Blasts from the Past

So much has happened in just a few years that it is interesting to look at some of the responses we had from health professionals way back in what seems like the mists of time (1993).

In July '93, HRT specialist Mr. Malcolm Whitehead[1] replied to a letter from a very early member of the group concerning her teenage CAIS daughter's HRT:

Thank you for your recent letter. I regret to say that, as far as I am aware, there are no data whatsoever on testicular feminisation syndrome and HRT. My approach to my patients with this condition is to treat them as though they had undergone a premature menopause .... You will have to spread your net far and wide if you wish to form a support group.[2] The condition is uncommon and most gynaecologists/endocrinologists only have one or two patients with the condition.

In Sept '93 we sent a standard letter to a number of UK clinicians, as follows:

Dear Dr _____,
A long overdue support group has recently been formed[3] for the Androgen Insensitivity Syndrome, AIS (otherwise known as Testicular Feminization Syndrome) under the wing of the national and well-respected Contact-a-Family organisation.

I am preparing literature to send to people who have AIS, to the parents of young children with AIS, and to doctors, and I am looking for specialists in one or more aspects of the condition from whom I can obtain information. The literature would be made available to those parents (or older patients) who have a) expressed a desire for full information on the condition, and b) have already received a full and truthful diagnosis of AIS (including the genetic make-up and the nature of the gonadal tissue).

There seems to be considerable disagreement amongst the medical profession on what should/shouldn't be revealed to the parents, and particularly to the child, in this condition and there seems to be virtually no published work on the psycho-sexual outcome in these patients. I am therefore interested to know what clinicians base their decisions on, in this regard. If you are directly involved in diagnosing/caring for such patients I should be very interested to hear of the policy you adopt in regard to giving or withholding information, and what provisions, if any, are provided in your establishment for psychological support and follow-up.

Yours sincerely etc.

Some of the more interesting/revealing/ useful responses are reproduced below.

A professor of paediatric research, Prof. Martin Brobow,[4] wrote:

Thanks for the note concerning the AIS Support Group. I am glad to hear of this initiative. It is a very sensitive and often poorly handled condition. I am not aware of anyone who has produced good objective data on optimum methods of handling these situations. I was very interested in the condition many years ago, but do not now see patients myself. I have, however, taken the liberty of passing copies of your letter to a few people in my School who may have ideas, and advising them to write directly to you if they have anything to say. *[They didn't – Ed.]*

Recently retired gynaecologist and intersex expert Prof. J. S. Scott[5] responded:

Thank you for your letter. I wish you success with your AIS support group though I do hope you are not under-estimating the difficulties. Some of these I think relate to the fact that AIS is not an entity and overlaps with other forms of anomalous sexual differentiation.[6]

In selected cases, it was for long my practice to introduce an individual with the condition, who was at a stage of having difficulty in coming to terms with the situation, to another with the same type of problem who was happily adjusted to life and flourishing in a rewarding career – a rather more personal and intimate form of mutual support than that offered by a group such as yours but with similar intentions .... I agree that there is considerable difference of opinion within the medical profession as to exactly what should be told to the individuals. Personally, I think that to a large extent this should depend on the individual patient. For example, I would not give the same explanation to a low IQ person as to one who had an honours degree in medicine or human biology – and I have been faced with cases in both these categories[7]....[8]

.... With regard to psychological support and follow-up, my practice was to see these patients *personally* until such time as they were settled into a marital – or so-called stable – relationship (if there is a difference), *or* I was satisfied that they had become completely adjusted to their condition and were settled in the single state. Even then, discharge would be with the proviso that they could at any time approach me directly if the situation should change.[9]

I think that one of the most important things to appreciate and get across is that AIS individuals, even though in the present state of knowledge they are not going to have natural children of their own, can frequently have very fulfilling lives and flourish in high level business or academic posts. They may also shine

1.Snr. Lecturer/Consultant Gynaecologist, King's College Hospital, Bessemer Rd., London SE5 9PJ.

2.We have. No problem!

3.It was in 1993 that the editor met the founder member (who had acted alone since 1988) and the group was really 'launched'.

