PROBLEMS OF BIRTH DEFECTS

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From Hippocrates to Thalidomide and After

Original papers with commentaries by
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INTRODUCTION

Surprisingly, the beginning of a modern approach to the problems of birth defects is relatively recent and dates from Gregg's classical report in 1941 that mothers who contracted rubella during the first trimester of pregnancy gave birth to infants with severe multiple anomalies. For the first time, an environmental agent was found to be teratogenic in man and was documented in a thoroughly convincing manner. Since then, many important discoveries and significant developments have been made, particularly in the areas of environmental teratogenesis, hereditary mechanisms, and prenatal diagnosis.

In recent years, there has been an impressive surge of interest in the causes and prevention of birth defects. Undoubtedly this resulted not only from the thalidomide tragedy, but also from the steady decline in infant mortality from other causes, such as infectious and nutritional diseases. The magnitude of the problem is emphasized by a recent report which estimated that at least 6% of all infants are born with some genetic or developmental anomaly. Furthermore, congenital defects contribute to approximately 20% of all neonatal mortality.

This collection of articles and commentaries is an integration of information from many disciplines, and presents a comprehensive survey of both recent and previously reported work related to the major aspects of birth defects. In particular, an attempt has been made to provide a critical assessment of current concepts and to identify areas in need of further investigation.

The scope of this volume and space limitations precluded discussion of and reference to all papers of relevance or importance: a work of the present nature must necessarily be selective. Some good papers have been left out or given relatively little consideration. It is my hope that the list of Further References will be consulted and should compensate for this lack of completeness. The important milestones in the investigation of birth defects are highlighted by the inclusion of the original reports or extracts from them. In some cases, however, more recent articles have been given preference to original scientific papers. These have been selected primarily for their appropriateness and depth of interpretation.

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