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I. Background

Spinal muscular atrophy (SMA) is a leading inherited cause of infant death and is second only to cystic fibrosis (CF) as a common, life-limiting autosomal recessive disorder. SMA affects all racial and ethnic groups and has an incidence of 1 in 11,000 live births.¹ In March 2017, the American College of Obstetricians and Gynecologists (ACOG) released a committee opinion (691) stating that carrier screening for SMA should be offered to all patients considering pregnancy or currently pregnant.² A similar carrier screening recommendation has been in place for CF since 2001.³

II. Methods

To assess the utilization of SMA carrier testing following a change in ACOG recommendations, a retrospective analysis of tests ordered at one clinical laboratory over a two year period was performed. Data were analyzed from healthcare practices ordering at least one single gene carrier test for SMA within the total study period. Among those that ordered single gene carrier testing for SMA, a subset of healthcare practices that also ordered single gene carrier testing for CF were identified. Assessing data from the subset of healthcare practices ordering both SMA and CF single gene carrier testing throughout the study period allowed for analysis of ordering trends comparing utilization of two carrier tests, both recommended by professional societies.



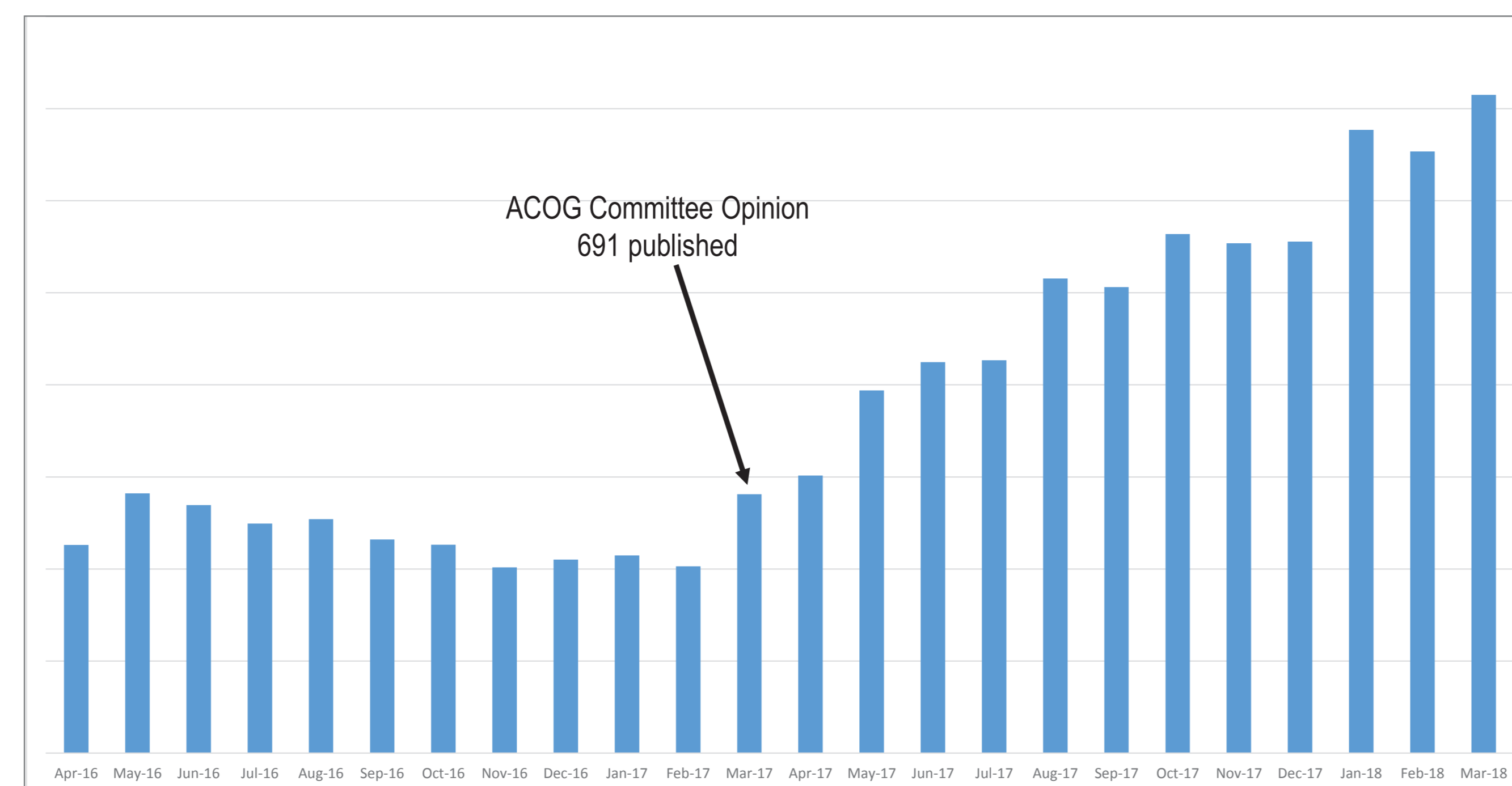
III. Results

I. SMA ordering trends before and after ACOG Committee Opinion 691

Figure 1 shows data collected from nearly 4,000 healthcare providers who ordered at least one SMA test in the study period.

- Nearly half the practices ordering testing (47.8%), had not ordered SMA screening before the updated guidelines in March 2017.
- On average, the laboratory received 1-2 SMA tests per month per healthcare practice before universal screening was recommended by ACOG.
- After the recommendations were updated to include SMA, the laboratory received 13-14 SMA tests per month per healthcare practice.
- There was a 68% increase in the number of healthcare practices ordering SMA carrier testing from the beginning of the study period to the end.