4.Paediatric Research Unit, Division of Medical and Molecular Genetics, Guy's Hospital, London Bridge, London SE1 9RT.

5.Author of Chapter 16 (Intersex) in Progress in Obstetrics and Gynaecology, Vol. 5, Ed: Studd J., Churchill Livingstone, Edinburgh, 1985. Had retired from Chair in Obstetrics and Gynaecology, Leeds General Infirmary.

6.Does it? CAIS seems pretty well-defined. Maybe there could be some overlap in PAIS?

7.There seems to be an assumption on the part of some clinicians that unless the patient has a high IQ and/or medical knowledge, she is not entitled to the openness and information that essentially provides the framework for *emotional* acceptance.

8.At this point he commented on adverse parental/ GP reactions (already quoted – together with similar comments from Derek Johnston, Cons. Paediatrician, University Hospital, Nottingham – in "Parental Attitudes" in $A^{L}I^{A}S$ No. 1, Spring 1995).

9.With no disrespect to this gynaecologist's good intentions, it is interesting to note that in the study of Langer et al (see " Vorsprung durch Technic?" in $A^{L}I^{A}S$ No. 7, Spring 1997) it is reported that "*in none of the cases .... did patients mention the gynaecologist as the person they would turn to first.*"

at various forms of athletics though with the risk, if not prudently advised, of suffering distress when at the level of international competition (sex testing in athletics should be banned in my view). AIS girls are often very handsome[1] and socially popular with both sexes. Marriage is frequently successful.

AIS paediatric/genetic expert Prof. Ieuan Hughes[2] wrote:

Thank you for your circular concerning the Androgen Insensitivity Syndrome (AIS) .... We have been involved with the investigation and management of this condition for several years. We also have a major research programme underway which examines the molecular genetics of the disorder, both the complete and partial forms .... I have also had a letter from Mrs. F_____ who I understand is helping with the establishment of the support group. She mentions 'carrier detection' and that is something we undertake on a fairly regular basis. As with all carrier detection work we prefer [first] to know the mutation in the index case[3] rather than embarking on a non-focused search [in the female relatives].

I am very interested in deciding what advice the medical profession should be giving to these patients, particularly when they reach young adult life. Several of my colleagues have suggested that some form of National Centre would be useful in that regard and if that were the case, I think we would be able to offer considerable support here in Cambridge. We already have a repository of samples[4] from nearly 200 families accompanied by quite detailed information about clinical and genetic items ....

Later, one of his researchers wrote:
.... We will publicize your support group by sending out a letter each time we are sent samples, which happens quite regularly[5]....

Reproductive biology researcher Dr. John Bancroft[6] wrote:

I was interested to receive your letter about the support group for Androgen Insensitivity Syndrome. It is true that very little has been published in the literature about the sexual development and behavioural consequences of this rare condition. I am rather embarrassed to say that a few years ago (I think it must have been around 1980) I interviewed a series of AIS women that were patients of Professor Polani at Guy's Hospital and also a number of controls. I must have seen about 15 AIS subjects and I never completed the study because of difficulty getting an appropriate control group .... They were all, of course, people who knew of the diagnosis although I do remember them telling me how they had reacted and how the diagnosis had been presented to them in various different ways.

Although I do not currently see patients with this condition, mainly because it is very rare and I do not run an endocrine clinic as such, I would have reasonably clear ideas how I would approach the issue of information to parents and the patient. My main uncertainty would be the timing of this as far as the AIS individual was concerned. My general line would be to emphasise the biological fact that whatever our chromosomes might be our development along male or female lines is very largely determined by whether we are exposed [and are sensitive to] testosterone during development. If we are not, then we are in a sense programmed to develop along female lines. Although in AIS the situation is not quite as simple as that because of the effects of Mullerian inhibiting factor,[7] it is, I think, a valid conclusion to make that an AIS person is almost to all intents and purposes a normal female except for her lack of uterus and her associated infertility and lack of menstruation.[8] I don't think the abnormality of the situation can be avoided but the essential femaleness of her being can, I think, quite legitimately be emphasised.

