SECTION 6
Counselling/education/screening

6.001
Defining the skills and knowledge base of genetic nursing practice
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The Delphi technique was used to determine the opinions of an expert practitioner panel regarding the core components of genetic practice and the knowledge base, skills and personal attributes required to deliver an optimum service. The items generated by the panel were the subject of iterative review to establish a consensus view. 96% consensus was achieved over three rounds. The main components of practice identified included interaction with clients involving information transfer and counselling around issues concerning psychosocial impact. Educational and research components of practice were also valued highly. Commensurate knowledge and skills were identified to support this practice most of which were considered to be embedded in prior clinical experience in other settings which could be developed as a practitioner in the specialty. Academic study to at least first degree level with the goal of a Masters degree was identified. This would fit in with the designated UKCC education requirements for specialist and advanced practitioners.

The results of the study can be used as a basis upon which to develop appropriate specialist educational programmes for future practitioners and to define competencies and standards which could be used to guide specification and appraisal of nursing contributions to genetic services.

6.002
The objectives of genetic counselling: a comparison of patients' and professionals' views
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In order to evaluate the effectiveness of a service, there needs to be agreed objectives. The aim of this study is to establish the extent to which there is a consensus about the objectives of genetic counselling amongst those who provide, use and purchase this service. A second aim is to seek the views of these groups about whether any differences found constitute a problem and, if so, what should be done to increase agreement. Questionnaires were sent using the Delphi technique to seven groups: clinical geneticists, genetic nurse counsellors, general practitioners, public health consultants, genetic centre outpatients, Genetic Interest Group members and medical ethicists. Of the 209 questionnaires sent, 121 were returned. Responses were coded into 15 categories with high inter-rater reliability. The five categories most frequently judged to be the most important objective were: give information (34 ranked 1), facilitate decision-making (22), assess risk (18), achieve understanding (14) and give support (9). The distribution of these categories varied across groups. Respondents have been asked to comment on whether they consider these distributions to be a consensus and to rank order the five most frequently cited objectives. The results of the second and final rounds of questionnaire circulation will be presented, and their implications discussed.

6.003
Genetic counselling: The psychological impact of meeting patients' expectations
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We know little about patients' expectations of genetic counselling, the extent to which their expectations are met and whether meeting patients' expectations is associated with better patient outcome. These questions were addressed in a study of 131 consultations. Patients came to genetic counselling expecting information (79%), explanation (53%), reassurance (50%), advice (50%) and help in making decisions (30%). The majority got what they were expecting: 74% had their expectation for information met, 56% had their expectation for explanation met, 60% had their expectation for reassurance met, 61% had their expectation for advice met and 73% had their expectation for help with making decisions met.

Meeting patients' expectations for reassurance and advice was associated with better emotional outcomes. Those who were expecting reassurance and got this were less concerned after the consultation than those who reported not having been given reassurance after expecting it. Those who were expecting advice and got this experienced a greater reduction in anxiety than those who did not feel they had been given advice after expecting it.

Meeting patients' expectations for information, explanation and help with decision-making was not associated with better outcomes. Explanations for these results and implications for the practice of genetic counselling will be discussed.

6.004
Educating the public about genetics: a niche for a high street Gene Shop.
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Public education forms an important strand of the human genome project.

This includes widening discussion of the moral and ethical aspects.

While many agencies are involved in genetic education, the vast majority, including clinical genetics departments, are patient-oriented. A non-commercial high street Gene Shop, as a community outpost of the hospital unit, should fill a niche in public education. A shop, such as the one for which we have funding through the EC, should increase general knowledge in an area which the public finds interesting if a little frightening. Staff will be from the department's field-workers, specialist health visitors and clinical geneticists, all used to fielding enquiries from the public.

Counselling as such will not take place but the shop will carry non-directive general information across the widest spectrum possible of human genetics Medical and paramedical knowledge should also increase. During a successful pre-run held in the hospital during Science Week in March, 260 people visited the shop. Most came with general enquiries. All felt that there was too little easily available information about genetic conditions. Placing the "shop" on a busy street, away from a clinical setting will allow a clearer assessment of its future role in public education.
6.005
Survey of attitudes of pregnant women towards Down's syndrome screening in ante-natal clinics
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Investigation and critical assessment of antenatal screening for Down's syndrome was carried out in response to concerns raised by health professionals about the way the service was delivered in Mid Glamorgan. Survey of health professionals revealed disparate policies and lack of staff training. Survey of mothers was also carried out to recommend a policy and implement change. Method used was semi-structured interview of 20 weeks pregnant mothers. Forty two women younger than 35 years were interviewed (20 screened; 9 unscreened; 5 refused screening; 3 terminated and 5 who delivered a Down's syndrome baby).

