

Contents

Original articles

- German family study on hereditary breast-ovarian cancer *U Hamann, H Becher, T Zimmermann, K Pella, G Bastert, J Chang-Claude* 633
- Complex genetic predisposition to cancer in an extended HNPCC family with an ancestral hMLH1 mutation *P Hutter, A Couturier, R J Scott, P Alday, C Delozier-Blanchet, F Cachat, S E Antonarakis, F Joris, M Gaudin, L D'Amato, J-M Buerstedde* 636
- Distribution of oculocutaneous albinism in Zimbabwe *P M Lund* 641
- Phenotypic variability in male patients carrying the mutant ornithine transcarbamylase (OTC) allele, Arg40His, ranging from a child with an unfavourable prognosis to an asymptomatic older adult *I Matsuda, T Matsuura, A Nishiyori, S Komaki, R Hoshida, T Matsumoto, M Funakoshi, K Kiwaki, F Endo, A Hata, M Shimadzu, M Yoshino* 645
- The deletion of six amino acids at the C-terminus of the $\alpha 1(\text{II})$ chain causes overmodification of type II and type XI collagen: further evidence for the association between small deletions in COL2A1 and Kniest dysplasia *A Winterpacht, A Superti-Furga, U Schwarze, H Stöss, B Steinmann, J Spranger, B Zabel* 649
- PAX genes and human neural tube defects: an amino acid substitution in PAX1 in a patient with spina bifida *F A Hol, M P A Geurds, S Chatkupt, Y Y Shugart, R Balling, C T R M Schrandt-Stumpel, W G Johnson, B C J Hamel, E C M Mariman* 655
- No founder effect in three novel Alzheimer's disease families with APP 717 Val \rightarrow Ile mutation *D Campion, A Brice, D Hannequin, F Charbonnier, B Dubois, C Martin, A Michon, C Penet, M Bellis, A Calenda, M Martinez, Y Agid, F Clerget-Darpoux, T Frebourg, and the French Alzheimer's Disease Study Group* 661
- Characterisation of four novel fibrillin-1 (FBN1) mutations in Marfan syndrome *L C Adès, E A Haan, A F Colley, R I Richards* 665
- Linkage of rheumatoid arthritis to the candidate gene NRAMP1 on 2q35 *M-A Shaw, D Clayton, S E Atkinson, H Williams, N Miller, D Sibthorpe, J M Blackwell* 672
- PCR assay confirms diagnosis in syndrome with variably expressed phenotype: mutation detection in Stickler syndrome *N N Ahmad, D M McDonald-McGinn, P Dixon, E H Zackai, W S Tasman* 678
- Uneven X inactivation in a female monozygotic twin pair with Fabry disease and discordant expression of a novel mutation in the α -galactosidase A gene *I Redonnet-Vernhet, J K Ploos van Amstel, R P M Jansen, R A Wevers, R Salvayre, T Levade* 682
- Investigation of an interleukin-4 promoter polymorphism for associations with asthma and atopy *A J Walley, W O C M Cookson* 689

Syndrome of the month

- Rett syndrome *A Clarke* 693

Short reports

- X chromosome inactivation pattern in female carriers of X linked hypophosphataemic rickets *K H Ørstavik, R E Ørstavik, J Halse, J Knudtzon* 700
- Gorlin syndrome associated with midline nasal dermoid cyst *E K Pivnick, A W Walter, M D Lawrence, M E Smith* 704
- Hidrotic ectodermal dysplasia of hair, teeth, and nails: case reports and review *L S Chitty, N Dennis, M Baraitser* 707
- Heterogeneity of phenotype in two cystic fibrosis patients homozygous for the CFTR exon 11 mutation G551D *R B Parad* 711
- Linkage of a medium sized Scottish autosomal dominant retinitis pigmentosa family to chromosome 7q *Z Mohamed, C Bell, H M Hammer, C A Converse, L Esakowitz, N E Haites* 714
- Genotype-phenotype correlation in von Hippel-Lindau disease: identification of a mutation associated with VHL type 2A *F Chen, L Slife, T Kishida, J Mulvihill, S E Tisherman, B Zbar* 716

Abstracts

- Medical genetics: advances in brief 718

Letters to the Editor

- Prevalence of 22q11 microdeletion *S Tézenas du Montcel, H Mendizabel, S Aymé, A Levy, N Philip* 719
- Should the 3C (craniocerebellocardiac) syndrome be included in the spectrum of velocardiofacial syndrome and DiGeorge sequence *M G Butler, P Mowrey* 719

Book reviews

- 720

Notices

- 720

BMJ PUBLISHING GROUP TAVISTOCK SQUARE LONDON WC1H 9JR