What I have learnt from talking to people with various types of abnormality of development is the sense of being 'nowhere' in terms of gender development and identity and I believe it is fundamentally important to correct that misunderstanding, even though they still have to come to terms with the consequences such as infertility which is unavoidable.

Paediatric endocrinologist Dr. Christopher Kelnar[9] responded:

Thank you for sending me the details about the AIS Support Group. I am sure that this will be a useful organisation. I have a number of families under my care with this problem and I think many, although perhaps not all of them, would be keen to know of others in the same situation. The question about when, or indeed whether, it is ever appropriate to tell a 'girl' or 'woman' that, for example, she has a male genetic make-up and even that she had testicles removed in infancy, is a very difficult one and certainly most families which I have been involved with have, I think rightly, been of the view that this is never appropriate. That is not to say that extensive and appropriate counselling should not be given in terms of infertility, etc.

You are right in saying that there is virtually no published work on the psychosexual outcome in these patients and this is obviously an important area to know about .... It is certainly an area we are keen to do more research in, although obviously it is a difficult one from many points of view[10].... I work very closely

1.Unfortunate choice of word in these circumstances? Maybe this alone indicates why doctors don't necessarily make good counsellors, no matter how well-meaning?

2.Prof. of Paediatrics, Addenbrookes Hospital, Hills Rd., Cambridge, CB2 2QQ.

3.The individual who actually has AIS.

4.We are not sure what samples this refers to. Adult endocrinologist Dr. Gerard Conway confirms that all AIS diagnostic genetic tests can be done on blood samples, or even on mouth brushes, and that genital skin samples, for example, would be of academic/research interest only – in seeing exactly how the tissue androgen receptor is affected by any given gene mutation.

5.One mother told us she was angry to find out that her CAIS child's surgeon had been approached directly by a research group, with a request to remove a tissue sample whilst her daughter was under anaesthetic for gonadectomy. The parents themselves had not been consulted directly.

6.Then head of the MRC Reproductive Biology Unit, Behaviour Research Group, Royal Edinburgh Hospital, Edinburgh, EH10 5HF (now Director, Kinsey Institute for Research in Sex, Gender and Reproduction, University of Indiana, Bloomington, Indiana, USA).

7.See "MIF Matters" in A[L]I[A]S No. 8, Summer 1997.

8.What about the issue of severe vaginal hypoplasia in some patients?

9.Consultant Paediatrician, Royal Hospital for Sick Children, Sciennes Rd., Edinburgh, EH9 1LF.

10.Yes, especially if you haven't told the patient her diagnosis, so she can't give fully-informed consent to taking part in any such study!

with colleagues in the Department of Child and Family Psychiatry, who are able to provide expert counselling in conjunction with myself and colleagues from Clinical Genetics, and I would certainly see this as an important part of one's role with these families.[1] I think that some problems arise when they move out of the paediatric age group, in terms of finding appropriate adult physicians who have insight into their problems, but that is another story.

Dr. David Grant,[2] an expert in intersex conditions in babies/children, did an excellent job in commenting on a multi-chapter 'AIS Review' document drafted by a member of the support group. In a letter of Aug '94 he wrote to her:

.... I am sorry that you had difficulty with my handwriting – I have tried to clarify my comments. The one I feel most strongly about is [your section] "The Parents' Role in CAIS" and the need to have open discussion without any 'dark secrets'. I think that this really needs to be stressed as strongly as possible ....

Consultant gynaecologist Mr. Keith Edmonds[3] wrote in Feb '95 (after we had approached the nursing sister who runs his department's support group for vaginal hypoplasia/aplasia patients,[4] asking permission to list her group in our first newsletter):

Thank you very much for writing to Miss Little and I wonder if I might reply on her behalf. I am a little concerned about the wording of your description of our clinic. The policy within the department is certainly not to tell women with AIS of their chromosomal or gonadal constitution unless they ask.[5] We always inform women of the gonadectomy that they had in childhood and the reason for the surgery.[6] I think this is important because I do not want our group or our policy to be misunderstood.

See also "Psychological Paternalism" in A^{L}I^{A}S No. 3, Winter 1995.

