

## Contents



This logo indicates an article that was published online first ahead of print

### Original articles

- 385** Mutation of *DNAJC19*, a human homologue of yeast inner mitochondrial membrane co-chaperones, causes DCMA syndrome, a novel autosomal recessive Barth syndrome-like condition K M Davey, J S Parboosingh, D R McLeod, A Chan, R Casey, P Ferreira, F F Snyder, P J Bridge, F P Bernier
- 394** Epsilon sarcoglycan mutations and phenotype in French patients with myoclonic syndromes S Tezenas du Montcel, F Clot, M Vidailhet, E Roze, P Damier, C P Jedynek, A Camuzat, A Lagueny, L Vercueil, D Doummar, L Guyant-Maréchal, J-L Houeto, G Ponsot, S Thobois, M-A Cournelle, A Durr, F Durif, B Echenne, D Hannequin, C Tranchant, A Brice, the French Dystonia Network
- 401** Genotype-phenotype correlation in Costello syndrome: *HRAS* mutation analysis in 43 cases B Kerr, M-A Delrue, S Sigaudy, R Perveen, M Marche, I Burgelin, M Stef, B Tang, O B Eden, J O'Sullivan, A De Sandre-Giovannoli, W Reardon, C Brewer, C Bennett, O Quarell, E M'Cann, D Donnai, F Stewart, R Hennekam, H Caveé, A Verloes, N Philip, D Lacombe, N Levy, B Arveiler, G Black
- 406** The phenotypic spectrum in patients with arginine to cysteine mutations in the *COL2A1* gene K P Hoornaert, C Dewinter, I Vereecke, F A Beemer, W Courtens, A Fryer, H Fryssira, M Lees, A Müllner-Eidenböck, D L Rimoin, L Siderius, A Superti-Furga, K Temple, P J Willems, A Zankl, C Zweier, A De Paepe, P Coucke, G R Mortier
- 414** The distribution of apolipoprotein E alleles in Scottish perinatal deaths J-C Becher, J W Keeling, N McIntosh, B Wyatt, J Bell
- 419** Mutations of the *RET* gene in isolated and syndromic Hirschsprung's disease in human disclose major and modifier alleles at a single locus L de Pontual, A Pelet, D Trochet, F Jaubert, Y Espinosa-Parrilla, A Munnich, J-F Brunet, C Goriadis, J Feingold, S Lyonnet, J Amiel
- 424** Cancer risks in first degree relatives of *BRCA1* mutation carriers: effects of mutation and proband disease status J Gronwald, T Huzarski, B Byrski, K Medrek, J Menkiszak, A N Monteiro, P Sun, J Lubinski, S A Narod

### Short reports

- 429** An inversion inv(4)(p12-p15.3) in autistic siblings implicates the 4p GABA receptor gene cluster J B Vincent, SI Horike, S Choufani, A D Paterson, W Roberts, P Szatmari, R Weksberg, B Fernandez, S W Scherer
- 435** Autosomal dominant optic atrophy associated with hearing impairment and impaired glucose regulation caused by a missense mutation in the *WFS1* gene H Eiberg, L Hansen, B Kjer, T Hansen, O Pedersen, M Bille, T Rosenberg, L Tranebjærg
- 441** Mutation in the epsilon subunit of the cytosolic chaperonin-containing t-complex peptide-1 (*Cct5*) gene causes autosomal recessive mutilating sensory neuropathy with spastic paraplegia A Bouhouche, A Benomar, N Bouslam, T Chkili, M Yahyaoui
- 444** A second locus for Aicardi-Goutières syndrome at chromosome 13q14-21 M Ali, L J Highet, D Lacombe, C Goizet, M D King, U Tacke, M S van der Knaap, L Lagae, C Rittey, H G Brunner, H van Bokhoven, B Hamel, Y A Oade, A Sanchis, I Desguerre, D Cau, N Mathieu, M L Moutard, P Lebon, D Kumar, A P Jackson, Y J Crow

### Letters to JMG

- 451** Gross rearrangements of the *MECP2* gene are found in both classical and atypical Rett syndrome patients H L Archer, S D Whatley, J C Evans, D Ravine, P Huppke, A Kerr, D Bunyan, B Kerr, E Sweeney, S J Davies, W Reardon, J Horn, K D MacDermot, R A Smith, A Magee, A Donaldson, Y Crow, G Hermon, Z Miedzybrodzka, D N Cooper, L Lazarou, R Butler, J Sampson, D T Pilz, F Laccone, A J Clarke
- 457** Decreased cellular uptake and metabolism in Allan-Herndon-Dudley syndrome (AHDS) due to a novel mutation in the *MCT8* thyroid hormone transporter C M C Maranduba, E C H Friesema, F Kok, M H A Kester, J Jansen, A L Sertié, M R Passos-Bueno, T J Visser

### JMG Unlocked


Articles carrying the



logo are available free via the journal's website: [www.jmedgenet.com](http://www.jmedgenet.com). Funding for this open access experiment is provided by the UK's Joint Information Systems Committee (JISC); see <http://jmg.bmjournals.com/cgi/content/full/42/2/97> for further information.


contd...

## Contents ...contd

- 461**  Cerebellar hypoplasia and quadrupedal locomotion in humans as a recessive trait mapping to chromosome 17p *S Türkmen, O Demirhan, K Hoffmann, A Diers, C Zimmer, K Sperling, S Mundlos*

---

### Electronic letters

- e18**  *LKB1* exonic and whole gene deletions are a common cause of Peutz-Jeghers syndrome *E Volikos, J Robinson, K Aittomäki, J-P Mecklin, H Järvinen, A M Westerman, F W M de Rooij, T Vogel, G Moeslein, V Launonen, I P M Tomlinson, A R J Silver, L A Aaltonen*
- e19** Inversion polymorphisms and non-contiguous terminal deletions: the cause and the (unpredicted) effect of our genome architecture *R Ciccone, T Mattina, R Giorda, M C Bonaglia, M Rocchi, T Pramparo, O Zuffardi*
- e20** Novel locus for X linked recessive high myopia maps to Xq23-q25 but outside MYP1 *Q Zhang, X Guo, X Xiao, X Jia, S Li, J F Hejtmancik*
- e21** Novel splice isoforms for NLGN3 and NLGN4 with possible implications in autism *Z Talebizadeh, D Y Lam, M F Theodoro, D C Bittel, G H Lushington, M G Butler*

- e22** Mutational spectrum of *COH1* and clinical heterogeneity in Cohen syndrome *W Seifert, M Holder-Espinasse, S Spranger, M Hoeltzenbein, E Rossier, H Dollfus, D Lacombe, A Verloes, K H Chrzanowska, G H B Maegawa, D Chitayat, D Kotzot, D Huhle, P Meinecke, B Albrecht, I Mathijssen, B Leheup, K Raile, H C Hennies, D Horn*

---

### Online mutation reports

- e23** Oculopharyngeal muscular dystrophy: a point mutation which mimics the effect of the *PABPN1* gene triplet repeat expansion mutation *D O Robinson, A J Wills, S R Hammans, S P Read, J Sillibourne*
- e24** Mutations responsible for Larsen syndrome cluster in the *FLNB* protein *D Zhang, J A Herring, S S Swaney, T B McClendon, X Gao, R H Browne, K E Rathjen, C E Johnston, S Harris, NMCain, C A Wise*

---

### Book review

- 393** Oxford desk reference: clinical genetics *Ian Young*

---

### Miscellaneous

- 418** Correction