

NEWSLETTER

Society for the Study of Behavioural Phenotypes

Number 11

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Annual Review

This year is our Silver Anniversary. Twenty-five years ago, in 1971, William Nyhan coined the phrase “behavioural phenotypes” in reference to the extreme behaviour disorders he witnessed in certain major biological genetic disorders, particularly the Lesch-Nyhan Syndrome to which he gave his name. Behavioural phenotypes as an idea might have hit its quarter century this year, but our Society is, of course, rather younger. So what is the news of the SSBP this year?

First of all, membership is up. The growth is modest, but it is there, and the trends we have seen in recent years continue. Most pleasingly, we are receiving more enquiries outwith the UK. This has been a major element in our recruitment strategy in recent times, and it is gratifying to see some fruit in this connection. The recent meeting in Maastricht has obviously been of enormous influence here. We look forward to further growth arising from this year’s meeting in the Republic of Ireland. If we were to select “target groups” for recruitment now, it seems to

me that there are two particular specialities we should bear in mind. These are Genetics and Child Health. For, while we have grown in number in such areas as Psychology and Psychiatry, we have not been so successful in the two former. Some campaigning in both of these key fields is now a high priority.

Secondly, there has been something of an explosion of publication by our members and colleagues. There is scarcely a major journal which might conceivably have an interest in our work which has not published at least one informative review on behavioural phenotypes. As an outcome measure of sorts we can take some satisfaction here, for it is one of our declared aims to see knowledge and information on behavioural phenotypes be advanced and disseminated. In all of this the Society’s own role as an independent publication house continues. The sales of our collections of abstracts of previous conferences continues, along with our other review materials and research tools. This year has also seen the publication of the first book on

behavioural phenotypes, which I am pleased to say seems to be enjoying some success. How quickly will the first revision be required?

This year's major event is now fast approaching. Our international meeting in Dublin (14th - 16th November) symbolises a new direction in the Society's development. I refer here to the focus on Clinical Treatment and Case Management. By taking this line, in addition to providing a forum for update and stimulation of new findings in Behavioural Phenotypes, we can confidently look forward to a most successful meeting. And the next twenty-five years of growth of Behavioural Phenotypes.

Gregory O'Brien



Treasurer's Report

The audited accounts for 1st September to 31st August 1995 were presented at the Annual General Meeting of the Society held in Edinburgh on Thursday,

16th November 1995. It was reported at that time that there had been an excess of expenditure over income of £2, 159 in the previous financial year. The total amount at the end of that year held in the Society's accounts was £5, 911.

The Society had committed considerable expenditure to its first overseas conference which was held in Maastricht. This conference was a great success but it was noted at the time that it carried a greater financial risk. Whilst the finances of the Society remain adequate, there is continuing concern as the regular income obtained from subscription fees is insufficient in itself to cover the day-to-day running costs. It was noted at the Annual General Meeting that the decision to pay for secretarial support had been of great assistance to the Society as a whole but we needed to ensure that our income was sufficient to support this. Additional income is obtained through conferences and sales of catalogues and abstracts. This income cannot be relied upon on a regular basis but if conferences do not make a profit and if we do not raise additional income from these sources then the Society would be financially non-viable.

Since the Annual General Meeting we have continued to attract a small number of new members and we have also committed ourselves to sponsor a number of people to attend the Annual general Meeting to be held in Dublin in November 1996. At present the membership of the Society is 83. The meeting in Dublin has attracted considerable sponsorship from Janssen Cilag for which we are very grateful. However, we are dependent upon this

meeting making sufficient money to ensure that the Society continues to cover its costs for this and future financial years.

I would like to express my thanks to Robbie Patterson, Academic Secretary at the Section of Developmental Psychiatry at the University of Cambridge for her continuing support in the management of the Society's accounts and in the administration of the Society's affairs in general.

Tony Holland
Treasurer, SSBP

SSBP Bursaries

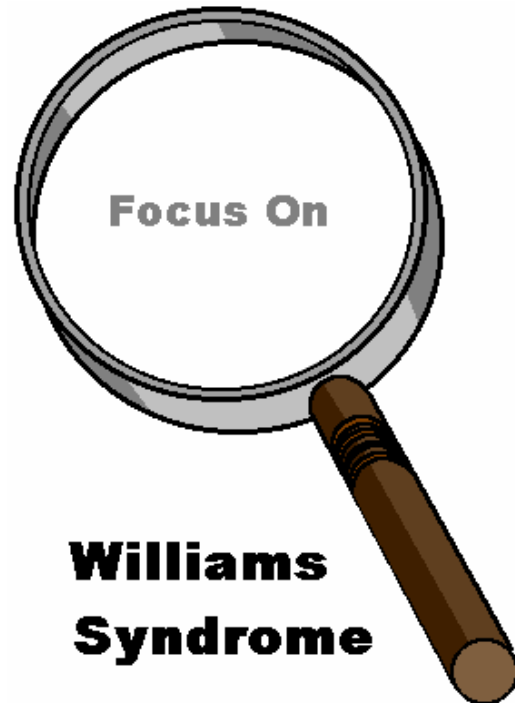
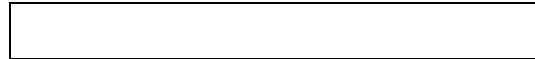
The Society is keen to encourage junior members of staff and people on limited research budgets to attend their meetings.

