H49 Case Report: Institutional Experience With the Molecular Autopsy and Its Obstacles

Lorraine Lopez Morell, MD, Wake Forest Baptist Hospital, Dept of Pathology, Medical Center Boulevard, Winston-Salem, NC 27157; and Jerri McLemore, MD, Wake Forest School of Medicine, Dept of Pathology, Medical Center Boulevard, Winston-Salem, NC 27157*

After attending this presentation, attendees will better understand the ideal work-up in sudden unexplained toddler and young child deaths. Attendees will be more aware of the financial obstacles in place in the United States.

This presentation will impact the forensic science community by increasing awareness and opening a discussion about needed genetic testing in cases of sudden toddler and young child death.

According to the Center for Disease Control and Prevention Wonder database, sudden unexplained toddler and young child deaths, one to four and five to nine years old, respectively, account for 0.9/100,000 deaths per year (2011).[1] Like unexplained infant deaths, the cause for sudden unexplained toddler deaths can be elusive with no anatomic findings to explain the demise. "In fact, even after gross and histologic examination, at least 3% and perhaps as much as 53% of sudden deaths involving previously healthy children, adolescents, and young adults have no identifiable morphological abnormalities found at autopsy, remain unexplained, and are classified as autopsy-negative Sudden Unexplained Death (SUD)."[2] In cases of sudden toddler/child death, the ideal postmortem work-up would be similar to unexplained infant deaths and include a radiologic skeletal survey, bacterial and viral tissue cultures for microbiology, a full autopsy with detailed histological examination, review of medical and family history, and a thorough scene investigation. Because many cases have no findings whatsoever to explain the sudden death, blood and tissue samples should be retained for possible future studies, including genetic testing. The number of sudden unexplained toddler deaths in the United States due to genetic abnormalities is largely unknown. In general, medical examiners/coroners across the United States do not have easy access to molecular genetic testing due to financial constraints. The cost per positive diagnosis for long-QT syndrome alone can run up to $45,000.[3] As a result, opportunity to provide a definitive cause of death to the family is lost, as well as any chance of protecting other potentially at-risk individuals. As the molecular age of medicine advances, forensic pathology in the United States is slowly being left in the proverbial dust.

Within a few months' span, the Wake Forest Baptist Health (WFBH) department of pathology autopsy service autopsied two toddlers, ages two and four years of age, who had unexpected cardiopulmonary arrest and who were resuscitated and briefly placed on extracorporeal membrane oxygenation before being pronounced. Both toddlers had siblings; one had a twin. Full autopsies were performed but showed no clear grossly or histologically apparent etiology for their death except for a suggestion of possible cardiac fibroelastosis in one case. These two deaths were referred to the Pediatric Collaborative Care group at WFBH whose staff is composed of pediatricians in all subspecialties, medical geneticists, and other specialties as warranted for assessment. Circumstances and autopsy findings in both cases suggested a possible underlying genetic cause. This presentation highlights the current difficulties in obtaining funds in a cash-strapped medical examiner/coroner and hospital system and the various possibilities that may mitigate this problem.

References:

1. Centers for Disease Control and Prevention, National Center for Health Statistics. Underlying Cause of Death 1999-2011 on CDC WONDER Online Database, released 2014. Data are from the Multiple Cause of Death Files, 1999-2011, as compiled from data provided by the 57 vital statistics jurisdictions through the Vital Statistics Cooperative Program. Accessed at http://wonder.cdc.gov/ucd-icd10.html on Jul 30, 2014 12:16:37 PM
2. Tester D, Domingo A, Ackerman J, et al. Cardiac Channel Molecular Autopsy: Insights From 173 Consecutive Cases of Autopsy-Negative Sudden Unexplained Death Referred for Postmortem Genetic Testing
3. Winkel B, Larsen M, Olesen M, Tfelt-Hansen J, Banner J: The prevalence of mutations in KCNQ1, KCNH2, and SCN5A in an unselected national cohort of young sudden unexplained death cases. *J Cardiovasc Electrophysiol* 2012;23:1092–1098.

Molecular Autopsy, Sudden Unexplained Death, Genetic Testing

Copyright 2015 by the AAFS. Unless stated otherwise, noncommercial *photocopying* of editorial published in this periodical is permitted by AAFS. Permission to reprint, publish, or otherwise reproduce such material in any form other than photocopying must be obtained by AAFS.