# Journal of Medical Genetics



## **Contents**

#### Review



Double outlet right ventricle: aetiologies and associations D Obler, A L Juraszek, L B Smoot, M R Natowicz

#### **Commentary**

**498** The fragile X prevalence paradox *P J Hagerman* 

#### **Original articles**

- 500 Noonan and cardio-facio-cutaneous syndromes: two clinically and genetically overlapping disorders A-M Nyström, S Ekvall, E Berglund, M Björkqvist, G Braathen, K Duchen, H Enell, E Holmberg, U Holmlund, M Olsson-Engman, G Annerén, M-L Bondeson
- 507 A novel A121T mutation in human cationic trypsinogen associated with hereditary pancreatitis: functional data indicating a loss-of-function mutation influencing the R122 trypsin cleavage site P Felderbauer, J Schnekenburger, R Lebert, K Bulut, M Parry, T Meister, V Schick, F Schmitz, W Domschke, W E Schmidt
- 513 A systematic review of the clinical validity and clinical utility of DNA testing for hereditary haemochromatosis type 1 in at-risk populations *J Bryant, K Cooper, J Picot, A Clegg, P Roderick, W Rosenberg, C Patch*
- 519 X-linked ichthyosis (steroid sulfatase deficiency) is associated with increased risk of attention deficit hyperactivity disorder, autism and social communication deficits L Kent, J Emerton, V Bhadravathi, E Weisblatt, G Pasco, L R Willatt, R McMahon, J R W Yates

### **August 2008 Vol 45 No 8**

Termination of damaged protein repair defines the occurrence of symptoms in carriers of the m.3243>G tRNA<sup>Leu</sup> mutation R G E van Eijsden, L M T Eijssen, P J Lindsey, C M M van den Burg, L E A de Wit, M E Rubio-Gozalbo, C E M de Die, T Ayoubi, W Sluiter, I F M de Coo, H J M Smeets

#### **Letters to JMG**

- 535 Molecular basis of the Li–Fraumeni syndrome: an update from the French LFS families

  G Bougeard, R Sesboüé, S Baert-Desurmont,

  S Vasseur, C Martin, J Tinat, L Brugières,

  A Chompret, B Bressac-de Paillerets,

  D Stoppa-Lyonnet, C Bonaïti-Pellié,

  T Frébourg, the French LFS working group
- reduction in spermine synthase protein function and a severe form of Snyder–Robinson X-linked recessive mental retardation syndrome

  G de Alencastro, D E McCloskey, S E Kliemann,
  C M C Maranduba, A E Pegg, X Wang,
  D R Bertola, C E Schwartz, M R Passos-Bueno,
  A L Sertié

#### **PostScript**

**544** Correspondence



This article has been chosen by the Editor to be of special interest or importance and is freely available online.



Articles carrying the Unlocked Logo are freely available online under the BMJ Journals unlocked scheme.

See http://jmg.bmj.com/info/unlocked.dtl



This journal is a member of and subscribes to the principles of the Committee on Publication Fitnes

www.publicationethics.org.uk

