

Document for BAPS from the Ethics Advisory Committee

Androgen insensitivity syndrome:

How much to tell families of girls with inguinal herniae regarding the possible diagnosis.

Background:

Androgen Insensitivity Syndrome (AIS) is a relatively rare X-linked abnormality of the development of the genitalia resulting in an essentially external female phenotype but with testes as gonads and no uterus or Fallopian tubes and a short vagina. The condition usually presents with primary amenorrhoea. However, on some occasions the first manifestation may be the finding of inguinal herniae in a prepubertal girl. Most authors consider that unilateral herniae do not represent a high risk of being associated with the syndrome but that the presence of bilateral herniae necessitates investigation.

Certain specialists dealing with anomalies of gender differentiation have suggested there is a high chance of AIS being present even with a unilateral hernia in a girl (Viner R M et al Arch Dis Child 1997; 77: 305 – 309). This is not universally agreed but the true incidence is not known.

This paper sets out not to decide upon clinical decisions as to who should be investigated for AIS but what information should be imparted to families if the investigations are considered by the treating clinician as advisable. There may be need for a research project to define the incidence of AIS presenting with a hernia or herniae.

Recommendations from the debate:

1. Covert investigations should not be undertaken. However, it must be up to the clinician to decide what detail of explanation is appropriate for each family. For example it may well be deemed appropriate to say that 'with herniae in girls there may be problems with internal structures and though rare one should exclude such problems'. Any specific questions from the family would need to be answered fully. For other families it may be appropriate to define fully the issues from the outset. A child of 'Gillick' competence may not wish her parents to know and this should be respected.
2. Presuming that all girls with bilateral inguinal herniae should be assessed (and have agreed) then the clinician should decide upon the optimum least invasive test compatible with achieving the answer with sufficient certainty for acceptable clinical practice. In the paper quoted above it is suggested that chromosome analysis should be performed on all such children. The clinician must decide whether this would cause undue anxiety in which case a less emotive investigation such as ultrasound examination of the pelvic organs might be deemed acceptable. (This

investigation is believed to be highly sensitive and specific in infancy and children from the age of 5 years but may be less accurate in the intervening years. It is currently being evaluated.) It remains the clinician's responsibility to decide whether or not to investigate for a contralateral hernia if only one is evident at presentation.

3. Evaluation at operation by identifying the presence of a Fallopian tube on one or other side provides conclusive evidence that AIS does not exist in that patient. Previous practice of performing a rectal examination to feel for the uterus if a Fallopian tube is not identified is not now considered ethical, even under anaesthesia.
4. The clinician must be aware of the ethical dilemma of investigating or not investigating the patient for AIS:
 - The diagnosis if proved may not lead to any immediate benefit to the patient or alteration in management. It could be argued for some families that investigations under such circumstances are not in the patient's interest. It must be remembered that there have been reported rare instances of prepubertal malignancy in the gonad of these patients.
 - The converse opinion may be more relevant where withholding knowledge of a possible serious condition may be unacceptable.
 - The clinician needs to be aware of the family history for example of first or second degree female relatives without children.
 - The established diagnosis of AIS must influence sex education of the individual.
 - The timing of imparting clinical information is always of concern to clinicians and may be particularly difficult in this instance. In most instances the intention to undertake investigations should be intimated and discussed at the earliest opportunity.

A.E.MacKinnon 15/11/98

AIS Recommendation

For comments on the site email webmaster@baps.org.uk

AISSG UK Note:

The above article was originally at: <http://www.baps.org.uk/documents/AIS.html>