## Programme

### Monday 28 September 1998

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
</tr>
</thead>
<tbody>
<tr>
<td>08.00-10.00</td>
<td><strong>CLINICAL GENETICS SOCIETY - Council Meeting</strong> Room V044 Vanbrugh College</td>
</tr>
<tr>
<td>09.00-17.00</td>
<td><strong>Genetic databases / Cyber Cafe</strong> Throughout the conference in the Exhibition area there will be a CyberCafe to demonstrate genetic sites on the World Wide Web.</td>
</tr>
<tr>
<td>10.00-12.30</td>
<td>Concurrent sessions</td>
</tr>
</tbody>
</table>

### FISH: present and future

**Exhibition Centre Room:** PX001  
**Chair:** Dr John Wolstenholme and Mr John Barber

- **Introduction** Dr John Wolstenholme / Mr John Barber

- **10:05 M-FISH** Dr Michael Speicher
- **10:30 RxFISH** Dr Johannes Wienberg
- **10:55 CGH for constitutional abnormalities and prospects for CGH to DNA microarray targets** Dr Saeed Ghaffari
- **11:15 Aspects of FISH and Leukaemias** Dr Christine Harrison
- **11:30 Aspects of FISH/GH and solid tumours** Dr Seamus O'Neill
- **11:45 Constitutional cytogenetic amplification** Mr John Barber
- **12:00 3-D organisation of chromosomes in the interphase nucleus** Dr Emanuela Volpi

### Practical approaches to methylation and imprinting

**Exhibition Centre Room:** PL001  
**Chair:** Dr John Harvey

- **10:00 Overview** Dr Bernhard Horsthemke

- **Invited contributions**

- **10:55** Invited contributions (SP1)
- **11:00** Invited contributions (SP2)
- **11:15** Invited contributions (SP3)
- **11:30** Invited contributions (SP4)

### Predictive testing protocols: who benefits?

**Central Hall (10:30 am start)**  
**Chair:** Mrs Alison Lashwood

- **10:30** Invited contributions (SP5)
- **10:45** Invited contributions (SP6)
- **11:00** Invited contributions (SP7)
- **11:15** Invited contributions (SP8)
- **11:30** Invited contributions (SP9)
- **11:45** Invited contributions (SP10)
- **12:00** Invited contributions (SP11)
- **12:15** Invited contributions (SP12)
- **12:30** Invited contributions (SP13)

### Additional Events

- **12.30 ASSOCIATION OF GENETIC NURSES AND COUNSELLORS** Business Meeting, PL001 Exhibition Centre
- **12.30 CLINICAL MOLECULAR GENETICS SOCIETY** Council Meeting, PT103 Exhibition Centre
- **12.30 SPECIALIST REGISTRARS IN CLINICAL GENETICS** (meeting for trainees and trainers) Room PX001 Exhibition Centre
- **12.30-14.00** LUNCH AND POSTER VIEWING (Exhibition Centre)
14.00-17.15 Symposium: The genetics of human behaviour
Room: Central Hall
Chair: Professor Pat Jacobs & Professor Ian Craig

14.00 (SP14) Behavioural phenotype in velo-cardio-facial syndrome Dr Keiran Murphy
14.30 (SP15) The Cognitive processes underlying behaviour in Williams syndrome Dr Annette Karmiloff-Smith
15.00 (SP16) Behavioural aspects of Smith Magenis syndrome Ann C M Smith
15.30-16.15 TEA AND POSTER VIEWING (Exhibition Centre)
16.15 (SP17) Progress in understanding the genetic basis of epilepsy Dr Louise Bate
16.45 (SP18) New horizons: DNA pooling - an approach to genome wide screening for behaviour genes Professor Michael Owen
17.15 BRITISH SOCIETY FOR HUMAN GENETICS - Annual General Meeting Central Hall
18.00 WINE TASTING around TRADE EXHIBITION - Exhibition Centre
19.30-20.30 DINNER

Tuesday 29 September 1998

09.00-12.45 Symposium: Human Evolution
Room: Central Hall
Chair: Professor Tom Strachan & Dr Mike Jackson

09.00 (SP19) The sons of Adam and the daughters of Eve Professor Bryan Sykes
09.35 (SP20) Karyotype evolution in mammals revealed by cross-species chromosome painting Dr Fengtang Yang
10.10 (SP21) Could variable imprinting mediate transgenerational adaption Professor Marcus Pembrey
10.45-11.30 COFFEE AND POSTER VIEWING (Exhibition Centre)
11.30 (SP22) The behaviour of trinucleotide repeats and other micro satellites Dr David Rubinsztein
12.05 (SP23) Large scale sequence comparison of mouse and human alphaglobin gene clusters Dr Jonathan Flint
12.45-14.00 LUNCH AND POSTER VIEWING

14.00 The Carter Lecture
Room: Central Hall
Chair: Professor Dian Donna

(SP24) The evolution of modern humans Professor Kenneth K Kidd, Yale University

15.00-15.45 TEA AND POSTER VIEWING (Exhibition Centre)
### Concurrent sessions

#### Exhibition Centre Room: PL001
**Chair: Dr Jill Clayton-Smith**

- **15:45 (SP25)**
  - Outcome of pregnancy in women with epilepsy with regard to malformation
  - Montgomery, Tara; Fairgrieve, S; Jonas, P; Burn, J; Lynch, SA

- **16:00 (SP26)**
  - Fetal glucokinase gene mutations result in reduced birth weight
  - Ellard, Sian; Beards FE; Ballantyne E; Appleton M; Harvey R; Hattersley AT

- **16:15 (SP27)**
  - An integrated map of segmental aneuploidy associated with congenital malformations.
  - Brewer, Carole; Holloway, S; Zawalnyski, P; Schinzel, A; FitzPatrick, D

- **16:30 (SP28)**
  - Xp deletions associated with autism in two females
  - Thomas, Simon; Sharp, A; Browne, C; Dennis, N

- **16:45 (SP29)**
  - A Locus for Primary Microcephaly maps to 8p22-pter
  - Jackson, Andrew; McHale, D; Karban, G; Mueller, R; Markham, A; Lench, N; Woods, G

