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**1.1 Introduction**

The surgical correction of birth defects helped create the speciality of paediatric surgery during the middle of the last century. Around this time, pioneering neonatal operations were successfully performed to allow survival of babies with conditions like oesophageal atresia or congenital diaphragmatic hernia (CDH). Indeed, along with innovations such as parenteral nutrition, the concentration of surgical, anaesthetic, nursing and critical care expertise now allows high survival rates to be achieved for many previously fatal anomalies. For certain conditions that have high mortality and morbidity, fetal surgery aims to further reduce the harm of birth defects.

**1.2 Birth Defects Are Leading Causes of Global Infant Mortality**

Given the huge progress made in the treatment of infectious diseases in particular, birth defects are now emerging as the leading cause of infant mortality. Moreover, this state of affairs pertains not only to places with expensive healthcare systems but in fact anywhere that infant mortality rates have significantly fallen. Hence, as progress against other infant killer diseases continues it is likely that birth defects will gradually become one of the most significant global causes of infant mortality. In addition, birth defects are a leading contributor to both premature birth (itself a major cause of infant mortality) and chronic disability (with its substantial personal and societal costs). Tragically, many such problems are already preventable; for example, the birth defects associated with congenital rubella syn-
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drome may be virtually eradicated by an effective programme of maternal immunisation. Furthermore, a subset of neural tube defects continues to occur due to inadequate implementation of pre-conceptual folate prophylaxis. However, the epidemiological challenges for clinicians extend beyond the known, preventable defects to unsolved conditions and their changing circumstances (for instance, the increasing prevalence of gastroschisis Fig. 1.1).

Fig. 1.1 Gastroschisis – a birth defect on the move? Data from birth defect registries indicate a real and unexplained increase in gastroschisis prevalence. It remains to be seen if the severity of gastroschisis is also increasing: in this example, in addition to the gut, most of the liver lies outside the neonatal abdomen (image used with permission, courtesy of the author)

1.3 Birth Defects Epidemiology and Teratology

Although birth defects have been described with horror and fascination since antiquity, teratology and scientific birth defects epidemiology date, like paediatric surgery, from the mid-twentieth century. Key historical developments include the recognition of congenital rubella syndrome (noted by clinical ophthalmological examination) and the thalidomide disaster (phocomelia and other defects associated with maternal thalidomide administration for morning sickness). These episodes vividly illustrated the devastating consequences of prenatal infection and drug exposure. In addition, these chastening experiences highlighted the urgent need to formalise birth defects surveillance. Such monitoring of birth defects can now be said to serve a range of important purposes including early warning of an outbreak, identification of possible environmental or genetic causes, rational planning for neonatal surgical provision, facilitation of prenatal counselling based on accurate data, establishment of associations between birth defects and comparison of outcomes.

1.4 Causation of Birth Defects Remains Often Complex and Uncertain

Before considering the methods of birth defects surveillance, it is worth sketching the developmental biology that underpins birth defects from a surgeon’s perspective. Causes of birth defects can be classified as parental, fetal and environmental. Examples of the former include the impact of maternal and paternal age on the prevalence of Downs and Aperts Syndromes, respectively. Alternatively, maternal diseases such as diabetes are well-described risk factors for the formation of birth defects. Fetal causes include genetically determined inborn errors of metabolism such as those causing intersex anomalies in congenital adrenal hyperplasia, chromosomal lesions such as Downs Syndrome, Edwards Syndrome etc. and twinning with its increased risk of birth anomalies. Environmental causes include those related to prenatal addiction and drug exposure, such as alcohol, smoking, thalidomide, valproate, phenytoin, warfarin etc., as well as the impact of intrauterine infections, such as toxoplasmosis, rubella and cytomegalovirus. The impact of assisted reproductive technologies such as in vitro fertilisation and intracytoplasmic sperm injection on the prevalence of birth defects are actually quite difficult to assess. The suggestion that anomaly rates are higher in such assisted pregnancies needs to contend with the confounding increased rates of multiple pregnancy. Also, given the parents’ need to use assisted reproductive technology, it may be that they are importantly different to parents conceiving naturally; increased anomaly risk could therefore be due to parental abnormality and predisposition rather than a result of the techniques themselves.

Other environmental contributors to birth defects include “endocrine disrupters”; these oestrogenic compounds are conjectured to contribute to anomalies of sexual development in fetal males (e.g. hypospadias)