

The Emerging Role of Forensic Pathology in Diagnosing Rare Diseases as Causes of Death

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Introduction

Forensic pathology plays an indispensable role in modern medicine and the legal system. It is a specialized branch of pathology focused on determining the cause of death in cases where the circumstances are suspicious, unexplained, or unexpected. Traditionally, forensic pathologists have been at the forefront of identifying common causes of death, such as trauma, poisoning and infectious diseases. However, over the past few decades, there has been a growing recognition of the importance of forensic pathology in diagnosing rare diseases as the underlying causes of death. The role of forensic pathology is evolving due to the increasing complexity of medical conditions and the rise in recognition of rare diseases, many of which may mimic common causes of death. Advances in molecular diagnostics, genetics and medical imaging have allowed forensic pathologists to identify previously undiagnosed conditions, shedding light on the mystery surrounding seemingly unexplained fatalities. The importance of forensic pathology in diagnosing rare diseases is critical not only for the understanding of the disease itself but also for helping to prevent future deaths, guide medical treatments and, in some cases, provide legal clarity in cases of medical malpractice or wrongful death [1].

This explores the emerging role of forensic pathology in diagnosing rare diseases as causes of death. It examines how advancements in forensic medicine, new technologies and interdisciplinary collaboration are enhancing the ability of forensic pathologists to detect rare diseases, the challenges they face and the broader implications of these discoveries for both medical practice and the legal system [2].

Description

Forensic pathology, by definition, involves the application of pathology techniques to determine the cause of death, especially in cases where the death is suspicious, unexplained, or potentially the result of a criminal act. Traditionally, forensic pathologists have focused on causes of death that are common and relatively easy to identify. Trauma includes blunt force trauma, sharp force trauma and gunshot wounds. Identifying these injuries often involves external examination and autopsy, which provide clear evidence of cause of death. Forensic toxicology is a sub-discipline within forensic pathology, where pathologists test for toxins, drugs, or alcohol that could have led to death. Infections such as pneumonia, meningitis, or sepsis can cause death and their identification may rely on microbial cultures and tissue examination. However, rare diseases were often overlooked or misdiagnosed due to the challenge of identifying these conditions in post-mortem examinations, especially when the signs are subtle or mimic more common diseases. Rare diseases, also referred to as orphan diseases, are conditions that affect a small number of people and may have obscure or atypical symptoms that make them difficult to diagnose even during life. Over the past few decades, there has been a significant

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increase in the awareness of rare diseases, in part due to advancements in genetics, molecular biology and diagnostic tools. Many rare diseases have genetic origins and some manifest only in specific circumstances, making them elusive to detection in clinical practice. Some of these diseases, however, can present as life-threatening conditions and in some cases, they may be the underlying cause of death [3].

Advances in genetic testing and molecular diagnostics have revolutionized the ability to identify rare diseases. The use of next-generation sequencing (NGS), whole-genome sequencing and targeted gene panels allows forensic pathologists to detect genetic mutations that could lead to conditions such as mitochondrial diseases, rare metabolic disorders, or genetic syndromes like Marfan syndrome or Ehlers-Danlos syndrome. The autopsy remains one of the most critical tools in forensic pathology. Forensic pathologists employ detailed examination techniques, including histopathological analysis, immunohistochemistry and electron microscopy. When dealing with rare diseases, these advanced autopsy techniques can help identify changes in tissues and organs that might indicate an underlying rare disease, even if the cause of death initially appeared to be something else. Integration of Interdisciplinary Expertise: As the field of forensic pathology evolves, collaboration between forensic pathologists, geneticists and clinicians is becoming more common. This interdisciplinary approach allows for a deeper understanding of rare diseases and better identification of subtle manifestations that could point to rare genetic or metabolic disorders [4].

One of the most important aspects of forensic pathology is its role in preventing future deaths. By identifying rare diseases post-mortem, forensic pathologists can contribute valuable information that may lead to earlier diagnoses in other family members or even in the broader community. When a rare disease is identified as the cause of death, forensic pathologists can provide genetic testing for surviving family members, helping to identify those who may be at risk of developing the same condition. Identifying rare diseases through forensic pathology can also provide crucial information for public health authorities. Recognizing patterns in rare diseases can help inform policies regarding screening, prevention and early diagnosis, which could ultimately save lives. Forensic pathology can also contribute to the broader field of medical research. By identifying previously undiagnosed rare diseases, forensic pathologists can help researchers gather data that may lead to the development of better diagnostic tools, treatments and preventive measures [5].

Conclusion

Forensic pathology is no longer limited to investigating common causes of death such as trauma, poisoning and infections. With the rise of rare diseases, forensic pathologists are playing an increasingly important role in diagnosing these conditions as causes of death. Through advancements in genetic testing, molecular diagnostics and interdisciplinary collaboration, forensic pathologists are now able to identify previously undiagnosed rare diseases that may have contributed to a person's death. While challenges remain in diagnosing rare diseases, the role of forensic pathology in this field is set to grow. As medical knowledge and technology continue to evolve, forensic pathologists will be better equipped to detect and diagnose rare diseases, potentially preventing future deaths and contributing to advances in medical science. By improving our understanding of these conditions, forensic pathology can make a meaningful difference not only in the field of law and medicine but also in the lives of families affected by these conditions.

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Conflict of Interest

The author declares there is no conflict of interest associated with this manuscript.

References

1. Patone, Martina, Xue W. Mei, Lahiru Handunnetthi and Sharon Dixon, et al. "Risks of myocarditis, pericarditis and cardiac arrhythmias associated with COVID-19 vaccination or SARS-CoV-2 infection." *Nat Med* 28 (2022): 410-422.
2. Schwab, Constantin, Lisa Maria Domke, Laura Hartmann and Albrecht Stenzinger, et al. "Autopsy-based histopathological characterization of myocarditis after anti-SARS-CoV-2-vaccination." *Clin Res Cardiol.* 112 (2023): 431-440.

3. Kogan, Evgeniya, Yuriy Berezovskiy, Olga Blagova and Anna Kukleva, et al. "Morphologically, immunohistochemically and PCR proven lymphocytic viral peri-, endo-, myocarditis in patients with fatal COVID-19." *Diagn. Pathol* 17 (2022): 31.
4. Robledo-Carmona, Juan, Maria V. Ortega-Jimenez, José M. García-Pinilla and Blanca Cabra, et al. "Severe cardiomyopathy associated to cocaine abuse." *Int J Cardiol* 112 (2006): 130-131.
5. Dettmeyer, R., M. Schlamann and B. Madea. "Cocaine-associated abscesses with lethal sepsis after splenic infarction in an 17-year-old woman." *Forensic Sci Int* 140 (2004): 21-23.

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