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

Original articles
Frequency of arylsulphatase A pseudodeficiency associated mutations in a healthy population
M L Barth, C Ward, A Harris, A Saad, A Fensom

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

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 Strochman

FISH detection of trisomy 21 in interphase by the simultaneous use of two differentially labelled cosmids contigs
A F Davies, L Barber, M Murer-Orlando, M Bobrow, M Adinolfi

Anticipation in X of overlap of chromosome 21 in patients with familial Mulvihill-Smith syndrome:
JR Rubinsztein, J Leggo, S Goodburn, T J Crow, R Lofthouse, L E DeLisi, D E Barton, M A Ferguson-Smith

Asymmetry and skin pigmented anomalies in chromosome mosaicism

Genetic epidemiology of single gene defects in Chile
R Cruz-Coke, R S Moreno

McLelland-Smith syndrome: case report and review
O Bartsch, K-D Tympner, E Schwinger, R J Gorlin

Familial half cryptic translocation t(9;17)
A Köhler, J Hain, U Müller

Homozygosity for a new mutation (Ile119→Met) in the insulin receptor gene in five siblings
J Hone, D Accili, L I Al-Gazali, G Lestringant, T Orban, S J Taylor

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 Levinisky, C Kinnon

Autosomal dominant simple microphthalmos
E M Vingolo, K Steindl, R Forte, L Zompatori, A Iannaccone, A Sciarra, G Del Porto, M R Pannarale

Syndrome of the month
Cleft hand/foot: clinical and developmental aspects
P W Buss

Brief papers
A cluster of cystic fibrosis mutations in exon 17b of the CFTR gene: a site for rare mutations
B Mercier, W Lissen, O Novelli, L Kalaydjieva, M de Arce, N Kapranov, N Canki, Kain, X Estivill, A Palacio, S Cashman, A Savov, M P Audrezet, B Dallapiccola,
J Liebaers, I Queré, O Raguënes, C Verlingue, C Féraud

Diaphragmatic herniae and translocations involving 8q22 in two patients
J Temple, J C Kirk, B R James, D Burge

An interstitial deletion of chromosome 7(q35)
K Fagan, C Kennedy, L Roddick, A Colley

Letters to the Editor
The Pallister-Hall syndrome
A Sama, J D T Mason, K P Gibbin, I D Young, M Hewitt

Familial Pallister-Hall syndrome
L G Biesecker, K Topf, J M Graham Jr

"CATCH 22" sans cardiac anomaly, thymic hypoplasia, cleft palate, and hypocalcaemia:
cAch22. A common result of 22q11 deficiency?
A Lipson, B Emanuel, P Colley, K Fagan, D A Driscoll

Skeletal malformations and polycystic kidney disease
A E Turco, B Peissel, S Rossetti, P P Pignatti, E M Padovani, G P Chiaffoni

Notices