#### Exhibition Centre Room: PX001
**Chair: Miss Wendy Johnston**

- **15:45 (SP31)**
  - Genetic testing considerations in patients with cancer
  - France, Elizabeth; Gray, J; Tischkowitz, M; Brain, K; Sampson, J; Anglim, C; Clarke, A; Parsons, E; Mansel, R; Barrett-Lee, P

- **16:00 (SP32)**
  - Prenatal diagnosis: describing informed decision making
  - Bekker, Hilary; Hewison Jenny; Thornton Jim

- **16:15 (SP33)**
  - BRCA1 Testing - Case Studies, the Male Perspective
  - Rae, Linda; Turnier, G; Woods, C G; Chu, CE; Bishop T

- **16:30 (SP34)**
  - Genetic counselling for Cystic Fibrosis in consanguineous Pakistani Muslim families with Congenital Bilateral Absence of the Vas Deferens: Experience in the Yorkshire Regional Genetics Service
  - Ahmed, Musthag; Chu, CE; Ellis, LA; Robson, F; Taylor, G

- **16:45 (SP35)**
  - Attitudes of deaf, hard of hearing and hearing adults towards genetic testing for inherited deafness
  - Middleton, A; Hewison, J; Mueller RF

- **17:00 (SP36)**
  - Is the uptake of pre-natal diagnosis among the Pakistani populations of Yorkshire and Northern Pakistan a reflection of changing transcultural beliefs about reproduction?
  - Karban, Gulshan; Mason, GC; Raashid, Y; Hussain, J; Chu, C; Woods, CG; Mueller, RF

#### Room: Central Hall
**Chair: Dr Rob Elles**

- **15:45 (SP37)**
  - A liver specific ABC transporter gene (SPGP) is mutated in chromosome 2q linked progressive familial intrahepatic cholestasis
  - Strautnieks, Sandra; Bull, L; Dahl, N; Sokal, E; Ling, V; Tanner, M; Freimer, N; Gardiner, RM; Thompson, RJ

- **16:00 (SP38)**
  - Molecular genetic analysis of von Hippel-Lindau (VHL) disease
  - Maher, Eamonn R; Rehal, P; McMahon, R; Rose, S; Littleboy, S; Richards, FM; Webster, AR; Woodward, ER; Whittaker, J; Macdonald, F

- **16:15 (SP39)**
  - The APC variants I1307K and E1317Q are associated with colorectal tumours but not always with a family history
  - Frayling, Ian; Beck, NE; Ilyas, M; Dove-Edwin, I; Bell, JA; Williams, CB; Hodgson, SV; Bodmer, WP; Tomlinson, IPM;

- **16:30 (SP40)**
  - Germline E-cadherin gene mutations in familial gastric cancer
  - Richards, Frances M; McKee, SA; Cole, TRP; Evans, DRG; Jankowski, JA; McKee, C; Maher, ER

- **16:45 (SP41)**
  - TSC1 and TSC2 mutations in 153 sequentially ascertained unrelated patients with tuberous sclerosis: implications for diagnostics and counselling
  - Sampson, Julian; Jones, A; Shyam Sundar, M; Thomas, M; Maynard, J; Tomkins, S

- **17:00 (SP42)**
  - Emery-Dreifuss muscular dystrophy: mutation spectrum and phenotype in X-linked families
  - Yates, John; Bagshaw, J; Aksmanovic, V; Coomber, E; McMahon, R; Whittaker, J; Morrison, P; Kendrick-Jones, J; Ellis, J

### Additional Information

- **17:30**
  - ASSOCIATION OF CLINICAL CYTOGENETICISTS - Annual General Meeting  Central Hall

- **19:30**
  - CONFERENCE SOCIAL PROGRAMME: CARIBBEAN EVENING
Wednesday 30 September 1998

09.00-10.30 Concurrent Sessions

Multiple endocrine neoplasia  
(organised in conjunction with Cancer Family Study Group)  
Room: Central Hall  
Chair: Prof John Burn

9.00  
Molecular Genetics & Clinical Genetics of MEN 1  
Prof Rajesh V Thakker

9.30  
Molecular Genetics & Clinical Genetics of MEN 2  
Prof Bruce Ponder

10.00  
Aspects of Treatment in MEN Syndromes  
Prof John Farndon

Cytogenetics

Room: PX001  
Chair: Dr Alan McDermott

9.00  
Colour banding identifies cryptic chromosome rearrangements in ten cases of myeloid malignancies  
Harrison, Christine; Yang, F; Butler, T; Cheung, K-L; Gibbons, B; Ferguson-Smith, M

9.15  
Applications of Comparative Genomic Hybridization in Constitutional Chromosome Studies  
Stallings, Raymond; Breen, CJ; Hall, K; Dunlop, A; Green, AJ

9.30  
Cytogenetic prenatal diagnosis in the UK: results from UKNEQAS external audit: 1987-1997  
Waters, Jonathan On behalf of UK National External Assessment Scheme (NEQAS) in Clinical Cytogenetics

9.45  
A molecular and FISH analysis of structurally abnormal Y chromosomes in patients with Turner syndrome  
Robinson David; Dalton P; Jacobs, PA; Mosse, K; Power, M; Skuse, D; Crolla J

10.00  
Clark, Alan; Moore, L; Martin, F.A; Gaunt, K.L

10.15  
Terminal deletions of the short arm of chromosome 1: three new cases  
Morgan, Deborah; Willatt, L; Firth, H; Kelsall, A

10.30-11.15 COFFEE AND POSTER VIEWING
11.15-12.45 Concurrent sessions

Room: Central Hall
Chair: Professor Dian Donnai

11:15 (SP52)
Clinical genetics services for common disorders
Hughes, Helen; Harper, P.S.