Origins of "Don't Tell"

Two AIS women, whose extended email discussions appear later in this issue (see "1) Reading the Words" on page 117), and others had the following discussion with an adult endocrinologist in mid '97:

The UK CAIS woman wrote:
Dr. Gerard Conway[7] tells me he has been doing a search of the literature to try and find out who was the first person to firmly advocate in print that an AIS patient should not be told the full details of her diagnosis. He seemed to be homing in on John Money but we discussed this and neither of us were too clear on this since Money does go to some length to suggest suitable ways of conveying quite detailed diagnostic information even if he recommends talking about a 'broken X' rather than a Y chromosome etc., and he has written a lot of good stuff on psychological counselling methods.

Our US representative replied:
When I was rooting around in medical libraries almost 20 years ago, trying to find out 'the truth' about my condition, I remember reading stuff that predated Money which said the patient would kill herself if she found out.

The PAIS woman in the US replied:
I wouldn't pin this on Money, who seems to be the scapegoat for nearly everything nowadays. As you noted, he does advocate trying to candy-coat the facts with that 'broken X chromosome' story, but he does not advocate actually concealing them. I looked up one of the early papers on AIS, by Morris.[8] This is considered one of the pioneering articles on the condition, so much so that AIS has sometimes been called 'Morris Syndrome'. Morris wrote an additional paper in the same journal in 1963.[9] On page 1209 it says "*It goes without saying that it would be unwise to inform the patient of the true state of affairs ....*"

I know how much trouble this kind of recommendation has caused, but to be fair to Morris, one needs to realize that he probably considered this a kinder alternative to telling the patient that she was 'really' a man, or even to telling her she should try to live as one, which was sometimes done in the early twentieth century. The text immediately following the above reads "*.... To go one step further, in view of the long and happy marriages some of these patients have had, it seems unfortunate that in some cases the doctor has felt it necessary to tell the patient she must never marry. It seems only necessary to state that child-bearing is impossible. Whatever the legal, moral or religious view of this unusual circumstance may be, where a male is married to a female with male gonads, the obvious humane attitude is not to interfere and by such meddling produce a psychiatric casualty and perhaps a suicide.*"

Dr. Conway replied:
Many thanks .... I too conclude it is Morris. The woman he refers to as having a suicidal psychosis had a tumour and was not likely to have known the [AIS] 'diagnosis' so it was never a good paradigm on which to base the instruction. I agree he was never to know the repercussions of this instruction, as he had only just grasped the physical features of the condition. Rather than 'pin it on him', I think it is revealing of the medical profession how avidly they took up the instruction and stuck to it without question for 40 years. Medics were able to avoid responsibility, felt relieved [at not having to face a difficult situation], and so they spread the word .... Money goes some way to undoing the wrong by saying that each woman should be told the degree of detail deemed appropriate (quite how the deeming was to be done is not clear).

Our 49 year-old UK PAIS member (see "Some Old friends" in A^{L}I^{A}S No. 8,

1.But counselling on infertility issues only?

2.Honorary Snr. Lecturer, Institute of Child Health, 30 Guilford St., London WC1N 3JH.

3.Consultant Obstetrician and Gynaecologist, Queen Charlotte's and Chelsea Hospital, Goldhawk Rd. London W6 0XG.

4.A group which meets twice a year to support vaginal hypoplasia patients (other conditions as well as AIS) in their use of vaginal pressure dilation (see "Support Groups" in A^{L}I^{A}S No. 1, Spring 1995).

5.You mean like not telling someone they have a notice on their back saying "I am an idiot" unless they ask you whether they've got a sign on their back saying "I am an idiot"?

6.How can you truthfully inform someone of the reason for surgical removal of an organ if you have a policy of not telling them the true nature of the organ that's removed?

7.Senior Lecturer in Medicine, Dept. of Endocrinology. The Middlesex Hospital, Mortimer Street, London W1N 8AA.

8.Morris: The syndrome of testicular feminization in male pseudo-hermaphrodites. Am. J. Obs. and Gyn., Vol. 65, No.6, June 1953, pp.1192-1211.

9.Vol. 87, No.6, pp.731-748.