Diseases of development: leveraging developmental biology to understand human disease

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Developmental disorders, often called ‘birth defects’, represent the number one cause of infant mortality in the USA and are nearly as lethal in the UK and Australia. The incidence of birth defects is highest in countries with the lowest income per capita. Owing to the highly heterogeneous nature of these conditions, they are commonly thought to be very rare. Collectively, however, they represent the leading biological cause of death for children of all ages. Indeed, in the USA, congenital anomalies are more than twice as lethal as pediatric cancers. These conditions all share an origin in embryonic development, so developmental biologists have a key role to play in addressing this important biomedical problem. In this Special Issue of Development, we highlight that role by presenting a collection of articles in diverse formats that are focused on developmental disorders.

The issue starts with a series of four Spotlight articles (Beames and Lipinski, 2020; Khokha et al., 2020; Leslie, 2020; Link and Bellen, 2020) designed to bridge the still-too-wide gaps that separate developmental biology, human genetics and epidemiology. The Special Issue also includes a short Primer article (Bruneau, 2020) on congenital heart disease, the most common form of congenital anomaly, and a longer Review (Bagnat and Gray, 2020) on the mechanisms controlling spine straightness, highlighting the often overlooked fact that developmental processes continue to play key roles in children and adolescents.

The core of this Special Issue is a series of 20 Research articles and reports that provide a broad view of the state of the field. Using mice, chickens, fish, frogs and human cells, the authors of the studies published here reveal new insights into conditions ranging from structural anomalies, to neurodevelopmental disorders, to pediatric cancers. As genetic causes are only one piece in the developmental disorder puzzle, the role of environmental factors and how these interact with genetic variation is also highlighted. Importantly, these papers also run the gamut from fundamental investigation of developmental mechanisms to direct modeling of genetic lesions known to underlie a human disease, further highlighting the ever-blurring lines between basic and clinical developmental biology.

We hope you enjoy reading this Special Issue and would like to thank everyone – authors and reviewers – who contributed to it. We also hope you will consider sending your next manuscript on this fundamental topic our way!

References


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