



NEWSLETTER

Society for the Study of Behavioural Phenotypes

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ANNUAL REVIEW

The SSBP continues to thrive with another busy and productive year. After several years' absence, Professor Greg O'Brien, one of the founding members of the SSBP and a former Honorary Secretary, was welcomed back onto the Committee. Our membership list has also increased to 140 members and we continue to attract members from a wide range of complementary disciplines.

In November 2004, the Society's 8th International Symposium was held in the Hotel Gran Via, Barcelona, Spain. The theme of the conference was Fragile X syndrome and we were made very welcome by the local organisers Drs Josep Artigas and Carme Brun. They exhorted us to "Let ourselves be swept by the wave of life that flows along the Ramblas, stimulating our senses and arousing thousands of sensations!" Conference attendees are invited to contact the

Editor to confirm and give details of such experiences!

For those of us coming from Northern Europe, the novelty of sitting in the warm sunshine in November was a real treat. In addition, Barcelona is a magnificent city - I counted many old and new converts to the joys of Gaudi! The conference hotel was located in the heart of Barcelona, a stone's throw away from most of the major sights.

The opening session on Genetics featured keynote lectures on the genetics of Attention Deficit Hyperactivity Disorder (Professor Anita Thapar, Cardiff University, UK), mental retardation (Dr Isabel Tejada, Hospital de Basurto, Bilbao) and murine models of behaviour (Dr Xavier Estivill, Centre for Genomic Regulation, Barcelona). There were two wonderful sessions devoted to Fragile X, the main theme of the meeting, with keynote lectures on 25 years of X-linked mental

retardation (Professor Jean Pierre Fryns, University of Leuven, Belgium), a longitudinal developmental perspective of Fragile X (Dr Jeremy Turk, St George's Medical School, London, UK) and recent molecular advances in Fragile X (Professor Paul Hagerman, University of California at Davis, USA).



(Robbie Fountain, Louisa Nespoli and Jenny Wishart enjoying the Barcelona Conference dinner).

Many of you will remember that the Les Fountain Travel Bursary was established last year in memory of Robbie's husband Les who died tragically in 2003. The winner of the inaugural Bursary was Katy Berg from the University of Birmingham for her study "Behavioural characteristics of Lowe Syndrome" which she presented at the meeting. She outlines her study and experiences of the Barcelona meeting elsewhere in this newsletter.

The conference dinner usually manages a surprise or two – who can forget the previously undiscovered skill of Greg O'Brien as a tour guide for the Newcastle conference dinner?! The logistics of getting from the conference hotel to the waterfront proved beyond the capacity of many SSBP committee and other members. A decision to use

the underground resulted in a frantic dash to the tube to discover too late that we were going in completely the wrong direction! Suffice to say that, as always, the organisational skills of Robbie Fountain saved the day!

A new initiative this year included a presentation by Stewart Einfeld, host of the forthcoming 9th International Symposium to be held in Cairns, Australia October 6th-8th 2005. In addition to Stewart, the conference is being organised by Randi Hagerman, Jim Harris, David Dossetor and members of the Committee. The theme of this meeting will be "Treatment of Behavioural Problems in Neurodevelopmental Disorders". During Stewart's presentation, we were treated to fantastic visual displays of the great barrier reef, crocodiles and tropical rainforests and led to a great rush for members to mark off the dates in their diaries.

See you in Cairns!

Professor Kieran Murphy
Honorary Secretary



*(Delegates attending the Barcelona meeting enjoying the Conference dinner at Magnum 7 Portes).
[photos courtesy of Jim Harris]*

Treasurer's Report

Once again as I write this report for the Newsletter our accounts are soon to be audited and the results will be presented at the AGM in Cairns. From our July bank statements we have just over £19,000 in our accounts with £12,000 allocated for the payment of an invoice for administrative costs. Thus, we have funds of approximately £7,000, which is a little under the amount that we might consider to be a comfortable financial buffer. However, we have paid some of the Cairns' conference expenses in advance and this is our normal financial lowpoint in the year as we await income from registrations.