Only 5% of mothers refused screening, they were better educated and of higher social class. Tests were presented as routine, but all mothers wanted to be offered the choice to be screened. Twenty three percent of them, who were better educated, of either low or high social class, would not terminate an affected foetus. Organisation of second trimester termination of pregnancy, and staff communication skills, especially in delivering risk estimate were criticised. Recommendations were made to implement The Royal College of Obstetricians and Gynaecologists's minimum standards for contracting including individually-tailored information packages, offering women the choice to be screened and training a "screening midwife" in each unit.

6.006
Is A Clinical Genetics Service better than no service at all? A "Controlled trial".
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A population survey of Huntington disease (HD), was undertaken in both Northern Ireland (NI) and the Republic of Ireland (ROI) from 1990-1994. Northern Ireland has had a genetics service for over 25 years; the Republic of Ireland has had no genetics service. The difference in outcomes of the survey serve as an indicator of the relative utility of genetic services.

Methods: Multiple ascertainment methods were used: postal surveys of all GP's, secondary case tracing and record linkage.

Results: Minimum population prevalences were 6.4x10-5 (NI), and 2.9x10-5 (ROI). GP response rates were 60-70% (NI) and 5% (ROI). Multiple ascertainment of previously recorded cases occurred in NI; most ROI cases were recorded only from one source. Virtually total ascertainment was obtained in NI based on an indirect mutation rate of 1.0x10-6 (3%) which is comparable to recent molecular data. Ascertainment in ROI is therefore grossly incomplete.

Conclusions: Gathering data is facilitated by a genetics service. Functions of a service include organisation of genetic registers, tracing families, liaison with other specialities for diagnosis, screening, and education, and enabling patients to obtain information about the disease they may have, so that diagnosis is accurate and life choices can be made from an informed standpoint.

6.008
Syndrome Diagnosis - The Parent's View
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We have completed a study using in-depth interviews and psychometric tests in a group of 30 mothers of children aged 5-10 with multiple congenital anomalies (17 with a syndrome diagnosis and 13 without). We were interested to investigate to what extent the presence or absence of a dysmorphic diagnosis has made a difference to the child and family. All the mothers in the diagnosed group stated that they were pleased to have a diagnosis for their child, and 77% of the mothers in the undiagnosed group wished they had a diagnosis. While 50% of mothers did describe the diagnosis as a label, many saw this as useful. 70% of the mothers in the diagnosed group felt that a diagnosis had helped them to come to terms with their child's problem. In the undiagnosed group, 92% (compared with 41% in the diagnosed group) felt they still had unanswered questions and half of these felt the knowledge must exist to answer them. More mothers in the undiagnosed group went on to have further children after the birth of their affected child and this was not dependent upon degree of risk. These and other findings will be presented.

6.009
Content and provision of genetic counselling: general practitioners' knowledge and opinions
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Debate is continuing as to how the anticipated increase in demand for genetic counselling will best be met. Some feel that it will be necessary for primary care to become more involved than it is at present. The majority of discussion so far has been amongst those involved in clinical genetics and those general practitioners (GPs) with a special interest in this area. The aim of this study is to assess the views of a sample of GPs as a contribution to this debate. A postal questionnaire was devised and 300 randomly-chosen GPs from four health districts were ascertained for study participation. Initial results suggest a good level of knowledge of the services provided by a clinical genetic service. When asked to consider five imaginary patient scenarios, most GPs indicated that they already provide many elements of genetic counselling for their patients. The majority felt that they would still refer on for specialist confirmation of the advice they had given. When asked about the future involvement of GP practices in the provision of some genetic services such as population screening, the majority felt that the main limitations to this were time and knowledge and that these would be difficult to overcome.
6.010
Audit of Telephone Activity by Genetic Nurse Specialists
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Face to face contact with individuals and families attending a Clinical Genetics Service is well documented and acknowledged. It was felt that telephone counselling activity, whilst time consuming, is less well recorded and is not represented within contract activity. In an attempt to obtain some baseline information about the extent and the purpose of telephone contact with families so that a more complete record of the professional input to referred families might be provided, an audit of this activity was undertaken by genetic nurse specialists attached to the Nottingham service. For one week all telephone activity was recorded and the results have given an indication of the number of calls made and received, the length and purpose of the calls and the time of day when this activity is greatest. We plan to use this as a baseline for future audits and as a focus for more complete documentation of work done with families as part of contract activity.

