

## Original articles

- A locus for primary ciliary dyskinesia maps to chromosome 19q M Meeks, A Walne, S Spiden, H Simpson, H Mussaffi-Georgy, H D Hamam, E L Fehaid, M Cheehab, M Al-Dabbagh, S Polak-Charcon, H Blau, A O'Rawe, H M Mitchison, R M Gardiner, E Chung 241
- Localisation of the gene causing diaphyseal dysplasia Camurati-Engelmann to chromosome 19q13 K Janssens, R Gershoni-Baruch, E Van Hul, R Brik, N Guañabens, N Migone, L A Verbruggen, S H Ralston, M Bonduelle, L Van Maldergem, F Vanhoenacker, W Van Hul 245
- Mutation screening in Rett syndrome patients F Xiang, S Buervenich, P Nicolao, M E S Bailey, Z Zhang, M Anret 250
- Genetic heterogeneity of Usher syndrome type II: localisation to chromosome 5q S Pieke-Dahl, C G Möller, P M Kelley, L M Astuto, C W R J Cremers, M B Gorin, W J Kimberling 256
- Report of five novel and one recurrent *COL2A1* mutations with analysis of genotype-phenotype correlation in patients with a lethal type II collagen disorder G R Mortier, M Weis, L Nuytinck, L M King, D J Wilkin, A De Paepe, R S Lachman, D L Rimoin, D R Eyre, D H Cohn 263
- Universal primer quantitative fluorescent multiplex (UPQFM) PCR: a method to detect major and minor rearrangements of the low density lipoprotein receptor gene K E Heath, I N M Day, S E Humphries 272
- Parental origin and mechanisms of formation of cytogenetically recognisable de novo direct and inverted duplications D Kotzot, M-J Martinez, G Bagci, S Basaran, A Baumer, F Binkert, L Brecevic, C Castellan, K Chrzanowska, F Dutly, A Gutkowska, S B Karaüzung, M Krajewska-Walasek, G Luleci, P Miny, M Riegel, S Schuffenhauer, H Seidel, A Schinzel 281
- An unbalanced submicroscopic translocation t(8;16)(q24.3;p13.3)pat associated with tuberous sclerosis complex, adult polycystic kidney disease, and hypomelanosis of Ito B H J Eussen, G Bartalini, L Bakker, P Balestri, C Di Lucca, J O Van Hemel, H Dauwerse, A M W van den Ouwendijk, C Ris-Stalpers, S Verhoef, D J J Halley, A Fois 287
- Clinical and radiological assessment of a family with mild brachydactyly type A1: the usefulness of metacarpophalangeal profiles C M Armour, D E Bulman, A G W Hunter 292

## Letters to the Editor

- Can hair be used to screen for breast cancer? A Howell, J G Grossmann, K C Cheung, L Kanbi, D G R Evans, S S Hasnain 297
- Mutation analysis of *SMAD2*, *SMAD3*, and *SMAD4* genes in hereditary non-polyposis colorectal cancer S Roth, M Johansson, A Loukola, P Peltomäki, H Järvinen, J-P Mecklin, L A Aaltonen 298
- Novel mutation in the *MYOC* gene in primary open angle glaucoma patients J P C Vasconcellos, M B Melo, V P Costa, D M L Tsukumo, D S Bassères, S Bordin, S T O Saad, F F Costa 301
- Three novel *SALL1* mutations extend the mutational spectrum in Townes-Brocks syndrome C Blanck, J Kohlhase, S Engels, P Burfeind, W Engel, A Bottani, M S Patel, H Y Kroes, J M Cobben 303
- Genotype-phenotype correlation in three homozygotes and nine compound heterozygotes for the cystic fibrosis mutation 2183AA→G shows a severe phenotype M O Kilinç, V N Ninis, A Tolun, X Estivill, T Casals, A Savov, E Dagli, F Karakoç, M Demirkol, G Hüner, F Özkinay, E Demir, J L Seculi, J Pena, C Bousono, J Ferrer-Calvet, C Calvo, G Glover, I Kremenski 307
- Alkaptonuria in Italy: polymorphic haplotype background, mutational profile, and description of four novel mutations in the homogentisate 1,2-dioxygenase gene B Porfirio, I Chiarelli, C Graziano, A Mannoni, A Morrone, E Zammarchi, D Beltrán-Valero de Bernabé, S R de Córdoba 309
- Rough skin, brittle hair, and photosensitivity: a mild phenotypic variant of trichothiodystrophy R Savarirayan, R J M Gardner, R D Sinclair, M McDowell, J E Cleaver 312
- The first description of lethal pterygium syndrome with facial clefting (Bartsocas-Papas syndrome) in 1600 P D Turnpenny, R Hole 314
- Sex reversal and diaphragmatic hernia in phenotypically female sibs with normal XY chromosomes S Manouvrier-Hanu, R Besson, L Cousin, C Jeanpierre, N Kacet, M Cartigny, L Devisme, L Storme, B de Martinville, P Lequien 315
- Clinical variability of Stickler syndrome with a *COL2A1* haploinsufficiency mutation: implications for genetic counselling J Faber, A Winterpacht, B Zabel, W Gnoinski, A Schinzel, B Steinmann, A Superti-Furga 318

