MONITORING HUMAN BIRTH DEFECTS: METHODS AND STRATEGIES

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1. Introduction

Since the rationale and goals of a paper in multidisciplinary settings are often unclear to those outside the author's specialty, a statement of purpose may not be out of place. My interest is in the diminution of birth defects and detection of preventable environmental causes of such events.

This paper has been written assuming the reader has no previous knowledge of birth defects. Definition of medical terms not provided in the text are not crucial to the argument, but may be found in any medical dictionary. The record of the New York State Birth Defects Institute's Symposium of October, 1970 will present in much greater detail some of the themes discussed here [10].

Some of the material treated here was originally presented in more condensed form at a National Foundation Symposium in New York City on Environment and Birth Defects, January 27, 1971.

2. Definition and incidence of birth defects

A major human birth defect may be defined as an anatomical structural variant that produces a significant clinical or cosmetic effect. This definition is, of course, a somewhat loose one in that what may be abnormal in one setting may be acceptable in another. For the purposes of monitoring as discussed here, this vagueness will not be a problem. But it should be pointed out that reports of incidence of total birth defects in various groups cannot be compared unless the precise defects scored by the authors and their method of ascertainment are specified [11]. For the purpose of this discussion, the incidence of infants with major defect detectable at birth will be assumed to be about two per cent. The incidence of particular malformations is of course much rarer. Order of magnitude estimates for some of the most frequent major malformations are (in contemporary U.S.A.), anencephaly $1 - 2 \times 10^{-3}$, mongolism 10^{-3}, spina bifida $0.5 - 2.0 \times 10^{-3}$, polydactyly (whites) $0.4 - 0.7 \times 10^{-3}$, polydactyly (blacks) $5 - 10 \times 10^{-3}$. Some might call the latter a minor malformation. (The definition of a minor birth defect is given in Section 8.) But most types of major