This year the Society will be making available 10 bursaries of £100 each to assist people to attend the Dublin conference.

Applications should be made to:

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Research Report: The Adjustment Difficulties and Needs of Adults with Williams Syndrome

Most studies investigating the characteristics of Williams syndrome have focused on the needs and difficulties of children, and until recently little was known about the long-term course of the condition. As the Williams syndrome children studied in the 1980's are now reaching adulthood, and as more adults with the syndrome are being newly diagnosed, research into their needs has become a priority for the Williams Syndrome Foundation.

Dr Orlee Udwin and Dr Patricia Howlin recently completed a two-year study of the behavioural and cognitive functioning and adjustment difficulties of adults with this condition. The study was funded by the Williams Syndrome Foundation, which identified 185 individuals aged 18-years and over through their register. A representative sample of 70-adults was selected for detailed assessment by research worker Mark Davies, who undertook detailed interviews with the individuals, their parents, and other relevant keyworkers/supervisors and/or employers, and administered cognitive and educational assessments to the adults.

Most of the adults had moderate learning difficulties; only 4% of the group had severe learning difficulties, while 10% had borderline cognitive abilities. The majority could read at a basic level; they achieved an average reading age of 9-years and 5-months for Reading Accuracy, and 8-years 6-months for Reading Comprehension. Twenty-three of the adults had been assessed 8-years previously, when in their early to mid-teens, as part of a study of the cognitive abilities and behavioural characteristics of children with Williams syndrome. Comparisons of their tested IQs on these two occasions allow us to conclude that in the case of Williams syndrome (unlike some other conditions) there does not appear to be a decline in the rate of cognitive development over time. Comparisons of reading, spelling and arithmetic scores attained at first and second testing periods revealed only modest increases in Reading Accuracy and Spelling scores, and little change in

Arithmetic test scores, suggesting that individuals with Williams syndrome do not make much more progress in their educational attainment with reading, writing and numeracy skills beyond their early teenage years.

The rates of health problems reported by parents and carers were much lower than American studies of adults with Williams syndrome have suggested. The most likely explanation is that the American studies were of small samples of individuals who were attending medical institutions because of ill health. However, it is important to note that only a minority of our sample were receiving regular, routine health checks. Thus it seems that both parents and health professionals require further information about possible health risks associated with ageing in this population, and they need to be reminded of the recommendation that all adults with Williams syndrome should have at least annual checks of cardiac and kidney function, and blood pressure.

The majority of adults in this study attended day centres, adult training centres or further education courses, and lived at home with their families. Only one had an independent part-time job and 4% lived independently; but even they needed supervision and support from parents and community workers to help them cope with the demands of daily life. Interviews with families and care workers revealed problems in many different behavioural domains, the most frequently occurring being anxiety, restlessness and distractibility, social disinhibition, irritability and anger, and preoccupations and obsessions with particular topics (eg

illness, disasters), objects (eg cars, electrical appliances) or people eg media personalities, a neighbour). In over 80% of cases these difficulties were also reported by supervisors and keyworkers to limit performance in work placement. On the other hand overactivity, which is a prominent feature in affected children, was not a problem for the adults.

In sum, many of the characteristics described as being typical in children with Williams syndrome were found to be common in our sample of adults with this condition as well. The adults continue to show high levels of behavioural disturbances, and require supervision and support with daily living and occupations. As the majority remain living at home, their families continue to shoulder the main burden of their care for many years. However, other features were less in evidence in the adults; the incidence of hyperactivity was particularly low, and physical complications were also less frequent than expected.

Following this study, we have produced booklets of advice on adults with Williams syndrome - Guidelines for Families and Professionals and Guidelines for Employers and Supervisors. These will be available shortly from The Williams Syndrome Foundation, The Little Ruin, Edge Road, Edge, Stroud, GL6 6NE. Other publications arising from this research will be available in due course.

Dr Orlee Udwin

Consultant Clinical Psychologist (Child Health)

Honorary Senior Lecturer in Child Psychology

Deletion of Elastin Gene on Chromosome 7 Reported in Williams Syndrome

Brewer, Morrison and Tolmie (1996) have published a study of sixteen children and adolescents with Williams syndrome, using the chromosome fluorescence in situ hybridisation (FISH) technique employing the elastin gene probe. In each case a fluorescent signal was detected on one chromosome 7 homologue only, indicating deletion of the elastin gene.

When the chromosomes of a child in whom an earlier diagnosis of Williams syndrome was judged to be doubtful were examined, no deletion was detected.