Last year we raised the membership fee and this has helped us to build up funds independent of conference income. However, we have noticed that payment of membership fees is slower this year and it is perhaps too early to say what impact the increase in fees has had on membership numbers. On a more positive note we continue to recruit more members and have been able to offer a very low registration fee for our members that includes meals and the conference dinner. Additionally, the hard work of Stewart Einfeld in gaining sponsorship for the Cairns conference has helped enormously and the Society is very grateful to Stewart for achieving the high level of sponsorship.

At this year's AGM I will present a budget for the coming year that will cover our running costs set against membership fees but not including conference income and expenditure. This is so that we can clearly identify the additional income that conferences should generate to keep us afloat and allow us to keep registration fees low for members and offer a rich

programme of scientific and social events. Putting aside next year's Dublin conference, the estimated income for the Society will be £8,250 whilst expenditure will be £11,300. Thus the conference balance needs to be at least +£3,000. Thanks to Kieran Murphy's early work on sponsorship we are confident that we can once again offer value for money for our membership and hope to see you in Dublin.

Prof. Chris Oliver
Honorary Treasurer (August, 2005)

Cairns Meeting details:

The 9th International Meeting will be held in Cairns, Australia at the Novotel, Palm Cove, from October 6th-9th, 2005, with meetings actually held on the 6th-8th.



Contact the Society's Office or check on the Web site for further information.

Advance Notice for the SSBP 11th Scientific Meeting 2006

The Society will have its next Scientific Meeting (the 11th) in Dublin, returning to that marvellous city after an absence of 10 years. The meeting

dates are October 19th to 20th, 2006, with an Educational Day taking place on October 18th and departures on October 21st.



The National Gallery of Ireland

The venue will be the beautiful National Gallery of Ireland and the Conference Dinner will be ably hosted by Prof. Kieran Murphy at the nearby Royal College of Surgeons in Ireland.

While some locations offer charm on a par with Ireland, few cities are as renowned as Dublin for its strong tradition of arts, culture, entertainment and hospitality. While Dublin continues to embody these characteristics, it has also become a thoroughly modern thriving cosmopolitan city of 1.5 million inhabitants. It is easily accessible through its airport located 10 km to the north of the city and is the main gateway to Ireland, with excellent direct links to most major European and North American airports.

For more general information on Dublin, visit the Dublin tourism website on www.visitdublin.com or www.dublinfo.com.

Further details of the meeting will be posted on the SSBP website (www.ssbp.co.uk) in due course

Conference or membership information

Further information regarding any meeting or about the membership of the SSBP is available from:

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The 24-Hour Sponsored Pool Competition

So here I am at 3.00am in the kitchen at the University Sports and Social Club cooking the third round of bacon rolls for the night. Six players are out in the main bar just over half way through the 24-hour marathon! It's July 2nd & 3rd and the night is warm and close and the kitchen's getting far too hot for my taste.

Two pots of tea, several yoghurts and bananas later I decided it is time to wake up Sue. She's been in the back Snooker room ensconced on one of the covered Snooker tables for a couple of hours. "Cup of Tea Mrs H-M?" I say brightly! "It's just coming up to 7.00am". Particularly for those of a sensitive nature I won't print the reply!

We'd been sitting in a pub nattering about this and that on a February evening trying to find away of cheering up the dank darkness outside when someone said that Les always knew ways of having fun. It was always something he was good at and could find a funny side to most things. He

was always at his best over a game of pool. Shall we do something to raise some money for his charity? That was it! The plan went into action.

Stuart Collins' friend and work colleague in the White Horse pub where Les worked in the evenings and weekends took over the planning of the competition. We would try to find eight players for a Pool Competition – we would get sponsorship for the players and see what funds could be raised for the **Les Fountain Bursary Fund** of the SSBP.

As time went on when we were talking to friends and former work colleagues it became obvious that many wished to donate something, other than cash, to raise funds so the Charity Auction went into swing. Chris Herbert, aka the SSBP Web Master was recruited as one of our players and enthusiastically set about making a new section of the SSBP web site showing the players' names and how to sponsor someone playing in the competition. He also donated the services of his dad, Dennis, as the Auctioneer. The other players were Chris's colleague John Walsh, Stuart Collins the organiser, Rory Matthews (my colleague Sue's son), Danny Davies our youngest player who is Stuart's nephew and of course Chris Fountain (Les's son) who was playing to uphold the family name.

