

Contents




Review

- 545** Cargos and genes: insights into vesicular transport from inherited human disease
P Gissen, E R Maher

Original articles

- 556** Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children
F Zahir, H V Firth, A Baross, A D Delaney, P Eydoux, W T Gibson, S Langlois, H Martin, L Willatt, M A Marra, J M Friedman
- 562** The *C20orf133* gene is disrupted in a patient with Kabuki syndrome
N M C Maas, T Van de Putte, C Melotte, A Francis, C T R M Schrandt-Stumpel, D Sanlaville, D Genevieve, S Lyonnet, B Dimitrov, K Devriendt, J-P Frys, J R Vermeesch
- 570** Contribution of the N-acetyltransferase 2 polymorphism *NAT2*6A* to age-related hearing impairment
E Van Eyken, G Van Camp, E Franssen, V Topsakal, J J Hendrickx, K Demeester, P Van de Heyning, E Mäki-Torkko, S Hannula, M Sorri, M Jensen, A Parving, M Bille, M Baur, M Pfister, A Bonaconsa, M Mazzoli, E Orzan, A Espeso, D Stephens, K Verbruggen, J Huyghe, I Dhooze, P Huygen, H Kremer, C W R J Cremers, S Kunst, M Manninen, I Pyykkö, A Lacava, M Steffens, T F Wienker, L Van Laer
- 579** Cowden syndrome and Bannayan–Riley–Ruvalcaba syndrome represent one condition with variable expression and age-related penetrance: results of a clinical study of *PTEN* mutation carriers
K L Lachlan, A M Lucassen, D Bunyan, I K Temple

Articles carrying the  logo are freely available online under the BMJ Journals unlocked scheme, see <http://jmg.bmj.com/info/unlocked.dtl>

Short report

- 586** Genetic screening for pheochromocytoma: should *SDHC* gene analysis be included?
M Mannelli, T Ercolino, V Giachè, L Simi, C Cirami, G Parenti

Letters to JMG

- 588** Mutations of the *Birt–Hogg–Dubé* gene in patients with multiple lung cysts and recurrent pneumothorax
Y Gunji, T Akiyoshi, T Sato, M Kurihara, S Tominaga, K Takahashi, K Seyama
- 594** The spectrum of vascular anomalies in patients with *PTEN* mutations: implications for diagnosis and management
W-H Tan, H N Baris, P E Burrows, C D Robson, A I Alomari, J B Mulliken, S J Fishman, M B Irons
- 603** Genetic association analysis of inositol polyphosphate phosphatase-like 1 (*INPPL1*, *SHIP2*) variants with essential hypertension
A C Braga Marçano, B Burke, J Gungadoo, C Wallace, P J Kaisaki, P Y Woon, M Farrall, D Clayton, M Brown, A Dominiczak, J M Connell, J Webster, M Lathrop, M Caulfield, N Samani, D Gauguier, P B Munroe
- 606** Insulin-degrading enzyme is genetically associated with Alzheimer's disease in the Finnish population
S Vepsäläinen, M Parkinson, S Helisalmi, A Mannerman, H Soininen, R E Tanzi, L Bertram, M Hiltunen

Online mutation report

- e88** New *PPARG* mutation leads to lipodystrophy and loss of protein function that is partially restored by a synthetic ligand
A Lüdtke, J Buettner, H H-J Schmidt, H J Worman