6.011
Genetic Registers for Primary Care
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90% of UK Practices are computerised (latest figures from NHS Executive). A small number of GPs are paperless, the majority record on computer and in the Lloyd George records. 82% of Practices have a modern reflecting the NHS drive towards electronic links, with Health Authorities 70%, path labs 14% and Hospital Departments 3%. 1 in 10 GPs use the Internet. Primary Health Care Teams use onscreen protocols for new patient registration checks, diabetic, asthma care and well person screening. Brooklands Medical Practice developed a protocol for antenatal care, incorporating information on screening for Downs, Neural Tube Defect and CF Carrier Screening. Increasing use of the computer in the consulting room for recording consultations, prescriptions, background health data, previous medical history, lab and X-ray reports will result in extensive patient data bases and the ability to access instant genetic registers using precise Read codes. New generations of medical computer programmes will enable family history data to be linked within families, alerting GPs to significant family history. Developments in Primary Care need to be matched by initiatives within departments of Medical Genetics, so that use is made of the links for developing protocols for appropriate genetic referrals and informing primary care of recent advances, with the implications for screening and patient management.

6.012
Investigation and Counseling of a Homologous Robertsonian Translocation Carrier with a
Cytogenetically Normal Offspring
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A female carrier of a homologous Robertsonian translocation for chromosomes 21 was ascertained after the birth of a baby girl with Down syndrome. Chromosome analysis of her 18 month old son showed a normal male karyotype. The parents insisted that they were the natural parents of both children and requested counselling on their risk of Down syndrome in future pregnancies. Possible aetologies of the normal male karyotype were investigated. Division of the Robertsonian translocation, mosaicism with a normal cell line in the mother or abnormal cell line in the son, nullisomic ova fertilised by disomic sperm or compensatory isodisomy of the paternal chromosome 21 were evaluated by cytogenetic banding studies and molecular microsatellite analysis. Discrepancies in the inheritance of microsatellite alleles identified the child as having no genetic relationship to either parent. Directive, sensitive counselling was essential to identify the true risk of Down syndrome to all offspring without confronting the issue of apparent adoption.

6.013
Improving information giving in clinical genetics: the
effectiveness of tape recording the clinic consultation
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Thirty seven clients who attended a clinical genetic consultation, between November 1995 and February 1996, participated in a randomised, controlled, experimental design study to investigate one method of improving information giving in clinical genetics clinics. All clinic consultations were audio-tape recorded. Clients were randomly allocated into two groups; those who took the tape recording home and those who did not. Pre-clinic, clinic and post-clinic anxiety levels were measured using The Spielberger State-Trait Anxiety Inventory (STAI). The level of genetic knowledge was assessed pre and post clinic. Qualitative data was collected on the use of, and client's views of the tape recording. Results show that those clients who had received a tape recording were able to recall significantly more information than those who had not. There was also a significant reduction in post-clinic anxiety levels for those clients who received a tape recording to take home, compared with the no tape group. The home visit by the nurse had the effect of increasing anxiety scores over time. The tape was favoured by those who had received it.
6.015

A review of the effect of surveillance on outcome for women considered to be at increased risk of developing breast cancer.

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The study was conducted to determine whether close surveillance is of benefit to women who are at increased risk of developing breast cancer. "At risk" included those with a family history, previous histological features associated with a pre-cancerous condition, or recurrent cysts. 141 women who had attended the benign clinic between 1974 & 1994 and had subsequently developed breast cancer were studied. Four age matched control groups were used. It was clear that surveillance patients had a better stage at presentation. Screened patients were also less likely to have an inoperable tumour (7.6% of the follow up group compared with 14.7% of the controls; p2 = 0.031). Tumours were smaller in the screened group (median sizes 2.5cm v 2.55cm; Kruskal-Wallis p2 statistic = 5.375, df=1, p=0.0204). The survival was not significantly different in the two groups. (logrank p2 statistic 0.99, df=1 p=0.32) Survival in the surveillance group at five years was 79% (95% CI 72-87) and at ten years 65% (95% CI 71-79). Survival in the control group at five years was 75% (95% CI 71-79) and at ten years 60% (95% CI 55-65). This study offers some evidence suggesting that increasing follow up regime may be of benefit to "at risk" patients in detecting cancers at an earlier stage. A larger study could help confirm these findings.

6.016

Assisted reproduction in male cystic fibrosis patients with congenital bilateral absence of the vas deferens.

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With the help of modern comprehensive management in cystic fibrosis (CF), life expectancy of CF sufferers has increased considerably to the fourth decade during the last ten years. Congenital bilateral absence or hypoplasia of the vas deferens (CBAVD) is a well known complication in male CF sufferers accounting for azoospermia in almost all cases. The male CF patients have very little hope to father a child of their own, whilst the natural pregnancy in female CF sufferers has been reported in number of cases. We describe successful application of percutaneous epididymal sperm aspiration (PESA) for sperm aspiration in two male CF patients (delta F 508/1138insG and delta F508/R117H) with CBAVD. Their partners are negative for these and other common CFTR mutations. Both couples received detailed genetic counselling including information on the scope of assisted reproduction in having their own child. The ethics of assisted reproduction were debated in view of the father's limited life expectancy. Both couples elected to attempt assisted reproduction by in vitro fertilization (IVF), using the technique of intra cytoplasmic sperm injection (ICSI). It is recommended that all male CF patients should receive information on PESA, ICSI and IVF to enable them to make an informed decision to father a child.

