively person who gets on well and knows how to get a good deal on things? you will be responsible for organising the location of the conferences and annual fundraiser, whilst arranging for the fundraiser you will be required to work closely with the fundraiser to ensure that all things go according

to plan, and also running things by the chairperson, If you answered yes to any of these questions then contact Christine telling her of your interest in the position of events organiser.