

EDITORIAL

Can birth defects be prevented?

Birth defects were recognized at birth or soon afterwards in just over 1% of a sample of 10,000 babies born in Port Moresby General Hospital, Papua New Guinea (1). Compared with the outcomes of similar studies in other countries, this prevalence appears quite low, but this should not be interpreted as a reason for inaction. A significant number of abnormalities pass undetected at birth, especially those of internal organs such as the heart. They will become apparent only later, and it is to be expected that the real occurrence of birth defects in this population is significantly higher than the prevalence recorded at birth.

Birth defects are an important public health issue. They have considerable consequences for the affected babies by contributing to infant mortality and handicap, and they influence the lives of family members and the provision of health care and education.

At the present time, substantial reductions in infant mortality are being achieved in Papua New Guinea through general improvements in health and living conditions, and it is to be expected that improvements in maternal health will be reflected in some reduction in the number of babies born with birth defects, especially those linked with environmental triggers such as disease and poor diet. However, in other countries it has been found that as infant mortality decreases, the proportionate contribution of birth defects to childhood problems increases, with the result that the health and educational services experience increasing pressure on them to provide special care for those affected (2).

Fortunately, this situation may not be inevitable: there is now growing confidence that a worthwhile proportion of birth defects can be prevented. Three degrees of prevention are discussed in the literature:

- ‘primary prevention’ means that measures are taken to prevent defects from arising in the first place
- ‘secondary prevention’ is concerned with prenatal diagnosis of defects and treatment or selective termination of affected babies
- ‘tertiary prevention’ refers to effective treatment after an affected baby has been born.

There has been the suggestion that more than 50% of birth defects are already preventable to some degree, provided that well-developed health care facilities are available and that the population is aware of factors that can cause abnormal development (3). This may be an overly optimistic goal in the Papua New Guinean context, where the prerequisites for prevention are still being established. Nonetheless, prevention even on a small scale is a worthwhile goal.

Clearly, primary prevention is the best option of the three, although our ability to achieve this is limited by our incomplete understanding of the causation of birth defects, both in Papua New Guinea and elsewhere. Although genetic and environmental factors have been identified in relation to some birth defects, the majority remain incompletely explained, and the aetiology of birth defects in Papua New Guinea has still to be investigated.

Secondary prevention reduces the number of babies born with birth defects, but inevitably raises complex ethical issues when decisions are made about affected babies. It also requires the widespread availability of expensive prenatal diagnostic facilities.

Tertiary prevention (treatment) can be effective in selected cases, as for example when a baby with cleft lip and palate has surgical correction and is then able to lead a

normal life, but in other cases the correction is less successful or may lie beyond the capacity of existing facilities.

Primary prevention begins from the premise that a woman in good health and living in a non-hazardous environment has a better chance of conceiving and giving birth to a normal baby than someone who is unwell or exposed to harmful teratogens. Knowledgeable health care workers can play an important part in achieving these aims by giving prospective mothers suitable advice and care before conception and during pregnancy (4).

Even when there is an existing maternal health problem, the risks of birth defects can be diminished by appropriate measures. For example, if a diabetic mother-to-be has poor glycaemic control in early pregnancy, her baby has increased risks of birth defects and spontaneous abortion. Both of these risks can be reduced if there is strict glycaemic control, especially if control is established before conception (5).

In the case of another metabolic disorder, phenylketonuria (PKU), considerable progress has been made in detecting the condition neonatally and then preventing mental retardation by provision of a special diet (6). However, as a result there are now increasing numbers of women with PKU who go on to become mothers themselves, and without suitable preparation they are at risk of having babies with microcephaly, mental retardation and cardiac defects as a result of their higher-than-normal blood levels of phenylalanine. Women with PKU should be helped to restrict their phenylalanine intake before and during pregnancy to reduce the risks for their babies (7).

Congenital hypothyroidism is another condition that can result in mental handicap if untreated, but which can be satisfactorily reversed if identified sufficiently early. In many countries, neonatal screening is now mandatory (8), and prophylactic iodine supplementation programs have proved beneficial in regions where goitre is endemic (9).

Three years ago the Department of Health in the United Kingdom issued a booklet advising potential mothers to supplement their usual dietary intake of folic acid as a way of

preventing neural tube defects such as spina bifida and anencephalus (10). While the early indications are that this is indeed an effective preventive strategy, it is unlikely to prevent all occurrences of neural tube defects. This is because neural tube defects (and some other birth defects) are very probably the result of a subtle interaction between environmental triggers and the genetic susceptibilities of individual babies. However, there seems to be little doubt that attention to diet in a more general sense will play an important part in primary prevention.

In some industrialized societies there is growing public concern about the presence of potentially toxic and teratogenic substances in the working and living environment, and the possibility that these may affect prenatal development (11). The issues are complex and conclusions hard to draw, but already employers are being pressured to introduce 'fetal protection policies' for their employees.

There is now considerable evidence that cigarette smoking during pregnancy has adverse effects on the baby and placenta. For example, the likelihood of a low birthweight baby is doubled for a mother who smokes, and there are increased risks of placental abruption, miscarriage, perinatal mortality and sudden infant death syndrome (12). Therefore it is sensible to strongly discourage this habit in women who are pregnant or who might become pregnant, even though smoking has not been linked with major developmental defects other than retarded growth.

Genetic counselling has an important preventive role already, and its contribution will become even more significant and effective as our understanding of genetic processes accelerates. A molecular revolution has swept through biology, and it is now possible to contemplate gene replacement in inherited conditions that have previously been considered untreatable. However, the ethical problems raised by genetic counselling and intervention are complex, and the development of new interventions sometimes outstrips our ability to make considered judgements about their use.

Given that some birth defects are preventable, and given the range of possible

preventive measures that are now being tested, it will become possible to devise appropriate preventive strategies for Papua New Guinea. Health promotion and education are parts of this process, and will benefit from the efforts of knowledgeable medical practitioners and educationalists. Sound aetiological and epidemiological studies in the Papua New Guinean context will also be needed to provide a basis from which to develop preventive measures.

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