Some names given to proteins and genes are extremely tedious but Lucina was taken by the idea of a sonic hedgehog. The sonic hedgehog gene (Shh) (could it be the gene rather than the hedgehog that’s sonic?) is the human homologue of a fruit fly gene that encodes inductive signals controlling development of the early embryo. It is on chromosome 7 (7q 36) alongside the gene (Hpe3) implicated in some families with holoprosencephaly. In Chicago (New England Journal of Medicine 2003; 348: 1449–54) a couple’s first two children had holoprosencephaly. Analysis of four African-American siblings from the more severely affected child showed an SHH nonsense mutation. This allowed preimplantation genetic diagnosis and the birth of a healthy child.

The upper limit set by the US Centres for Disease Control and Prevention for acceptable blood lead concentration has been lowered progressively since 1970 from 60 μg/dl (2.9 μmol/l) to 40 μg/dl, 30 μg/dl, 25 μg/dl, and (since the early 1990s) 10 μg/dl. Now it seems that there may be no completely safe blood lead concentration. Two studies reported side by side in the New England Journal of Medicine show effects at blood lead concentrations of less than 10 μg/dl. In the first study (2003;348:1866–74, see also editorial, ibid: 1703–4) there were similar results. The incidence in infancy (8 per 100 000 children per year) is much higher than in later childhood. The overall annual incidence was 1.24 per 100 000 children up to 10 years of age (Australia) and 1.13 per 100 000 up to 18 years (USA). The condition was more common in black children (indigenous children in Australia) than in white children. The most common type of cardiomyopathy was dilated cardiomyopathy (50% in Australia, 51% in USA) followed by hypertrophic (25% and 42%), restricted (3% in both countries), and unspecified (13% and 4%). Overall there was a male preponderance (but not for dilated cardiomyopathy in Australia). Familial cases were common (20% in the Australian study). Most cases were idiopathic but specific causes included neuromuscular disorders, lymphocytic myocarditis (40% of the Australian children with dilated cardiomyopathy who had a biopsy), inborn errors of metabolism, and malformation syndromes (most commonly Noonan’s syndrome). Neuromuscular disorders most commonly cause dilated cardiomyopathy while inborn errors and malformation syndromes are more often associated with hypertrophic cardiomyopathy. Myocarditis causes dilated cardiomyopathy.

Inherited long QT syndrome is usually caused by a mutation in one of six sodium or potassium channel genes (LQT1–LQT6). About half of the carriers of these mutations never develop symptoms and only about 4% die suddenly. Many (about one third) have a QT interval within the normal range. Researchers in Italy (New England Journal of Medicine 2003;348:1866–74, see also editorial, ibid: 1837–8) have reported on 647 patients from 193 families with mutations at the LQT1, LQT2, or LQT3 loci. The risk of syncope, cardiac arrest, or sudden death before the age of 40 years and before treatment was 30% (LQT1), 46% (LQT2), and 42% (LQT3). Risk depended on genotype, QTc, and sex. The authors of this paper propose risk stratification using these variables but the writer of the perspective article casts doubt on the usefulness of such an approach.

Many adolescents take up smoking and many adolescent girls have a preference for thinness rather than plumpness. Smoking and weight concerns have been associated in cross-sectional studies but there have been few prospective cohort studies. In Massachusetts (Tobacco Control 2003;12:289–95) 273 female adolescents aged 12–15 years at baseline were followed up for four years. Subjects who at baseline said that they believed it important to be thin (84% of subjects) were significantly more likely to become established smokers than those who did not. This is consistent with other evidence that smoking is an aid to thinness.

Studies on adults have suggested that fruits, vegetables and, more recently, beta-carotene might protect against asthma. Now a birth cohort study in the Netherlands (Thorax 2003;58:567–72) has provided evidence that milk fats might be protective in young children. In a cohort of 4237 children daily consumption of full cream milk or butter at age 2 years significantly reduced the prevalence of asthma or wheezing at age 3 years. Brown bread had a similar effect but consumption of fruits, vegetables, margarine, or fish had no apparent effect. The associations were unexplained.