The authors concluded that firm clinical diagnosis had correlated with elastin gene deletion in 16/16 cases of Williams syndrome, and that detection of such hemizyosity by FISH constituted a useful confirmatory diagnostic test.

Brewer, C.M., Morrison, N. and Tolmie, J.L. (1996) Clinical and molecular cytogenetic (FISH) diagnosis of Williams syndrome. *Archives of Disease in Childhood*, **74**: 59-62.

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Conference Report: First
Congress of the European
Association for Mental Health in
Mental Retardation, September
13th-16th 1995, Amsterdam.**

This congress was organised by the European Association founded in 1993 to promote international co-operation and exchange of knowledge and expertise in the field of mental health care for people with mental handicap. The Congress attracted delegates from different disciplines with varied assessment and therapeutic approaches from all over Europe and North America.

In his opening address, Dr Anton Dosen of the Netherlands emphasised the need for appropriate training of professionals and administrators to ensure that people with mental handicap have access to the same range of mental health services as people without mental handicap. He reiterated the need to approach mental health as a health issue, and not simply an illness issue related to disruptive activity by the individual. Mental health is a pre-requisite of full normalisation.

The Congress approached this theme through keynote presentations, round-table discussions, symposia, workshops and free paper and poster presentations. The content of these activities included diagnostic criteria, assessment

instruments and methods, specific disorders (e.g. autistic spectrum disorders), treatment methods (from psychotherapy to chemotherapy) and administrative structures to deal with the mental health needs of this population. Genetically transmitted conditions (and those hypothesised as such) and their mental health implications were discussed in various fora.

Dr J. Turk presented a pre-congress course on the Fragile-X syndrome. Jeremy addressed the diagnosis and intervention implications of current knowledge of the behavioural phenotype. A keynote address on "Behavioural Phenotypes and Psychiatric Diagnosis in Persons with Mental retardation" allowed Jeremy to enlarge on the importance of the behavioural phenotype area with reference to other syndromes.

Another keynote address by Professor C. Gillberg referred to studies which suggest that the behavioural syndrome of autism is caused by medical conditions in about 25% of cases. By using the term "medical conditions" he was referring to genetically transmitted conditions such as tuberous sclerosis. However, children seen in child psychiatric clinics who are diagnosed as having an autistic spectrum disorder probably have lower rates of associated "medical conditions".

A round-table discussion covered the area of behavioural phenotypes under the title "Genetic and Biological Disorders". In this discussion, Dr S. Bernard discussed the Prader-Willi syndrome

including the aetiology and phenotype. The latter includes the well known behaviours of over-eating and associated temper tantrums if thwarted. Other mental health issues including self-injurious behaviour and an increased likelihood of psychosis in adulthood were represented from research findings. Dr Van Beeckelaer-Onnes presented the move from phenotype to genotype in the pervasive developmental disorders. This approach is receiving more attention despite the lack of hard markers. Dr R. Collacott outlined the vulnerabilities of people with Down syndrome to certain types of mental health problems, namely dementia of the Alzheimer type and depression. He related this to over-expression of the gene coding for amyloid precursor protein and a relative hyperserotonergia. The final speaker in this symposium, Dr B. Moes, presented research findings demonstrating a specific social-emotional and behavioural profile associated with the Fragile-X syndrome in adults with mental handicap. These findings included a high proportion of Fragile-X adults displaying specific behaviour belonging to the spectrum of pervasive developmental disorders. However, the core features of autism were not found in the study group of adults with Fragile-X syndrome. The adults in their group had a positive approach to their environment and to others, with average interpersonal and communicative competencies.

These findings supported the hypothesis that, though people with Fragile-X syndrome have traits that suggest an association with autism, there are marked differences that suggest

otherwise. This symposium emphasised the connection between genes and both conditions which cause mental handicap and mental health problems. This supports the importance of the Behavioural Phenotype in assessment, treatment and service planning.

In other sessions, the prevalence of autism and autistic-like conditions in people with Down syndrome (Dr D.W. Kraijee), the presentation and therapy of depression in children with Down syndrome (Dr R. Cocchi) and early intervention to promote mental health in children with Down syndrome (Dr G. Albertini *et al*) were presented in free papers.

In the closing keynote address of the Congress, Dr R. Fletcher of the National Association of the Dually Diagnosed (USA) followed Dr Dosen's opening theme and called for more co-operation between community service providers and mental health professionals and between researchers and clinicians to develop training, assessment and treatment approaches that will satisfy the special mental health needs of people with mental handicap.

On a personal note, the gathering of so many professionals of different disciplines to focus on the mental health needs of people with mental handicap provided a reassuring reminder of the growing interest in this area. Behavioural phenotypes were given due attention but I would like to see the next Congress containing more presentations on behavioural phenotypes and specific presentations in individual syndromes.

The next Congress is due to take place in
England in the Summer of 1997.

John Hillery

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