11:30 (SP53)
Genetic registers in the UK - where now?
Dean, John CS; Fitzpatrick, DR; Farndon, P; Kingston, H

11:45 (SP54)
The cost of providing genetic assessment and enhanced surveillance to women at increased risk of familial breast cancer: preliminary results from the TRACE Project Team.
Barton, Garry; Cohen, D; Gray, J; on behalf of the TRACE Project Team

12:00 (SP55)
A study of family history taking in general practice
Storrar, David; Howard, E; Sowerby, S

12:15 (SP56)
Telegenetics: Delivering genetic services at a distance. A pilot study across the Welsh mountains
Gray, Jonathon; Alderman, J; Hughes, H

12:30 (SP57)
Public Health Genetics: A New Discipline?
Zimmern, Ron

Exhibition Centre Room: PL001
Chair: Professor Andrew Read

11:15 (SP58)
Targeted disruption of Brca2 exon 11 gives rise to viable mice with embryonic growth retardation, developmental defects and thymic lymphomas
Thistlewaite, Fiona; Friedman, LS; Carlton, MBL; Collinge, WH; Ponder, BAJ; Evans, MJ

11:30 (SP59)
Identification of the human homologue of a mouse retinoic acid-induced gene in Xp22

12:00 (SP60)
Fine mapping of crossover breakpoints in subtelomeric DNA
Armour, John; Badge, RM

Exhibition Centre Room: PX001
Chair: Dr John Old

11:15 (SP64)
Variation in PLP gene duplications causing Pelizaeus-Merzbacher disease
Woodward, Karen; Kendall, E; Vetrie, D; Hodes, E; Malcolm, S

11:30 (SP65)
Cationic trypsinogen mutations in families with Hereditary Pancreatitis in the UK and Ireland
Rutherford, Sarah; Ellis, I; McDonald, F; Howes, N; Whitcomb, DC; Neoptolemos, JP; Mountford, RC

11:45 (SP66)
Identification of mutations in the EYA1 gene in Branchio-Oto-Renal (BOR) syndrome patients
Rickard, Sarah; Boxer, M; Trompeter, R; Bittner-Blindzicz, M

12:00 (SP67)
Sharing of PPT mutations between distinct clinical forms of neuronal ceroid lipofuscinoses in patients from Scotland
Greene, Nicholas D; Leung, KY; Munroe, PB; Stephenson, JBP; Crowe, YJ; Mole, SE; Gardiner, RM; Mitchison, HM

12:15 (SP68)
Mutation screening of vestibular schwannomas in individuals not fulfilling NIH criteria for NF2
Evans, Gareth; Purcell, S; Wu, C-L; Wallace, A; Neary, W; Lye, R; Ramsden, R

12:30 (SP69)
Novel HOX A13 mutations and the phenotypic spectrum of Hand-Foot-Genital Syndrome
Goodman, Frances; Donnenfeld, A.E.; Feingold, M; Fryns, J.P.; Hennekam, R.C.M.; Scambler, P.J.
12.45-14.00  LUNCH AND POSTER VIEWING

14.00-15.30  Concurrent sessions

**Exhibition Centre Room: PX001**
Chair: Dr Nick Dennis

14:00  (SP70)
Genetic investigation of isolated ocular and central nervous system haemangioblastoma cases
McKee, Shane; Webster, AR; McMahon, R; Rose, S; Kaur, P; Macdonald, F; Moore, AT; Maher, ER

14:15  (SP71)
Dilated Vestibular Aqueducts and their significance in deafness
Reardon, William; O Mahoney, C; Phelps, P; Coyle, B; Trembath, R

14:30  (SP72)
A unique family with Charcot-Marie-Tooth, sensorineural deafness, vocal cord palsy and anticipation maps to 17p11.2-p12
Kimonis, Virginia; Kovach, Margaret; Lin, Jing-Ping; Boydajiev, Simeon; Campbell, Kathleen; Frank, William; Gelber, David; Mazzeo, Larry; Jabs, Ethylin

14:45  (SP73)
Functional effects of a polymorphism of the collagen (I) alpha 1 gene (COL1A1) in osteoporosis
Hobson, Emma; Dean, V; Grant, SFA; Ralston, SH

15:00  (SP74)
Experience from over 250 molecular diagnostic tests for facioscapulohumeral muscular dystrophy (FSHD): the need for close clinical/laboratory liaison.
Tyfield, Linda; Stephenson, A; Jardine PE; Lunt PW

15:15  (SP75)
The molecular basis of malonyl-CoA decarboxylase deficiency
FitzPatrick, David; Hill, Alison; Tolmie, John; Thorburn, David; Christodoulou, John

**Room: Central Hall**
Chair: Dr Fiona Macdonald

14:00  (SP76)
Cloning and mutation analysis of the DKC1 gene responsible for X-linked dyskeratosis congenita
Knight, Stuart; Heiss, NS; Vulliamy, T; Stavrides, G; Greschner, S; Mason, P; Dokal, I; Poustka, A

14:15  (SP77)
Characterisation of the human myeloid leukaemia-derived cell line GF-D8 by multiplex FISH, subtelomeric probes & CGH
Tosi, Sabrina; Giudici, G; Scherer, SW; Rambaldi, A; Dirscherl, L; Biondi, A; Kearney, L

14:30  (SP78)
A novel method for increasing the productivity of mutation scanning methods
Wallace, Andrew; Wu, C-L; Elles, R

14:45  (SP79)
Identification of loci on chromosome 3p containing putative ovarian cancer tumour suppressor genes
Fullwood, Paul; Rader J; Martinez A; Broggin M; Morelli C; Latif F; Maher ER

15:00  (SP80)
Identification of putative coding sequences within the critical lung tumour suppressor region at 3p21.3: A comparison of various techniques
D.P. Macartney; F. Latif; S.E. Walsh; S. Sekido; M.H. Wei; F.M. Duh; L. Geil; D. Angeloni-Adreazoli; P. Byrd; E.R. Maher, J.D. Minna, M.I. Lerman.

15:15  (SP81)
Toutain, Annick; Burn, J

15.30  End of conference
Poster presentations

Biochemical genetics
01.01 Family study of isolated glycerol kinase deficiency Kidd, Alexa; Sargent, C; White, I; Auchtarlonie I; Kirk, J; Moore, S; Dean, J
01.11 The use of tissue specific promoters in the correction of OTC deficiency in mouse models Trainer, Allison H.; Akhurst, R.J.

