

Immunodeficiency Disorders and Molecular Techniques of Pediatric Pathology

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Received: November 16, 2021; Accepted: November 30, 2021; Published: December 07, 2021

Citation: Kayser K (2021) Immunodeficiency Disorders and Molecular Techniques of Pediatric Pathology. J Alzheimers Dis Parkinsonism S9: 033.

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Description

Pediatric pathology appears to be easily defined and narrow fields of medicine. However, we believe quite the opposite is true. Pediatric pathology is unique as a subspecialty of pathology in that it is defined by an age group rather than an organ system or process. As such, the subspecialty encompasses the study of disease from embryonic life to young adulthood. This aspect might be considered the vertical axis of pediatric pathology. Beginning at conception and terminating in late adolescence, the axis passes through the very dynamic stages of human development. With this realization, it becomes evident that pediatric pathology is a very complex subspecialty requiring a broad knowledge of normal and pathophysiologic processes that vary considerably through development. The consequent pathologic changes too can be different depending on the age at which injury occurs.

Because of social and economic factors, families in many countries have fewer children, so that the emphasis on the health and development of each individual fetus is raised [1]. Should a pregnancy fail, parent wish to know the cause of fetal loss. To examine the embryo and fetus, the pathologist must have a sound knowledge of embryology, genetics and the functioning of the maternal-paternal-fetal unit. Likewise, to examine congenital malformations at autopsy, the pathologist must have special knowledge and techniques so that the increasing number of known syndromes can be precisely diagnosed. It is of paramount importance of the family that a diagnosis be obtained on which genetic counseling can be initiated [2]. The perspective the pediatric pathologist acquires working in the fields of embryologic and fetal pathology, cytogenetics and teratology engenders a curiosity about developmental pathophysiology. Consequently, pediatric pathologists have contributed greatly to these areas of research.

During the newborn period and infancy, the pediatric pathologist's attention in the past was largely focused on genetic diseases, including the aforementioned problem of malformations, and on infectious diseases, metabolic disease and pathology of iatrogenic origin. Although a presumptive diagnosis can be made on clinical grounds, it is the role of the pediatric pathologist and laboratory colleagues to make the definitive diagnosis [3].

Recent studies also demonstrated the integral relationship of pediatric pathologists and biochemical and molecular pathologists in diagnosing metabolic disorders, also illustrated the infectious diseases and immunodeficiencies that have an effect solely or predominantly in early life. In late infancy and childhood, the pediatric pathologist plays a prominent role in the diagnosis and research of tumors. The tumors that occur in infants and children are distinct from those that develop in adults and they often exhibit unique clinical behavior so that specific therapeutic protocols are required. Today, light microscopic examination is more often the starting point in tumor diagnosis than the endpoint. Cytogenetic, flow cytometry, electron microscopy and molecular pathologic examination are now routinely required in the classification of many childhood malignancies [4]. It is the responsibility of the pediatric pathologist to ensure that appropriate evaluation is undertaken for each tumor.

Adolescent medicine is emerging as an area of great clinical concern. The pediatric pathologist and colleagues in the pediatric laboratory will find their services required, for example, in the investigation of infectious disease, adolescent nutritional disorders and substance abuse. Similar to infectious disease and oncologic pathology, forensic medicine spans the entire age range encompassed by pediatric pathology [5]. Because children can die unexpectedly and violently at any age, the pediatric pathologist must have knowledge of forensic medicine. Indeed, because of the uniqueness of pathology in early life, it is the pediatric pathologist who is often called on to provide expert interpretation.

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