



Progressive cerebellocerebral atrophy is the name given to an apparently new syndrome by workers in Israel (*Journal of Medical Genetics* 2003;**40**:e96). They describe seven children from six families, all Sephardi Jews from Iraq or Morocco. The seven patients (five girls) had profound mental retardation, progressive spastic quadriplegia with joint contractures, and progressive microcephaly. Five developed generalised seizures in the second year of life but all seven had severe and widespread clonus. Irritability was a feature in six. Repeat MRI scans showed progressive cerebellar atrophy followed by cerebral atrophy involving both white and grey matter. No metabolic defect has been found. It is suggested that this is a new autosomal recessive syndrome affecting Sephardi Jews.

The transient gross ocular deviations, usually convergence, seen in many normal newborn babies are often regarded with some amusement by parents and others but do they have any significance? A group of 214 orthoptists have recorded observations on their own babies (*British Journal of Ophthalmology* 2003;**87**:1142–45). They found that babies who squinted occasionally (<15% of waking time) were less likely to develop later strabismus or refractive error than those who never squinted or had frequent squinting (>15% of waking time). It is suggested that neonatal squinting usually reflects an emerging and normally developing vergence system.

Some people have to spend so much on their health needs that they and their families are impoverished by it. Data from 59 countries reported from the World Health Organization (*Lancet* 2003;**362**:111–7) give rates of catastrophic household health expenditure (health spending equal to more than 40% of household income left after meeting basic needs) of up to 10.5%. Rates were greatest in some transitional countries (Azerbaijan, Ukraine, Vietnam, Cambodia) and some countries in Latin America (Argentina, Brazil, Colombia, Paraguay, Peru). The rates for the UK, USA, France, and Germany were 0.04%, 0.55%, 0.01%, and 0.03%. Three key conditions lead to catastrophic household health expenditure: payment for health services, low ability to pay, and lack of repayment or health insurance.

Accident and emergency departments have been accused of still failing to assess asthma severity properly. Records were

analysed for 255 attendances of 229 children aged 3–14 years with asthma at four London teaching hospitals (*Emergency Medicine Journal* 2003;**20**:329–31). Although heart rate, respiratory rate, and oxygen saturation were usually recorded (on 95%, 86%, and 97% of occasions respectively) peak expiratory flow rate (in children aged 5 years or older) and fraction of inspired oxygen were recorded at fewer than half of attendances. The authors of this paper call for 'interventions' to improve matters.

Etanercept and infliximab are both soluble tumour necrosis factor antibodies used in the treatment of polyarticular juvenile chronic arthritis. A 15 year old girl in Germany (*Annals of the Rheumatic Diseases* 2003;**62**:904–5) developed sterile, non-obstructive cholecystitis 14 weeks after beginning treatment with etanercept. Her abdominal symptoms resolved quickly on stopping the etanercept and she was then treated with infliximab. Nineteen weeks later her abdominal symptoms recurred and she had a cholecystectomy. There have been previous reports of two similar cases with infliximab.

Data about water fluoridation and stillbirths or congenital abnormalities have been sparse. A study in northeast England (*Journal of Epidemiology & Community Health* 2002;**57**:499–500), however, has shown no significant increase in stillbirths or certain congenital abnormalities in fluoridated areas. Data were collected over a period of 10 years (1989–98) from areas with no fluoridation (water fluoride <0.3 parts per million) and areas with full fluoridation (>0.9 parts per million). The two sets of areas did not differ significantly in rates of stillbirth and congenital abnormality (trisomies 21, 13, or 18, neural tube defect, Down's syndrome, or cleft lip and/or palate).

The risk of being admitted to hospital for a psychiatric problem is not increased among the survivors of childhood cancer, with the exception of those who have survived brain tumours (benign or malignant). In Denmark (*New England Journal of Medicine* 2003;**349**:650–7) 3710 survivors were followed up for an average of 15 years. The standardised hospitalisation ratio (SHR) for psychiatric disease was 1.3 but the excess risk was confined to survivors of brain tumours (SHR 1.8). Among these, the increased risks were for schizophrenia and related disorders (SHR 2.4), psychiatric disorders in somatic

disease (SHR 5.1), and psychoses of somatic, cerebral causes (SHR 7.7). There was no increase in admissions for depression or other affective disorders.

There is considerable variation in the severity of disease among patients with cystic fibrosis. Over 1000 disease-causing mutations of the cystic fibrosis transmembrane conductance regulator gene (*CFTR*) have been described and it is evident that genotype affects phenotype. A retrospective national cohort study in the USA (*Lancet* 2003;**361**:1671–6) including 17853 patients has related disease severity to genotype and *CFTR* function. Mortality rates were compared for patients with the 11 most common  $\Delta F508$  heterozygote genotypes and  $\Delta F508$  homozygotes. Four of the heterozygous genotypes were associated with a much lower standardised mortality than the homozygotes. Clinical phenotype (disease severity) was compared for 22  $\Delta F508$  heterozygous genotypes and the homozygous  $\Delta F508$  genotype. The same four heterozygous genotypes and one other were associated with less severe disease than the  $\Delta F508$  homozygous genotype. Disease severity was also related to *CFTR* functional classification, two of the five classes being associated with milder disease.

Methyl mercury in the human diet is almost entirely in fish and other seafood. Studies in the Faeroe Islands and in New Zealand have suggested that high maternal intake of methyl mercury during pregnancy might have an adverse effect on child neurodevelopment. These mothers, however, probably had very high methyl mercury intakes from pilot-whale blubber and shark meat. In the Seychelles mothers eat a lot of fish (12 fish meals a week) but the mercury content of fish is similar to that in most other parts of the world. Maternal hair methyl mercury content during pregnancy had little effect on children's neurocognitive performance at the age of 9 years (*Lancet* 2003;**361**:1686–92). Of 21 tests only two were related to prenatal maternal hair methyl mercury content. Increased methyl mercury exposure was associated with reduced performance on a grooved pegboard test using the non-dominant hand (almost certainly a chance finding) and with less hyperactivity. There is probably little neurodevelopmental risk from a maternal diet that contains relatively large amounts of sea fish unless the fish has an unusually high mercury content.