Cancer genetics and cytogenetics
02.01 A Cytogenetic study on fine needle aspirates in Non Hodgkin's Lymphoma Jameel, Tahir; Anwar, Masood; Ahmad, Manzoor
02.02 Review of paediatric tumours with 22q11 rearrangement Roberts, Paul; Annings, Jacob; Cullinane Catherine
02.03 The use of DNA extraction from tumour blocks in three breast/tumoral cancer families Hampson, Kim; Cole, T; Bourm, D
02.04 Assessment of the Cytologic 2.1 breast cancer susceptibility programme in selecting suitable families for mutation screening Broomfield, Angela; Stoppo, K; Bourm, D; MacDonald, F; Maher, E R
02.05 A novel gene, regulated by erbB2, encodes a nuclear protein which is specifically expressed in breast carcinoma Lu, Pei; Sundquist, K; Barkstrom, D; Poulsom, R; Hanby, A; Freemont, P; Taylor-Papadimitrou, J
02.08 An unaffected Ashkenazi Jewish woman presenting at the breast cancer clinic for predictive testing is found to carry two BRCA mutations. Haworth, Andrea; Short, J.P; Eales, R.A; Houlston, R.S; Murray, V.A; Taylor, R
02.09 von Hippel-Lindau Syndrome (VHL) surveillance in patients with an isolated VHL lesion and a negative family history Gardiner, Carol; Bumell, L; Turnier, G; & Chu, C E.
02.12 A qualitative study of psychological issues in male breast cancer France, Elizabeth; Gray, J
02.13 A survey of surgical approaches to women with a family history of breast cancer in Wales Daoud, Roaful; Gray, J; Mansel, R; Clarke, D; Dischowitz, M; Sweetland, HM
02.14 Presymptomatic genetic testing for women with a family history of breast cancer in Wales Gray, Jonathon; Myring, J; France, E; Sampson, J; Rogers, C; Mansel, R; Harper, P; on behalf of the TRACE Project Research Group
02.15 Replication errors in early onset and familial colorectal cancer Verma, Loveena; Brassel, C; Froggat, N; MacDonald, F; Bourm, D; Sanders, S; Williams, G; Welch, S; Evans, D G; Maher, ER
02.17 Duplication or translocation? Abnormalities of 1q. Roberts Kathryn; Moorman A.V; Bennett C; Butler T; Cheung K.L; Kasprzyk A; Martineau M; Pinson MP; Swanton S and Hancock C J.
02.21 A case of follicular lymphoma with double minutes and 3p26 amplification Gaffari, Saeed R; Boyd, E; Patterson, L; Connor, JM
02.22 Detection of Hyperdiploidy in Acute Lymphoblastic Leukaemia by Comparative Genomic Hybridisation Kim, Mee Hye; Stewart, J; Devlin, C; Boyd, E; Connor, JM
02.23 MEN2A presenting as bilateral phaeochromocytoma in father and son. Chu, Carol E; Pope, R M; Robinson, M D; Taylor, G R
02.24 Identification of a cryptic 15;17 translocation in a case of APL by Fluorescence in Situ Hybridisation Felix, Clive; Sharpe, C; Beddow, R; Hillman, P
02.26 Chromosome 7 abnormalities in sisters with Swachman-Diamond Syndrome and MDS Cunningham, Joan; Sales, M; Wilkie, R; Huntby, B; Prell, NR
02.27 Analysis of the prevalence of specific ancestral germline mutations in BRCA1 & BRCA2 & APC, in women affected with breast or ovarian cancer from the Ashkenazi Jewish population: correlation with pedigree data Hodgson, Shirley; Lewis, CM; Heap, E; Eales, R; Cameron, J; Ellis, D; Mathew, C; Solomon, E
02.28 A randomised controlled trial to evaluate the emotional impact of genetic assessment for familial breast cancer Brain, Kate; Gray, J; France, E; Anglim, C; Clarke, A; Parsons, E; Rogers, C; Mansel, R; Harper, P; On behalf of the TRACE Project
02.30 Preventative mastectomy in women with an increased lifetime risk of breast cancer Laloo, Fiona; Hopwood, P; Baldam, A; Brain, A; Howell, A; Evans, DGR
02.31 Accuracy of recording of family history of young breast cancer patients Laloo, Fiona; Zelton, A; O'Dair, L; Barr, L; Evans, DGR
02.32 A potential problem with detection of inv(16) by RT-PCR Hackwell, Stuart M; Robinson, D O; Harvey, J F; Ross, F M
02.33 Cytogenetic assessment of bone marrow proliferation in severe aplastic anaemia (SAA) Walther, Joachim-Ul; Fuehrer, M; Rampf, U; Bender-Goetze, Ch
02.35 Comparative Genomic Hybridization (CGH) Analysis of Neuroblastomas. Breen, Claire J; O'Meara, A; Stallings, R L.
02.37 A review of 25 cytogenetically abnormal Ewing tumour families comparing survival against cytogenetic findings and RT-PCR evaluation of EWS/FLI-1 transcript type. Morris, Stephen; Roberts, P; Wealdon, J; Anninga, J
02.38 Rapid mutation screening of the BRCA1 gene in early onset sporadic and familial breast cancer patients using chemical cleavage of mismatch analysis Ellis, David; Greenman, J; Izatt, L; MacDermot, K; Perrett, C; Evans, G; Hodgson, S; Mathew, C
02.39 Direct mutation screening in MEN2 and related syndromes: a report of the first four years Mcmahon, Robert; Littleboy, S M; Whittaker, J L
02.41 Investigation of candidate genes for low penetrance breast cancer susceptibility. Healey, Catherine; Dunning, AM; McBride, S; Pharoah, PDP; Gregory, J; Foster, N; Teare, D; Easton, DF; Ponder, B AJ
02.42 Identification of EX1 and EX2 gene mutations in families with Hereditary Multiple Exostoses Richards, Frances M; Rajapak, MH; Gerrand, C; Farnon, PA; Cole, TRP
02.44 Rearrangement of the MLL gene in AML patients without 11q23 abnormalities Abdou, S; Jadayel, D; Min, T; Dainton, J; Swansbury, J; Powles, R; Catovsky, D
02.45 A novel 10;11 translocation involving the MLL gene in AML M2. Emmerson, Julie; Davies, C J; Nokes, T J C; Patterson, K; Davies, H
02.46 Is the NF1 gene involved in genetic predisposition to neuroblastoma? McConville, Carmel; Mann, J P; Stillier, C A
02.48 Incidence of the t(13q7) APC gene mutation in UK Ashkenazi Jewish patients with a family history of breast cancer Rajapak, Helen; McKay, S; Evans, DGR; Burk, R; MacDonald, F; Maher, ER
Prenatal genetics/diagnosis and cytogenetics

