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 tissues 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;348:1517–26, see also perspective, ibid: 1515–16) 172 children had blood lead concentrations measured at 6, 12, 18, 24, 36, 48, and 60 months of age and Stanford Binet IQ at 3 and 5 years. IQ fell more (by 7.4 points) for an increase of blood lead concentration from 1 to 10 μg/dl than for an increase from 10 to 30 μg/dl (2.5 points). The second paper (ibid: 1527–36) reports data from the third National Health and Nutrition Examination Survey of 1988–94 (NHANES III) showing that girls with a blood lead concentration of only 3 μg/dl are shorter and have slight pubertal delay compared with girls with a concentration of 1 μg/dl. Though slight, the delays were significant for African-American and Mexican-American girls although not for white girls. After the removal of lead from petrol in 1979 the mean blood lead concentration in American children fell from 15 μg/dl (in 1978) to 2 μg/dl (in 1999).

There may be a link between Down's syndrome and neural tube defect (Lancet 2003;361:1331–5, see also commentary, ibid: 1316). In Israel 493 mothers had had a child with neural tube defect (445) or isolated hydrocephalus (48). Among their 1492 other pregnancies the expected number of cases of Down's syndrome was 1.87 but the actual number was 11. In the Ukraine 516 mothers had a child with Down's syndrome. Among their 1847 other pregnancies the expectedumber of neural tube defects was 1.37 and the actual number seven. It is suggested that defective folate metabolism could be the causal link and that periconceptional folate might prevent Down's syndrome as well as neural tube defects. (There must be data already available. Are babies with Down's syndrome more likely to have neural tube defects and has periconceptional folate supplementation affected the incidence of Down's syndrome?)

Epidemiological studies of cardiomopathy in children in Australia (New England Journal of Medicine 2003;348:1639–46) and the USA (ibid: 1647–55, see also editorial, ibid: 1703–4) have given 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.

Coronary artery dilatation in children is not always due to Kawasaki's disease. In Southampton (Heart 2003;89:595–6) three children had conical dilatation of the right coronary artery (base of the cone at the entrance to the artery) in association with bicuspid aortic valve. Other congenital aortic abnormalities, not present in all three patients, included subaortic stenosis (two patients), aortic stenosis, aortic incompetence, dysplastic aortic valve, small left ventricle, ventricular septal defect, patent ductus arteriosus, and coarctation of the aorta. The coronary lesion was detected soon after birth in two patients and after some years in the third. The authors of this report suggest that all patients with a bicuspid aortic valve should be monitored for the presence of coronary dilatation.

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 perspective, 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. Why this should be, however, was not clear since there was no significant association between taking up smoking and dietary behaviour or belief that smoking is an aid to thinness.

Studies on adults have suggested that fruits, vitamins C and E, and 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 almost 4000 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 are unexplained.