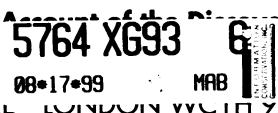




Journal of Medical Genetics
March 1998 Vol 35 No 3

Contents

Review article		
Recurrence risks in mental retardation	Y J Crow, J L Tolmie	177
Original articles		
Counselling issues in familial hypertrophic cardiomyopathy	B Yu, J A French, R W Jeremy, P French, D R McTaggart, M R Nicholson, C Semsarian, D R Richmond, R J Trent	183
How the magnitude of clinical severity and recurrence risk affects reproductive decisions in adult males with different forms of progressive muscular dystrophy	S Eggers, M Zatz	189
Genetic heterogeneity and HOMOGENEITY analysis in British malignant hyperthermia families	R Robinson, J L Curran, W J Hall, P J Halsall, P M Hopkins, A F Markham, A D Stewart, S P West, F R Ellis	196
Further refinement of Pendred syndrome locus by homozygosity analysis to a 0.8 cM interval flanked by D7S496 and D7S2425	M Mustapha, S T Azar, Y B Moglabey, M Saouda, G Zeitoun, J Loiselet, R Slim	202
Molecular pathology of familial hypertrophic cardiomyopathy caused by mutations in the cardiac myosin binding protein C gene	B Yu, J A French, L Carrier, R W Jeremy, D R McTaggart, M R Nicholson, B Hambly, C Semsarian, D R Richmond, K Schwartz, R J Trent	205
PCR based mutation screening of the laminin $\alpha 2$ chain gene (LAMA2): application to prenatal diagnosis and search for founder effects in congenital muscular dystrophy	P Guicheney, N Vignier, X Zhang, Y He, C Cruaud, V Frey, A Helbling-Leclerc, P Richard, B Estournet, L Merlini, H Topaloglu, M Mora, J-P Harpey, C-A Haenggeli, A Barois, B Hainque, K Schwartz, F M S Tomé, M Fardeau, K Tryggvason	211
Evidence of linkage of the inflammatory bowel disease susceptibility locus on chromosome 16 (IBD1) to ulcerative colitis	M M Mirza, J Lee, D Teare, J-P Hugot, P Laurent-Puig, J-F Colombel, S V Hodgson, G Thomas, D F Easton, J E Lennard-Jones, C G Mathew	218
Outcome of chromosomally normal livebirths with increased fetal nuchal translucency at 10-14 weeks' gestation	A F Brady, P P Pandya, B Yuksel, A Greenough, M A Patton, K H Nicolaidis	222
A new strategy for cryptic telomeric translocation screening in patients with idiopathic mental retardation	S R Ghaffari, E Boyd, J L Tolmie, Y J Crow, A H Trainer, J M Connor	225
A new approach to the elucidation of complex chromosome rearrangements illustrated by a case of Rieger syndrome	C Mackie Ogilvie, F L Raymond, R H Harrison, P N Scriven, Z Docherty	234
Syndrome of the month		
Costello syndrome	N Philip, S Sigaudy	238
Short reports		
Extensive germinal mosaicism in a family with X linked myotubular myopathy simulates genetic heterogeneity	M-C Vincent, C Guiraud-Chaumeil, J Laporte, S Manouvrier-Hanu, J-L Mandel	241
Molecular basis of variegate porphyria: a missense mutation in the protoporphyrinogen oxidase gene	J Frank, H Lam, E Zaider, M Poh-Fitzpatrick, A M Christiano	244
Septo-optic dysplasia and WS1 in the proband of a WS1 family segregating for a novel mutation in PAX3 exon 7	M L Carey, T B Friedman, J H Asher Jr, J W Innis	248
The first de novo mutation of the connexin 32 gene associated with X linked Charcot-Marie-Tooth disease	F Meggouh, A Benomar, H Rouger, S Tardieu, N Birouk, J Tassin, C Barhoumi, M Yahyaoui, T Chkili, A Brice, E LeGuern	251
Identification of a new missense mutation in MyBP-C associated with hypertrophic cardiomyopathy	J C Moolman-Smook, B Mayosi, P Brink, V A Corfield	253
Pancreatic exocrine dysfunction associated with mitochondrial tRNA^{Leu(UUR)} mutation	H Onishi, T Hanihara, N Sugiyama, C Kawanishi, E Iseki, Y Maruyama, Y Yamada, K Kosaka, S Yagishita, H Sekihara, S Satoh	255
Fuch's corneal dystrophy in a patient with mitochondrial DNA mutations	R L Albin	258
Abstracts		
Medical genetics: advances in brief		260
Letters to the Editor		
Is meconium ileus genetically determined or associated with a more severe evolution of cystic fibrosis?	M De Braekeleer, C Allard, J-P Leblanc, G Aubin, F Simard	262
UK centres are not following the Royal College of Pathologists' recommendations for storage of Guthrie cards: a national policy is needed	S Rahman, A Emery, J Poulton	263
Cyclopia and sirenomelia in a liveborn infant	M I Martínez-Frías, A García, E Bermejo	263
Book review		
Correcting the Blueprint of Life. An Historical Account of the Discovery of DNA Repair Mechanisms		264
Notice		264



J Med Genet: first published as on 1 March 1998. Downloaded from <http://jmg.bmj.com/> on February 8, 2023 by guest. Protected by copyright.