04.01 An investigation of interphase FISH methods to provide a rapid prenatal diagnosis of chromosomal abnormalities in uncultured amniocytes. Simpson, Peter; Milne, A; Waters, J; Davison, EV

04.02 A rare complex chorionic villus structural chromosome mosaicism involving isochromosome 18q. Ocraf, Kevin; Smith, N; Calvert, A; Ramsey, M; Gill, K; Munro, L; Heath, M; Wilkinson, A; Laister, C; Hamilton, R

04.03 Prenatal diagnosis of a duplication of chromosome 11p13-14 in the absence of major phenotypic abnormalities. Cooper, Clare; Ocraf, K; Crolla, J; Padfield, C J; Monk, A; Wilkinson, A; Whitehouse, M; Gill, K; Heath, M; Filer, A; Bruce, J

04.04 A molecular genetic protocol to identify maternal contamination in prenatal samples. Gardiner, Carol; Rowland, JS; & Woods, CG

04.05 Prenatal detection of trisomy 21 by QF-PCR: Preliminary results, a novel alternative to DNA extraction and a cautionary tale on marker selection. Bateman, Mark; Willatt, L; Whittaker, J

04.06 Early onset fetal hydrops and muscle degeneration in siblings due to a novel variant of type IV glycogenosis. Brunton LA; Cox PM; Bjelogric, P; Sewry, C; Ladda, E; Murphy, K

04.07 Pre- and post-natal karyotypic discordance for a de novo unbalanced structural chromosome abnormality - an unusual case resulting from apparent instability of the 21q telomeric region. Duckett, David; Healy, K; Steer, J; Creswell, L; Barrow, M

04.08 Development of an antenatal screening programme for congenital abnormalities in a South Wales District. Al-Jader, N; Layle, Crompton G

04.09 Prenatal detection of an unstable de novo dicentric chromosome, dic(14;15)(p11.1;p13), with duplication of the Prader-Willi/Angelman syndromes critical regions (PWASCR). Curtis, Merryl; Thompson, PW; Davies, SJ; Roberts, SH

04.10 Prenatal diagnosis and characterisation of a de novo supernumerary marker chromosome originating by presumptive 3:1 segregation and resulting in pure trisomy 10p. Morgan, Sian; Curtis, MA; Vujanic, GM; Procter, AM; Roberts, SH

04.11 Development of a rapid karyotyping method for use in prenatal diagnosis of high risk pregnancies. Boyle, Tracey; Ellis, K; Herbert, A; Crolla, J

04.12 Prenatal Diagnosis of Mosaic for Partial Trisomy 8: A Case Report. Jay, Anne; Roberts, E; Davison, EV

04.13 Rapid prenatal diagnosis in 3 cases of fluid non-amniotic in origin. Few, Graham; Roberts, E; Larkins SA; Kilby, M; Davison, EV

04.14 Mosaic trisomy 8 syndrome: A discordant amnioncentesis result in a twins pregnancy. Jeffries, Sally; Jay, Anne; Roberts, E; Davison, EV

04.15 Predictive value of common cytogenetic abnormalities detected following CVS: results from the Association of Clinical Cytogenetists UK Collaborative Study. Smith, Kim; Maher, E; Lowther, G; Hourihan, T; Wilkinson, T; Wolstenholme, J

04.16 Isolated fetal echogenic bowel as an indicator for molecular screening for cystic fibrosis. Norbury C; Gali; Boyd Patricia; Paola Gass Pirrone, Chamberlain Paul

04.17 Prenatal genetic diagnosis of coagulation factor VII deficiency in a consanguineous family. Lambert, Christopher T; Thompson, L J; Lalloz, M R A; Karban, G; Mueller, R F; Layton, D M

04.18 Lack of evidence for translocations between the FHS and associated loci at 4q35 and the telomere of 10q26. Rogers, Mark; Upadhyaaya M; Harper, PS

04.19 Prenatal diagnosis of chromosome abnormalities in the West Midlands 1988-1996 and the application of FISH technology. Roberts, Eileen; Elly A; Leedham P; Hutton M; Davison, EV

04.20 A retrospective study of follow-up data on cases of prenatally diagnosed mosaicism. Clark, Alan; Gaunt, K L

04.21 Prenatal diagnosis for Facioscapulohumeral muscular dystrophy(FSHD) UPADHYAYA, MEENA; MACDONALD, M; RAVINE, D

Programme S9

General cytogenetics

03.01 Investigation into the mechanisms of the origin of a rare case of mosaicism involving chromosome 18. Jackson, Ann; Donnai, D; Turner, H; Gaunt, KL

03.02 Chromosome 16 in a normal adult. Turnbull, Kate; Chandler, K; Turner, H; Gaunt, KL

03.03 Mosaicism for structural chromosome abnormality. Williams, J; Coles, P; Mattby, E

03.04 Two unbalanced chromosome 22 abnormalities with a possible linked origin in a mildly affected female. Steer, James; Duckett, D; Price, S; Willatt, L

03.05 The smallest cytogenetically detected terminal deletion of 8p reported to date. Browne, Caroline; Temple, K

03.06 A new variant of chromosome 16? Friend, Nicola; Barber, J

03.07 A rare case of true Trisomy 4 mosaicism detected by CVS. Davies, Teresa; Delmege, David; Beck, Jennifer; Kyle, Pippa; Porter, Helen

03.08 Possible false positive results in Fetal Blood Samples in a Twin Pregnancy. Davies, Teresa; Bagley, E; Kyle, P; Torge, H M; Newbury-Ecob, R; Berry, P; Delmege, D; Lowe, C

03.09 An infant with D2 abdominal distension, Dysmorphism and a Cytogenetically visible deletion of Chromosome 10 at q11.2. Douglas, A; Royston, B; Sweeney, E; Howard, PJ

03.10 Somatic mosaicism in Fanconi’s anaemia. Nobbs, Michael; Howell, RT

03.11 CGH defines 11q interstitial deletions and correlates deletions with phenotype. Power, Monica; Breen, CJ; Hegarty, AM; Ryan, F; Ryan, CA; Ahmad, S; Macken, SR; Green, AJ; Stallings, RL

03.12 Unexpected outcome of a ‘3-way translocation’ Sales, Mark; Siblebald, J; Pratt, N

03.13 A retrospective study of cytogenetic findings in patients referred for Fetal X screening. Bovey, Mary; Colgan, J M; Younger, R M; Lowther, G W