6.017

Managing families with children at risk of autosomal dominant polycystic kidney disease (ADPKD).

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A research clinic was established two years ago to assess children in families with autosomal dominant polycystic kidney disease (ADPKD). The functions of this were expected to be a) to monitor children at risk of ADPKD to estimate the frequency of asymptomatic but treatable complications; b) to make predictive testing available to families who wished to consider this; c) to assess the psychosocial impact on the families of the disease and of testing for the disease, and to examine family strategies for coping with the condition. In practice, the clinic has developed in a very different direction. Families with children at risk of ADPKD were contacted by letter, but the response rate has been low. In some families there have been unresolved issues relating to the affected parent and the way in which they became aware of the disorder. Some families have requested testing for ADPKD, but this has different meanings in different families - relating to genetic status or the presence of renal cysts or treatable complications. Some older children wish to participate in the discussion about testing. Differences between the parents have arisen in several families, with the interest in testing sometimes coming from the unaffected parent.

6.018

Testing children to identify carriers of balanced chromosomal translocations.

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The testing of children to identify carriers of genetic disorders is controversial, but there is little evidence of the long-term effects on families of such testing. This study set out to examine the consequences of identifying children as carriers of balanced chromosomal rearrangements by interviewing the tested individuals and their parents at least 10 years after the event. Members of 10 nuclear families were interviewed, and the conversations were recorded, transcribed and analysed. The analysis revealed that learning about carrier status causes a transient psychological disruption, which can be accompanied by feelings of discriminable stigmatisation. Some children were completely indifferent to their carrier status, although recognising that it might become relevant to them in the future, while others reported feelings and experiences of stigmatisation. One mother may have been over-protective towards her carrier son. Mothers who underwent prenatal testing experienced great emotional strain as a direct result of the procedure, which - for some - was not relieved by a favourable (unaffected) test result. Many of the parents expressed dissatisfaction with the genetic counselling they had received in the past.
6.019
Triplet repeat frequencies observed in Fragile X screening in the Yorkshire region 1994 to 1996
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Initial Fragile X (FX) screening in the Yorkshire region has been based upon PCR analysis of the FRAXA (CGG)n and the FRAXE (GCC)n repeats. The PCR products were analysed on an ABI 373 machine using GeneScanner 672 software. Samples were screened for the FRAXA repeat (600+ males and 300+ females) and the FRAXE repeat (450+ males and 150+ females). The frequency of FRAXA full mutations was 1.5% of the screened population. Premutation expansions made up 0.7% of the FRAXA screened population with intermediate expansions making up a further 4.5%. The frequency of FRAXE expansions was considerably lower with only 2.0% of the screened population carrying an intermediate expansion. However one full FRAXE expansion was observed along with two mosaic males within the intermediate expansion range. The frequency of intermediate alleles appears raised in the FRAX screened population compared to previously published control groups. These results are consistent with the suggestion that relatively large unmethylated expansions may play some role in mental impairment (Murray et al). Human molecular genetics, 1996. However we suggest that all females apparently homozygous for common FRAXA alleles continue to be screened by Southern blotting, as several apparently homozygous females were found to carry large expansions.

6.020
Sudden Unexpected Unexplained Death in Tuberous Sclerosis
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There is a small but recognised mortality in persons with tuberous sclerosis (TS), but sudden unexpected death is rare. We report the clinical and pathological details of 4 males and 6 females with TS who died suddenly and unexpectedly. 1 died during a fit, the remaining 9 unobserved, 7 while asleep. The age at death was from 7 months to 54 years. 8 of the 10 had learning difficulties and 7 of these 8 had epilepsy. Post-mortem examination was performed in 5, 3 had cardiac rhabdomyoma(ta) and 1 central nervous systems tubers. Although a cardiac arrhythmia or respiratory arrest during an epileptic fit are possible causes of sudden unexpected death, in none of the cases was a definite cause of death identified. We suggest that persons caring for individuals with tuberous sclerosis be aware of the small but significant risk of sudden unexpected death. Review of seizure control, anti-epileptic medications, along with detailed post-mortem examination of the central nervous system and cardiac conducting tissues is essential in persons with TS who die suddenly and unexpectedly in order to be able to determine possible causes of death and identify avoidable risk factors.