Annotation
National haemophilia B molecular genetic register  R Harris  601

Review article
A new strategy for the genetic counselling of diseases of marked mutational heterogeneity: haemophilia B as a model  F Giannelli, S Saad, A J Montandon, D R Bentley, P M Green  602

Original articles
X inactivation as a mechanism of selection against lethal alleles: further investigation of incontinentia pigmenti and X linked lymphoproliferative disease  A Harris, J Collins, D Vetrue, C Cole, M Bobrow  608
Chromosomal localisation of a pseudoautosomal growth gene(s)  T Ogata, C Petit, G Rappold, N Matsuo, T Matsumoto, P Goodfellow  624
Non-specific X linked mental retardation with aphasia exhibiting genetic linkage to chromosomal region Xp11  G N Wilson, C S Richards, K Katz, G S Brookshire  629
The acrocallosal syndrome and Greig syndrome are not allelic disorders  L A Brueton, K A Chotai, L van Herwerden, A Schinzel, R M Winter  635
Multipoint mapping of adult onset polycystic kidney disease (PKD1) on chromosome 16 P M Pignatelli, S E Pound, A D Carothers, A M Macnicol, P L Allan, M L Watson, A F Wright  638
Mutation analysis of 184 cystic fibrosis families in Wales  J Cheadle, J M. Myring, L Al-Jader, L Meredith  642
Identification of a new DMD gene deletion by ectopic transcript analysis  F Rininsland, A Hahn, S Niemann-Seyde, R Slomski, F Hanefeld, J Reiss  647
Cartilage-hair hypoplasia in Finland: epidemiological and genetic aspects of 107 patients O Mäkitie  652
Screening for phenylketonuria in a totalitarian state  L Kalaydjieva, I Kremensky  656

Syndrome of the month
Weyers’ ulnar ray/oligodactyly syndrome and the association of midline malformations with ulnar ray defects  P D Turnpenny, J C S Dean, P Duffy, J A Reid, P Carter  659

Short communications
X inactivation patterns in females with Alport’s syndrome: a means of selecting against a deleterious gene  D Vetrue, F Flinter, M Bobrow, A Harris  663
Autosomal dominant retinitis pigmentosa (ADRP): a rhodopsin mutation in a Scottish family C Bell, C A Converse, M F Collins, L Esakowitz, K F Kelly, N E Hailes  667

Case reports
New dysmorphic features in Rubinstein-Taybi syndrome  D Kanjilal, M A Basir, R S Verma, B K Rajegowda, R Lala, A Nagaraj  669
18p- syndrome and hypopituitarism  H G Artman, C A Morris, A D Stock  671

Dysmorphology reports
Bilateral retinopathy, aplastic anaemia, and central nervous system abnormailities: a new syndrome?  T Revesz, S Fletcher, L I Al-Gazali, P DeBuse  673
Unknown syndrome: nasal hypoplasia, sparse hair, truncal obesity, genital hypoplasia, and severe mental retardation  J-P Fryns, J Delooz, H Van Den Berghe  676

Abstracts
Medical genetics: advances in brief  678

Letters to the Editor
Beckwith–Wiedemann syndrome  A M Norman, A P Read, D Donnai  679
A mutation in exon 7 of the CFTR gene is common in the western part of France M P Audrezet, B Mercier, H Guillermot, C Ferenc  679
Williams syndrome and chromosome 18  F H Menko, P J H M Stouthart  679

Book reviews
BMJ PUBLISHING GROUP  TAVISTOCK SQUARE  LONDON WC1H 9JR