03.14 XX male and trisomy 21: an unexpected finding. Morgan, Deborah; Willatt, L; Owen, A

03.15 A DMD family with both an Xp inversion and duplication. Turner, Helen; McGaughran, J; Kingston, H; Snadden, L; Tolmie, J; Gaunt, L

03.16 Autistic twin sisters who both have a maternally inherited supernumerary derivative chromosome 15. Glancy, Mary; Power, MM; Hall, K; Webb, D; Broderick, AM; Green, AJ; Stallings, RL

03.17 Inherited interstitial deletion of proximal chromosome 4 short arm (p13p14) in a mother and son with normal intelligence. Ellis, Richard; Garrett, Christine; Bennett, Shona

03.18 Monosomy twins: Discordant for Pallister-Killian syndrome. Glancy, Mary; Hegarty, AM; Hall, K; McGuaid, S; Gorman, W; Green, AJ; Stallings, RL

03.19 A de novo balanced (X;22)(p11.2; q11.2) translocation in an adult female with a schwannoma. Smith, Elaine; Willatt, LR; Trump, D; Firth, H; Clegg, S; Leversha, M

03.20 Interstitial deletion of chromosome 5q(13q15) - a recognisable syndrome? Willatt, Lionel; Dyson, HM; Green, AJ; Tuck, SJ

03.21 The impact of computerised image analysis on FISH results. Daryani, YP; Patton, MA
Uniparental disomy studies in two cases of trisomy 16 mosaicism detected at amniocentesis Dicke, Jane S W; Clark, C; Kidd, A M J; Massie, D; Gray, E S; Dean, J C S

Detection of aneuploidy on uncultured amniocytes using FISH Hastings, Rosalind; Svennevik, C; Smith, M; Watson, S; Delhanty, J

Uniparental disomy: a clinical dilemma Ferriman, Emma; Rutter, A; Gardiner, CA; Woods, CG; Mason, GC

False positive results in the diagnosis of trisomy 18 on chorionic villus biopsy Ferriman, Emma; Linton, G; Woods, CG; Mason, GC

Transcervical retrieval of fetal cells in the first trimester of pregnancy and the results of isolation of fetal cesa by density gradient centrifugation and MACS Daryani, YP; Barker, G; Patton, MA

Clinical genetics

A new lethal short-rb skeletal dysplasia with microamelia and platyspondyly Slaney, Sarah F;(1,2); Sprigg A.A.; Davies N.P.; Hall C.M.

Terminp 2p deletion: Report of 2 cases Suri, Mohinsh; Morton, JEV; Davis, G; Larkins, S; Rodgers, CS; Holder, S

Incontinentia pigmenti presenting with recurrent subungual squamous cell carcinoma Shannon Nora; Giblin C A; Quarrall O W J; Page R E

A Genetic Family Register as a Model of Proactive Genetic Counselling Nicola Bradshaw; Holloway S; Kerzijn-Storrar L; Macleod R

Amplifications of the 15q11-13 region of maternal origin associated with a specific facial phenotype and autism. Gardiner, Carol; Robinson, M; Beddow, R A; J; Woods, CG

Unusual phenotype genotype correlation in a case of cystic fibrosis Law, Caroline; Lane, S; Robison, D

A Genetic Investigation of Children Attending Special Schools in Edinburgh Pearson Pauline; Halpin, S; Jackson, P; FitzPatrick, D

Study of individuals with balanced translocations of reproductive age Young, Dorothy; Fordye, A; Wright, A; Faulkner, C; Porteous, M

A Family with Hereditary Port-Wine Stain Berg, Jonathan (1,2); Quaba, A; Geogontopoulou A.; and Porteous M

Mutations of the cationic trypsinogen in British families with hereditary pancreatitis. Creighton, Jane; Lyall,R.; Wilson,D.I.Charnley, R.M.;CURTIS,A.

Partial trisomy 15 and 16 associated with 15p;16q translocation Magee, Alex; McManus D; Nevin NC

Monosoperoitcinoidontic dwarifism - clinical study of a new case Rusu, Cristina; Voloscuici, Milhia

INTERFAMILY VARIABILITY OF PHENOTYPE IN AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE TYPE 1 Haterboer, Nick; Lazaru, LP, Williams, AJ; Holmans, P; Ravine, D

Location of mutations within the PKD2 gene influences clinical outcome. Haterboer, Nick; Peters, D; San-Millan, JL; Bogdanova, N; Coto, E; Sagar-Malki, A; Torra, R; Krawczak, M; Ravine, D;

Clinical study of pulmonary lymphangioleiomyomatosis in patients with tuberous sclerosis Hancock, E; Tomkins, Susan; Sampson, J; Osborne, J

Okhiro syndrome and Acro-renal-ocular syndrome: two families with an overlapping phenotype Wadde, Kristin; Mohammed, S N; Calver, D M

A Cardiff pilot study for the investigation of analysis specific collagen mutations Tysoe, Carolyn J; Hills, N J; White, L J; Ravine, D; Pope, F M MRC Connective Tissue Group

17 Hydroxylase and then 21 Hydroxylase deficiencies in sibs: the dilemma of multiple autosomal recessive disorders in consanguineous pedigrees Gardiner, Carol; Woods, CG; Mueller, RF; Butler, G; Chatfield, S; Chu, C; Kabani, G; Ahmed, M

Autosomal Dominant Nephropathy with Rhabdomyolysis in Noonan Syndrome Len, Ruxandra; Sharland, M; Patton, M A

An unusual form of metaphyseal dysplasia with extraskeletal features DERRY, Christopher; COLLINS, A L.

Predictive testing for breast and ovarian cancer predisposition: What is the psychosocial impact? Foster, Claire; Watson, M; Baille, C; Eiles, R; Evans, G; Ashley, S; Davidson, J; Steering Group of the Cancer Research Campaign UK National Trial of the Psychosocial Impact of BRCA1/

Search of genetic factors underlying hereditary essential tremor Zagorovskaya, Tatiana; Druznia, E; Karabanov, A; Iliaionashkin, S; Ochvinikov, I; Ivanova-Smolenskaya, I.