My father donated a sum to help underwrite the competition which paid for the trophy for the winning player and most of the food eaten by the boys during the night.

Many local businesses contributed to the auction including meals for two at two local pubs, the White Horse, Barton and the Three Horseshoes, Comberton, many tickets to local

attractions such as the Farm at Wimpole Hall, the Anglo-Saxon village at Stow, and items for sales such as a local picture framer who donated the gift of having your favourite picture framed.



Left to right Chris Herbert (Web Master), Robbie Fountain, Rory Hampton-Matthews, John Walsh, Chris Fountain, Stuart Collins (winner) and Danny Davis

Various other items donated included a litre bottle of 10 year old malt whisky, a bottle of reserve Port and a bottle of Black Label Lanson Champagne.

Our day had been started at 12.00 noon by Committee member Petrus De Vries starting the 24-hour tournament. He returned in the evening to purchase many strange items (but also the Port!)

T-Shirts for the players had been printed and the 4 helpers had slightly different versions to wear, all advertising the SSBP. The T-shirts were made by Talking T's who also kindly donated one to be made up to order for the auction. Johnny Hon, a previous presenter in Dublin at an SSBP meeting, who has now moved on to greater things, donated funds to

underwrite the cost of the T-shirt printing.

The players themselves all raised between £100 to £150 each in sponsorship and some of this had been donated over the web site by SSBP members from as far a field as the USA.

The entertainment for the evening was supplied by a very talented trio of John Wright, Chris Newman and James Doherty, known collectively as the Beach Café Band. Their music took many of us back to beach holidays in warm sunny places!

All in all we had a great time and I was amazed to find at midday on the Sunday that the 6 players decided to finish 30 rounds of the rota to make it fair to all players (this meant going overtime by 2-3 minutes). The Tournament was won by Stuart Collins, but 2 of the others were equal second.

Many thanks go to Petrus, Sue and her daughter Charlotte who ran the bar in the evening, all those who sponsored players and donated goods for the auction or their time to help. Les would have greatly enjoyed the event. We raised just over £1100 for the Les Fountain Bursary Fund and hopefully this will be added to by the Inland Revenue since much of the money donated was done through Gift Aid.

Many thanks to all!

Robbie Fountain
SSBP Administrative Secretary

The Barcelona Meeting 2004 **Katy Berg, University of** **Birmingham, UK.**

Last year I was lucky enough to have been awarded the Les Fountain Bursary enabling me to attend the SSBP's 8th International Symposium. In doing so I gained two new experiences – the first was the opportunity to present a paper at an international conference and the second was the chance to visit the city of Barcelona. Both of these “firsts” will remain with me for some time as both the conference and the city made quite an impression on me.

In the weeks running up to the conference I became slightly anxious about giving my first paper presentation and spent quite a lot of time preparing, practising and worrying! Similarly, though to a much lesser degree, when visiting a new city for the first time I often feel uncertain of what to expect and like to prepare myself as much as possible by arming myself with a collection of maps, phrasebooks and guidebooks. Although my colleagues and I arrived in Barcelona after dark and were deposited from the airport bus in the middle of the city without much of an idea of where we were, I found my apprehension immediately quelled. There was something about the atmosphere of the city that immediately made you feel welcome and at home. A friendly, accepting and easygoing attitude seemed to be reflected in both the people and the pace of life in the city.

In the same way my apprehension about presenting a paper were somewhat (if not entirely!) alleviated on arriving at the conference and meeting some of the other delegates. The warm and enthusiastic welcome

from Josep Artigas (Unitat de Neuropediatria, Barcelona & Organising Committee member) reflected the atmosphere I'd experienced on arriving in Barcelona and the relaxed and good humoured way in which presenters dealt with the audiovisual equipment's climate adjustment problems made for a much less threatening environment.



