

Journal of Medical Genetics
September 1994 Vol 31 No 9

Contents

Review article

- The arylsulphatase A gene and molecular genetics of metachromatic leucodystrophy
M L Barth, A Fensom, A Harris 663

Original articles

- Frequency of arylsulphatase A pseudodeficiency associated mutations in a healthy population
M L Barth, C Ward, A Harris, A Saad, A Fensom 667
- A single origin for the most frequent mutation causing late infantile metachromatic leucodystrophy
J Zlotogora, Y Furman-Shaharabani, A Harris, M L Barth, K von Figura, V Gieselmann 672
- A gene for pachyonychia congenita is closely linked to the keratin gene cluster on 17q12-q21
C S Munro, S Carter, S Bryce, M Hall, J L Rees, L Kunkeler, A Stephenson, T Strachan 675
- FISH detection of trisomy 21 in interphase by the simultaneous use of two differentially labelled cosmid contigs
A F Davies, L Barber, M Murer-Orlando, M Bobrow, M Adinolfi 679
- Anticipation in Swedish families with bipolar affective disorder
P-O Nylander, C Engström, J Chotai, J Wahlström, R Adolfsson 686
- Study of the Huntington's disease (HD) gene CAG repeats in schizophrenic patients shows overlap of the normal and HD affected ranges but absence of correlation with schizophrenia
D C Rubinsztein, J Leggo, S Goodburn, T J Crow, R Lofthouse, L E DeLisi, D E Barton, M A Ferguson-Smith 690
- Asymmetry and skin pigmentary anomalies in chromosome mosaicism
C G Woods, A Bankier, J Curry, L J Sheffield, S F Slaney, K Smith, L Voullaire, D Wellesley 694
- Genetic epidemiology of single gene defects in Chile
R Cruz-Coke, R S Moreno 702
- Mulvihill-Smith syndrome: case report and review
O Bartsch, K-D Tympner, E Schwinger, R J Gorlin 707
- Familial half cryptic translocation t(9;17)
A Köhler, J Hain, U Müller 712
- Homozygosity for a new mutation (Ile¹¹⁹→Met) in the insulin receptor gene in five sibs with familial insulin resistance
J Hone, D Accili, L I Al-Gazali, G Lestringant, T Orban, S I Taylor 715
- Trisomy X in a female member of a family with X linked combined immunodeficiency: implications for carrier diagnosis
T Lester, M de Alwis, P A Clark, A M Jones, F Katz, R J Levinsky, C Kinnon 717
- Autosomal dominant simple microphthalmos
E M Vingolo, K Steindl, R Forte, L Zompatori, A Iannaccone, A Sciarra, G Del Porto, M R Pannarale 721

Syndrome of the month

- Cleft hand/foot: clinical and developmental aspects
P W Buss 726

Brief papers

- A cluster of cystic fibrosis mutations in exon 17b of the CFTR gene: a site for rare mutations
B Mercier, W Lissens, G Novelli, L Kalaydjieva, M de Arce, N Kapranov, N Canki Klain, X Estivill, A Palacio, S Cashman, A Savov, M P Audrézet, B Dallapiccola, I Liebaers, I Quéré, O Raguénès, C Verlingue, C Férec 731
- Diaphragmatic herniae and translocations involving 8q22 in two patients
I K Temple, J C K Barber, R S James, D Burge 735
- An interstitial deletion of chromosome 7(q35)
K Fagan, C Kennedy, L Roddick, A Colley 738

Letters to the Editor

- The Pallister-Hall syndrome
A Sama, J D T Mason, K P Gibbin, I D Young, M Hewitt 740
- Familial Pallister-Hall syndrome
L G Biesecker, K Topf, J M Graham Jr 740
- "CATCH 22" sans cardiac anomaly, thymic hypoplasia, cleft palate, and hypocalcaemia: cAtch 22. A common result of 22q11 deficiency?
A Lipson, B Emanuel, P Colley, K Fagan, D A Driscoll 741
- Skeletal malformations and polycystic kidney disease
A E Turco, B Peissel, S Rossetti, P F Pignatti, E M Padovani, G P Chiaffoni 742

- Notices** 742