Germline Mutations in patients with Multiple Endocrine Neoplasia Type I (MEN1) Lam, Wayne W K; Deeble, J; Chartton, R; Taylor, G; Belchetz, P; Fitzpatrick, D; Chu, CE

Autosomal dominant nephritis, macrothrombocytopathy and sensorineural deafness (Epstein syndrome) : 2 new families McGaughran, Julie, Perveren, R; Black, G

Utilization of predictive, prenatal and diagnostic testing for Huntington's disease in Johannesburg Kronberg, Jennifer; Krause, A

Germ-line mosaicism in Alcohol Related Oesdymopathy Aldred, Michellea; Bagshaw, RJ; Macdermot, K; Casson, D; Trehamb, RC

Brachyactyly Type B - further evidence for marked variability in clinical presentation Slavolneke, Anne; Donnai, D; Clayton-Smith, J

Carrier detection in Duchenne muscular dystrophy Butler, Allen; Barton, DE

Allele-Specific Reduction of 59 and DMAHP mRNA in DM Fibroblast Cell Lines Alawazzan, Madawi; Hamshere, M; Newman, E; Brook, JD

Pro-Thrombin mutation(2020A)associated with fatal venous thromboembolism in two families Shannon, M S; Thomson, P; Bevan, DH; Mereorea, J; Murday, VAM

West Midlands strategy for the management of familial cancer: One year follow-up data Sleighthome, Vicky; Maher, E R; Lawrence, G; Corder, A P; Harper, P H; Cole, T

Counselling/education/screening

Maternal serum screening for Down's syndrome: a survey of midwives' views Magny Dorothy, White I; Fairgrieve S

A Web site for Public Health Genetics Stewart, Alison; Zimmeren, Ron

Ethical and psycho-social issues encountered in a linkage study of a family with CADASIL (Cerebral autosomal dominant arteriopathy with sub-cortical infarcts and leukoencephalopathy) Lyall, Roz; Blank, C; Burn, D; Couthard, A; Keers, S; Bushby, K

A psychological model for parent child communication in familial breast cancer risk France, Elizabeth; Gray, J; Brain, K; Street, E

The current and future role of genetics services in primary care: the opinions of general practitioners in Wales Kirk, Meggie; tredale, R; Longley, M

Neonates with transient hypertrypsinemia and heterozygous for delta F508 are at risk of a second mild mutation Dalton, Ann; Evans, S; Boyne, J; Pollitt, RJ; Taylor, CJ

A ready-reckoner for rapid genetic analysis of complex traits. Pritchard, Dorian
Programme S11

06.27 The status of genetics education on diploma-level training courses for nurses in the United Kingdom. Kirk, Maggie

06.28 Evaluation of factors influencing interest in having a test for genetic susceptibility to breast and/or ovarian cancer and the psychological impact of such testing. Jacobo, Christine; Hodgson, S; Bish, A; Sutton, S; Ramirez, A

06.31 Intermediate zone alleles in Fragile X syndrome. Lindley, Victoria, MacDonald, F; Bullock, S; Larkins, S; Davison, E V

Epidemiology/population genetics

07.01 CFTR gene diseases. Schwarz, Martin; Malone GM; Andrew N; Super M

07.02 Prevalence of Mendelian genetic disorders. Al-Jader, N Layla; Clark A; Davies S; Ravine, D; MacMillan, J; Wilkie, A; Harper, P

07.11 Age and tissue specific variation of X-chromosome inactivation in normal women. Sharp, Andrew

07.13 Evaluation of the effects of systematic misclassification errors on genotype association analysis studies. Teare, M; Davenport, A.M.; Healey, K.; Easton, D.F.

07.14 Progression of mental and motor symptoms in Huntington's disease: correlations with CAG repeat length. Harper, Peter S; Stevens, M; Krawczak, M; Jones, A L

Gene structure and regulation

08.01 Functional analysis of the Huntington's disease gene promoter. Rubinstein, David C; Coles, R (1,2); Caswell, R

08.02 The analysis of a ribosome approach to target retained DMPK transcripts in Myotonic Dystrophy. Hamshere, M; Broek, JD

08.03 Functional analysis of DNA variants identified in the promoter/5'-UTR of the neurofibromatosis type 1 (NF1) gene. Osborn, Michael; Upadhyaya, M

08.04 Different types of mutation in KVLQTL1 underlie Romano-Ward and Jervell and Lange-Nielsen syndromes. Tyson, Jess; Tranebjerg, L; Bellman, S; Malcolm, S; Batten, J; Aslaksen, B; Sorland, S; Lund, O; Bilton-Gindlitz, M

08.05 Functional analysis of mutations in the L1 gene that cause neurological disease in man. De Angelis, Elena; Du, J-S; Watkins, A; MacFarlane, J; Yeo, G; Hicks, R; Kenrick, S

08.06 Analysis of mutations in Holt-Oram syndrome. Cross, Steve; Ching, Y H; Li, Q Y; Armstrong-Buissere, L; Packham, E; Ghosh, T; Yi, C H; Newbury-Ecob, R; Broek, J D

08.07 Analysis of Pilomatrixomas in Myotonic Dystrophy. Newman, Emma; Hamshere, M G; Alwazzan, M A; Broek, J D

Differential and development

09.02 Identification and mapping of Four New Human Members of the T-box Gene Family: HEOMES, TBX6, TBX18, and TBX19. Yi, Cheong-Ho; Terrett, J A.; Li, G Y.; Ellington, K.; Packham, E A.; Armstrong-Buissere, L.; McClure, P.; Slingsby, T.; Broek, J D.