Not having attended an international multidisciplinary conference before it was interesting to hear how researchers working in different fields and in different countries were approaching the same research questions from different angles. For example Paul Hagerman's (MIND Institute, University of California) description of his team's search for the mechanisms that lead to the silencing of the FMR1 gene in his paper "Fragile X: Molecular Insights into Clinical Involvement" and Isabel Tejada's (Unidad de Genetica, Hospital de Basurto, Spain) paper "Genetic Causes of mental retardation: the state of the art" in which she described how researchers in Spain had formed GIRMOGEN – a multidisciplinary collaborative research network. I was also able to learn about syndromes I had not previously come across such as Ring Chromosome 22 (Sarah Curran et al. Institute of Psychiatry, London) and Ring Chromosome 20 (Mary E O'Regan,

Royal Hospital for Sick Children, Glasgow) as well as hearing different perspectives on more well known syndromes such as Jeremy Turk's paper (St George's Hospital Medical School, London) Fragile X Syndrome: A longitudinal developmental perspective. In this paper he described how individuals with premutations present deficits similar to those seen in full mutation carriers and gave an overview of recent clinical research on the use of selected medications (alongside psychological, social and educational interventions) to treat certain challenging behaviours. He also discussed efforts to develop a model that could be used to conceptualise the developmental dynamics seen in Fragile X which incorporates genetic, neurological, cognitive, behavioural and environmental aspects.

So as a relatively inexperienced researcher new to the study of behavioural phenotypes it was a valuable experience to be able to see how all the different parts of the puzzle fit together. The conference highlighted for me the way in which researchers working in quite disparate fields all contribute to gaining a comprehensive understanding of the nature of the link between genes and behaviour.

By the end of my stay I had developed both a greater understanding of the different approaches relevant to the study of behavioural phenotypes and a real affection for the city of Barcelona. I look forward to exploring both areas further as I feel that there is much more to be discovered.

Publications by Members

Following is only a very partial list:

