Genetics education for physicians has been a popular publication topic in the United States and in Europe for over 20 years. Decreasing numbers of medical genetics professionals and an increasing volume of genetic information has created a dire need for increased genetics training in medical school and in clinical practice. This study aimed to assess how well pediatrics-focused primary care physicians apply their general genetics knowledge to clinical genetic testing using scenario-based questions. We chose to specifically focus on knowledge of the diagnostic applicability of Chromosomal Microarray (CMA) technology in pediatrics because of its recent recommendation by the International Standard Cytogenomic Array (ISCA) Consortium as a first-tier genetic test for individuals with developmental disabilities and/or congenital anomalies. Proficiency in ordering baseline genetic testing was evaluated for eighty-one respondents from four pediatrics-focused residencies (categorical pediatrics, pediatric neurology, internal medicine/pediatrics, and family practice) at two large residency programs in Houston, Texas. Similar to other studies, we found an overall deficit of genetic testing knowledge, especially among family practice residents and residents who attended medical school outside of the United States. Interestingly, residents who elected to take a genetics rotation in medical school scored significantly better than expected, as well as better than residents who did not elect to complete a genetics
rotation. We suspect that the insufficient knowledge among physicians regarding a baseline genetics work-up is leading to redundant (i.e. concurrent karyotype and CMA) and incorrect (i.e. ordering CMA to detect achondroplasia) genetic testing which is contributing to rising health care costs in the United States. Our results provide specific teaching points upon which medical schools can focus the education they provide about clinical genetic testing and suggest that increased collaboration between primary care physicians and genetics professionals would benefit patient health care overall.

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