Linkage mapping/marker polymorphisms

10.01 The Linkage Hotel at the Human Genome Mapping Project Resource Centre. Dearlove, Andrew; Thompson, A; Rhodes, M

10.02 Homozygosity mapping locates a gene for autosomal recessive spondylocostal dysostosis to chromosome 19q13.1-q13.3. Tumpenny, Peter; Bulman, MP; Frayling, TM; Abu-Nasra, K; Hattersley, AT; Ellard, S

10.11 Identification of locus specific single nucleotide polymorphisms (SNPs). Riley, John; Purvis, I J; Bhatti, S; Gibson, R; Goddard, N J; Hosking, L; John, I; Lumden, S; See, C G

10.12 Linkage studies to SCa6 in FHM families. Gordon, T; Cooke, A; Reid, E; Connor, JM

10.13 A genome - wide linkage study in a large Malignant Hyperthermia family. McCall, Sam; Hardy, Hall, W; J; Cuman, J L; Halsall, P; Hopkins, P M; Lench, N; Markham, A F; Ellis, F R

10.15 Mapping of a gene for spastic cerebral palsy which has a variable impact on the overall phenotype. McHale, Duncan; Mitchell, S; Bundey, S; Monirhan, L; Campbell, D A; Woods, C G; Lench, N J; Mueller, R F; Markham, A F

Physical mapping

11.11 The exclusion of three candidate genes from the pathogenesis of Noonan Syndrome by fluorescence in situ hybridisation. Ion, Ruxandra

The Noonan Syndrome Collaborative Research Group

Molecular cytogenetics

12.01 Molecular delineation of the 3p- syndrome. Green, Elaine K; Maliszewska, C; Waters, J; Laffl, F; Maher, ER

12.12 Identification of the Origin of Marker Chromosomes by CGH and In Situ Hybridisation in Patients with AML and MDS. Kim, Mee Hye; Stewart, J; Devlin, C; Boyd, E; Connor, JM

12.16 Translocation breakpoints mapped approximately 70-250 Kb from the TWIST gene in three patients with Saethre-Chotzen Syndrome. Patel, Poonvi; Reardon, W; Malcolm, S; Winters, R M

12.17 Identification and recovery of assaillant DNA in sexual assault cases. Bayoumy, Nagy; Pounder, D; Sales, M; Pratt, NR

12.20 Detection of Chromosome 16 Aberrations in AML Using a Micro-FISH Probe. Stefanou, Eunice-Geo; Boyd, E; Stewart, J; Connor, J M

12.21 Identification of the Origin of Double Minutes present in the Cells of an MDS Patient using Micro-FISH. Stefanou, Eunice-Geo; Boyd, E; Connor, J M

12.22 Identification of PLP gene duplications using Interphase Fluorescence In-Situ Hybridisation as an aid to diagnosis in patients with Pelizaeus-Merzbacher Disease. Palmer, Rodger; Woodward, K; Malcolm, S; Rao, K

12.23 1p deletion syndrome - report of a case and review. Slavotinek, Anne; Gaunt, L; Donnai, D; Clayton-Smith, J

12.24 Marker Chromosome 8 identified by FISH. Bhagwagar, Anj; Cockett, W

12.25 Chromosome unbalances are associated with poor prognosis in Chronic Lymphocytic Leukaemia. Hiorns, Lynne

British Human Genetics Conference 1998

J Med Genet 1998; 35; Supplement 1
**Mutation analysis**

14.01 Identification of a novel mt480delG mutation in Multiple Endocrine Neoplasia Type 1. Evans Steven; Curtis D; Dalton A; Cook J; Ross R; Quarrrell OWJ

14.02 Determination of SMNT and SMNc copy number in SMA families. Butler, Rachel; Mountford, R

14.04 New PCR-Based Test for Fragile X Mutation Detection. Cornley, Iain

14.06 Application of the protein truncation test (PTT) to mutation analysis in the neurofibromatosis type 1 (NFI) gene. Osborn, Michael; Upadhyaya, M

14.11 932delA - a novel LDLR mutation causing FH in N. Ireland shows evidence of common ancestral origin. Beattie, Diane; Martin, S; Ward, A.J.; Graham, C.A.; Young, I.S.; Nichols, D.P.

14.12 932delA - a novel mutation in the F8 gene. Hughes, Anne; Armstrong, DKB; McKenna, KE

14.13 Vohwinkel's keratoderma: a novel loricrin mutation. Hughes, Anne; Armstrong, DKB; McKenna, KE

14.14 Application of the protein truncation test (PTT) to mutation analysis in the neurofibromatosis type 1 (NFI) gene. Osborn, Michael; Upadhyaya, M

14.15 Characterisation of a second hit in a pheinxiform neurofibroma from a neurofibromatosis type 1 (NFI) patient. John, Alison; Ruggieri, M; Upadhyaya, M

14.20 Variation of site-specific methylation patterns in the factor VIII (F8) gene in human sperm DNA. Miller, David; Krawczak, M; Cooper, DN

14.21 Nearest-neighbour Effects upon Germline Nucleotide Substitution Rates in Human Genes. Krawczak, M; Ball, EV; Cooper, DN

14.23 Mutational screening in the flanking regions of CMT2A. Paton, Catherine; Knapps, S; Ireland, M; Pilla, G; O'Brien, C; Burn, J; Curtis, A; Lindsay, S

14.24 Genomic CTG/CAG repeat size is significantly enlarged and inversely correlated with age-at-onset in Type II diabetics. Morris, Alex; Tyson, Phillip; Hepburn, David; Mortimer, Anne

14.25 Cystic fibrosis analysis of a patient with a mutation in the exon 23 of the CFTR gene. Barton, David; Joyce, C; Barret, T; Harrison, R

14.26 Molecular analysis of exon 15 of the APC gene in 17 Northern Irish families with Familial Adenomatous Polyposis: 9 germline mutations identified by restriction digest, PTT, and sequencing. Malin, Edward; Nevin, N.C; Graham, C.A.; Bradley, A

14.27 Rapid Throughput Mutation Analysis of the Hereditary Non-polyposis Colorectal Cancer Genes hMLH1 and hMSH2. Barton, Geraint; Harvey, J. F.

14.28 Rapid Mutation Analysis of Charcot-Marie-Tooth Type 1. Silibourne, Julie; Hackwell, S.M.; Stevens, C.A.; Harvey, J. F.

14.30 Mutational analysis of the mucopolysaccharidoses disorders. Meaney, Cathy; Genet, S; Young, E; Winchester, B

14.31 Novel mutations in the 3' region of the Poly cystic Kidney Disease 1 (PKD1) gene. Sandford, Richard; Thomas, R; Whittacker, J; Bradley, J

14.33 Mutation screening strategy for craniofacial disorders. Monaghan, Gemma; Reardon, W; Malcolm, S; Boxer, M

14.34 Type VII Collagen (COL7A1) Mutation Screening in Severe Recessive Dystrophic Epidermolysis Bullosa Patients using the Protein Truncation Test and Fluorescent Chemical Cleavage of Mismatch. Whitlock, Neil; Meienio, J; McGrath, J; Eady, R; Mathew, C; Abb, S