

Clinical pathology selected abstracts

Editor: Deborah Sesok-Pizzini, MD, MBA, professor, Department of Clinical Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania, Philadelphia, and chief, Division of Transfusion Medicine, Children's Hospital of Philadelphia.

Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations

July 2019—Noninvasive testing for prenatal screening has been available through independent commercial companies and academic and hospital-based laboratories since 2011. In 2016, The American College of Medical Genetics and Genomics (ACMG) published an updated position statement on fetal aneuploidy, with specific recommendations for testing laboratories. Although some of the recommendations overlapped those of other organizations, the ACMG was unique in its recommendation that laboratories adhere to a specific format and content when reporting results. The authors studied U.S. commercial laboratories performing noninvasive prenatal screening (NIPS) as of Jan. 1, 2018. They excluded from the analysis umbrella companies, such as LabCorp, that did not directly sell commercially available NIPS and instead focused on their subsidiary testing companies. The authors collected patient education materials and sample reports for each NIPS test result via the companies' Web pages, exhibit booths at the 2018 ACMG annual meeting, and direct requests. They then divided into groups of two and each group analyzed one NIPS laboratory for compliance with the ACMG recommendations that pertained to laboratories in the 2016 ACMG position statement. A second analysis, in which one team member was assigned to analyze one or two of the recommendations across all NIPS laboratories, was subsequently performed. The authors examined 10 companies. Nine of the 10 reported fetal fraction in their reports, and eight of the 10 did not offer screening for autosomal aneuploidies beyond trisomy 13, 18, and 21. There was also inconsistency in the application and reporting of other ACMG recommendations. The authors concluded that the laboratories that were analyzed varied in their compliance with ACMG position statement recommendations. No company adhered to all ACMG laboratory guidance. The authors suggest that as laboratories continue to improve and revise their NIPS reports and communication with patients and providers, those findings may be helpful. In addition, consistency and transparency in test content, education, and reporting would benefit patients and providers.

Skotko BG, Allyse MA, Bajaj K, et al. Adherence of cell-free DNA noninvasive prenatal screens to ACMG recommendations. *Genet Med*. 2019. doi:1038/s41436-019-0485-2.

Correspondence: Dr. Brian G. Skotko at bskotko@mgh.harvard.edu

Red blood cells donated by smokers: an investigation of recipient transfusion outcomes

Blood collection facilities are not required to ask donors about cigarette smoking habits. However, previous studies have shown that red blood cells from chronic smokers have a 21.6 percent higher rate of baseline hemolysis and show a 281.7 percent increase in hemolysis when exposed to peroxidants relative to the rates in nonsmokers. Therefore, smokers may have higher hemoglobin levels, but smoking may adversely affect the quality of the donated RBCs due to higher carboxyhemoglobin (COHb) content and premature hemolysis. The authors of this study proposed that RBCs donated by smokers will have reduced efficacy due to reduced hemoglobin and hematocrit increments in recipients and increased COHb content. They tested RBC unit segments for nicotine and the major metabolite cotinine by mass spectrometry and then assessed COHb levels and investigated hemoglobin and hematocrit increments following transfusion of cotinine-positive and -negative units. They subsequently evaluated outcomes, retrospectively, in adult nonbleeding patients receiving one unit of RBCs. The results showed that 13 percent of the segments were positive for cotinine at levels consistent with current smoking. Of interest, the cotinine-positive RBCs showed significantly greater COHb content compared with cotinine-negative units (3.0 versus 0.8 percent). No significant change in vital signs or reactions was observed for the patients receiving cotinine-positive units. However, these units showed reduced hematocrit and hemoglobin increments (+1.2

percent and +0.4 g/dL) compared with cotinine-negative units (+3.6 percent and +1.4 g/dL). The authors concluded that the cotinine-positive units had greater COHb content and reduced hematocrit and hemoglobin increments following transfusion. They suggested that additional studies are needed, particularly in pediatric and neonatal patients because they may be more vulnerable to higher COHb content in donated RBCs. These cotinine-positive units may also have implications in chronically transfused patients, such as those with sickle cell disease or thalassemia, for which the efficacy of the incremental change in hemoglobin and hematocrit may be more significantly impacted.

DeSimone RA, Hayden JA, Mazur CA, et al. Red blood cells donated by smokers: A pilot investigation of recipient transfusion outcomes. *Transfusion*. 2019. <https://doi.org/10.1111/trf.15339>.

Correspondence: Dr. Robert A. DeSimone at rod9096@med.cornell.edu