Figure 1. Number of SMA carrier tests ordered over a two year period

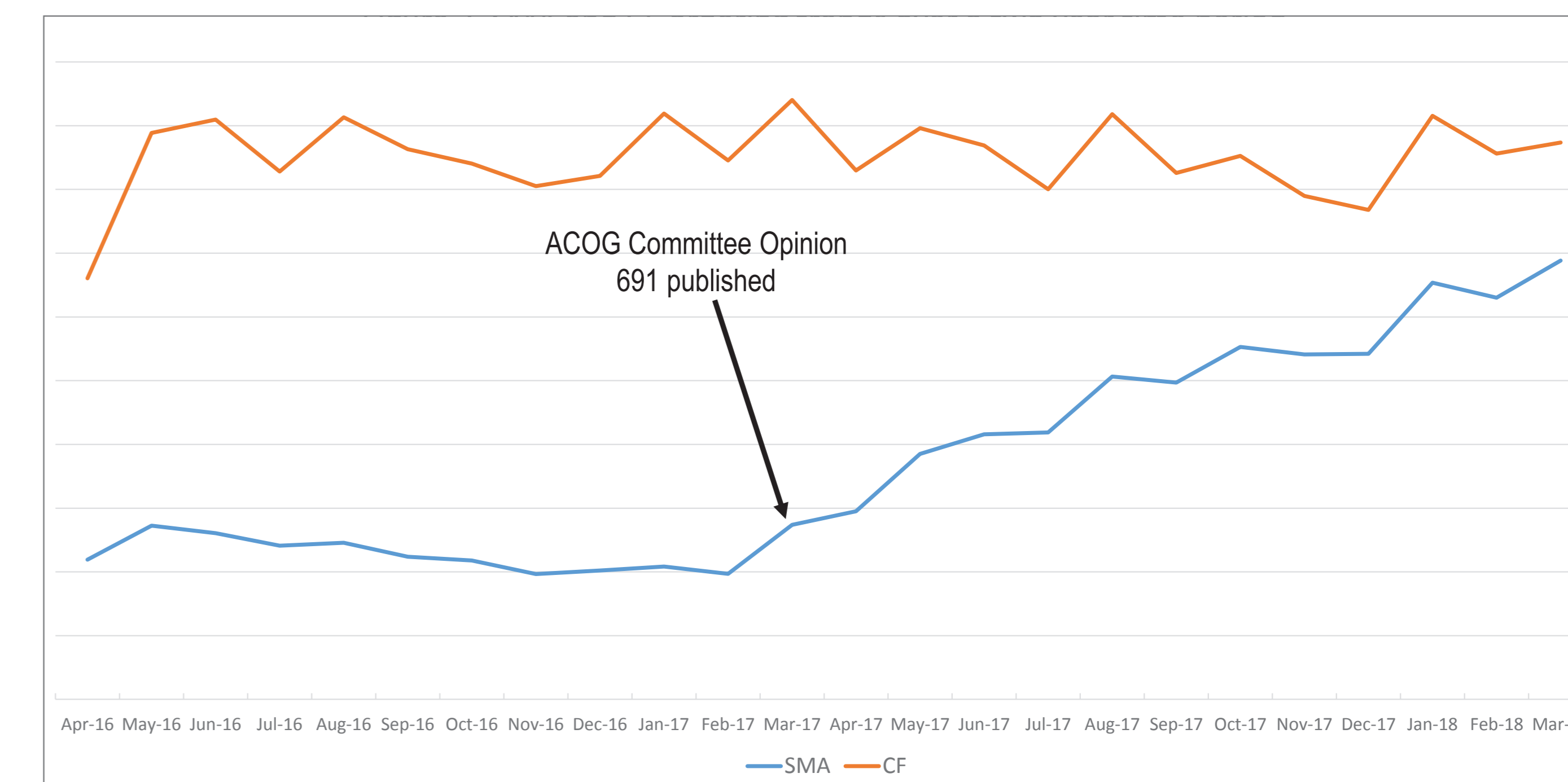


II. Comparison of SMA and CF ordering trends

Figure 2 compares the number of providers that ordered both CF and SMA carrier tests during the study period.

- Among practices regularly ordering carrier testing for CF, there was an increase in testing for SMA post-opinion.
- The number of CF tests received was 27% higher than SMA tests received, even one year after the updated committee opinion.

Figure 2. SMA and CF ordering trends over a two year time period



IV. Conclusions

Universal carrier testing for SMA, just like CF, is now recommended by both ACOG and ACMG. Post ACOG opinion, the number of single gene carrier tests for SMA has increased more than two and half times. Increased utilization is likely due in large part to the new recommendations from ACOG. However, despite the increase in SMA carrier tests and the increase in the number of ordering healthcare practices, the number of SMA tests post-opinion did not match the number of CF tests from healthcare practices sending both tests to our laboratory. These findings suggest increased adoption of SMA carrier screening guidelines, although not at the level of adoption of CF carrier screening. If full adoption had occurred, we would expect the number of SMA carrier tests to either exceed, or at least equal, the number of CF carrier tests ordered.

A limitation of this study is that it excludes healthcare practices ordering expanded carrier panels, which could capture additional data for increased adoption of SMA carrier testing.

Key takeaways:

- In March 2017, ACOG Committee Opinion 691 was published stating that carrier testing for SMA should be offered to all patients considering pregnancy or currently pregnant.²
- While there has been increased adoption of SMA carrier screening following societal recommendations, in the experience of one clinical laboratory it has not yet reached the expected level of adoption when compared with CF carrier testing.

V. References

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- American College of Obstetricians and Gynecologists, and American College of Medical Genetics. Preconception and prenatal carrier screening for cystic fibrosis. Clinical and Laboratory Guidelines. American College of Obstetricians and Gynecologists; Washington, DC: 2001.