- Barry RJ, Leitner RP, Clarke AR, Einfeld SL. Behavioral aspects of Angelman syndrome: a case control study. *Am J Med Genet A* 2005; 132: 8-12.
- Burd L, Freeman RD, Klug MG, Kerbeshian J. Tourette syndrome and learning disabilities. *BMC Pediatrics* 2005; 34: 1.
- Charman T, Howlin P, Berry B, Prince E. Measuring developmental progress of children with autism spectrum disorder on school entry using parent report. *Autism* 2004; 8: 89-100.
- Charman T, Taylor E, Drew A, Cockerill H, Brown JA, Baird G. Outcome at 7 years of children diagnosed with autism at age 2: predictive validity of assessments conducted at 2 and 3 years of age and pattern of symptom change over time. *Child Child Psychol Psychiatry* 2005;46: 500-513.
- de Ravel TJ, Thiry P, Fryns JP. Follow-up of adult males with chromosome 18p deletion. *Eur J Med Genet* 2005; 48 :189-93.
- Descheemaeker MJ, Ghesquiere P, Symons H, Fryns JP, Legius E. Behavioural, academic and neuropsychological profile of normally gifted Neurofibromatosis type 1 children. *J Intellect Disabil Res* 2005; 49 (Pt 1): 33-46.
- De Gregorio L, Jinnah HA, Harris JC, Nyhan WL, Schretlen DJ, Trombley LM, O'Neill JP. Lesch-Nyhan disease in a female with a clinically normal monozygotic twin. *Mol Genet Metab* 2005; 85: 70-7.
- de Vries P, Humphrey A, McCartney D, Prather P, Bolton P, Hunt A; TSC Behaviour Consensus Panel. Consensus clinical guidelines for the assessment of cognitive and behavioural problems in Tuberous Sclerosis. *Eur Child Adolesc Psychiatry* 2005; 14: 183-190.
- Didden R, Korzilius H, Duker P, Curfs L. Communicative functioning in individuals with Angelman syndrome: a comparative study. *Disabil Rehabil* 2004; 26: 1263-1267.
- Einfeld SL, Andrews D, Selig R, Williams G, Smith C. Schizophrenia in a 5-year-old. *Aust N Z J Psychiatry* 2005; 39: 312. [Letter, case report]
- Fisch GS. Syndromes and epistemology I: Autistic spectrum disorders. *Am J Med Genet A* 2005; 1; 135: 117-119.
- Freeman RD, Groenvelde M, Kozak FK. Hearing disorders. In *Cambridge Encyclopedia of Child Development*, ed. Brian Hopkins & Ron Barr, Cambridge University Press, pp. 437-441.
- Freeman RD. "Diagnostic criteria for Tourette's." Letter, *Journal of the American Academy of Child & Adolescent Psychiatry* 2005; 44: 209-210.
- Burd L, Freeman RD, Klug MG, Kerbeshian J. "Tourette syndrome and learning disabilities." *BMC Pediatrics* 2005; 5: 34.
- Goodlin-Jones BL, Tassone F, Gane LW, Hagerman RJ. Autistic spectrum disorder and the fragile X premutation. *J Dev Behav Pediatr* 2004; 25:392-398.
- Howlin P, Goode S, Hutton J, Rutter M. Adult outcome for children with autism. *J Child Psychol Psychiatry* 2004; 45: 212-229.
- Howlin P, Karpf J. Using the social communication questionnaire to identify 'autistic spectrum' disorders associated with other genetic conditions: findings from a study of individuals with Cohen syndrome. *Autism* 2004; 8: 175-182.
- Howlin P, Karpf J, Turk J. Behavioural characteristics and autistic features in individuals with Cohen Syndrome. *Eur Child Adolesc Psychiatry* 2005; 14: 57-64.
- Mathijssen IB, Fryns JP, Devriendt K, Sznajder Y, Van Eygen M. Iris heterochromia: a variable feature in Verloes-Koulistcher-oral-acral syndrome. *Am J Med Genet A* 2005; 132: 338-339.
- Moss J, Oliver C, Hall S, Arron K, Sloneem J, Petty J. The association between environmental events and self-injurious behaviour in Cornelia de Lange syndrome. *J Intellect Disabil Res* 2005; 49 (Pt 4): 269-277.
- Murphy KC. Annotation: velo-cardio-facial syndrome. *J Child Psychol Psychiatry* 2005; 46: 563-571.
- O'Malley K, Huggins J. Suicidality in adolescents and adults with fetal alcohol spectrum disorders. *Can J Psychiatry*. 2005 Feb;50(2):125. [Case Report]
- Prather P, de Vries PJ. Behavioral and cognitive aspects of tuberous sclerosis complex. *J Child Neurol* 2004; 19: 666-674.
- Schrander-Stumpel CT, Spruyt L, Curfs LM, Defloor T, Schrander JJ. Kabuki syndrome: Clinical data in 20 patients, literature review, and further guidelines for preventive management. *Am J Med Genet A* 2005; 132: 234-243.
- Schwartz PH, Tassone F, Greco CM, Nethercott HE, Ziaieian B, Hagerman RJ, Hagerman PJ. Neural progenitor cells from an adult patient with fragile X syndrome. *BMC Med Genet* 2005; 6: 2.
- Streissguth AP, Bookstein FL, Barr HM, Sampson PD, O'Malley K, Young JK. Risk factors for adverse life outcomes in fetal alcohol syndrome and fetal alcohol effects. *Dev Behav Pediatr* 2004;25:228-238.
- Tuffrey-Wijne I, Hollins S, Curfs L. Supporting patients who have intellectual disabilities: a survey investigating staff training needs. *Int J Palliat Nurs* 2005; 11: 182-188.
- Visser JE, Harris JC, Barabas G, Eddy GE, Jinnah HA. The motor disorder of classic Lesch-Nyhan disease. *Nucleosides Nucleotides Nucleic Acids* 2004; 23: 1161-1164.
- Vogels A, Fryns JP. Age at diagnosis, body mass index and physical morbidity in children and adults with the Prader-Willi syndrome. *Genet Couns* 2004; 15: 397-404.
- Woodbury-Smith M, Klin A, Volkmar F. Asperger's syndrome: a comparison of clinical diagnoses and those made according to the ICD-10 and DSM-IV. *J Autism Dev Disord* 2005; 35: 235-240.

From the Editor

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Roger Freeman, 2005