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June 30, 2014

Diana T. Yu, MD, MSPH, Chair  
Health Promotion Committee  
State Board of Health  
Post Office Box 47990  
Olympia, Washington 98504-7990

Dear Dr. Yu:

Thank you for your memo dated April 23, 2014 regarding our request for rulemaking for Chapter 246-680 Washington Administrative Code (WAC). I am responding to address your questions on prenatal carrier testing. For clarity, your questions are italicized.

*Are there standards used to make determinations on genetic screening testing inclusion?* Yes, we used Wilson and Jungner's screening criteria as published in "Principles and practice of screening for disease" 1968<sup>1</sup>. These criteria have long been considered the gold standard in making decisions about what to screen for within a population. The State Board of Health (SBOH) newborn screening criteria are also rooted in these.

Population screenings for genetic conditions were introduced around the same time as these screening criteria including prenatal screenings using maternal age, ultrasound and biochemical markers looking for Down syndrome and tube defects. Additional prenatal criteria were also utilized such as: Pregnant women will be fully informed and their autonomy respected; and prenatal testing for adult onset conditions is strongly discouraged.

*If so, how are these standards applied, changed, and/or accepted?* Professional organizations such as the American Congress of Obstetrics and Gynecology (ACOG), and the American College of Medical Genetics (ACMG) have committees and task forces that review requests to

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<sup>1</sup> Wilson JMG, Jungner G. Principles and practice of screening for disease Geneva: WHO; 1968.  
[http://whqlibdoc.who.int/php/WHO\\_PHP\\_34.pdf](http://whqlibdoc.who.int/php/WHO_PHP_34.pdf)

add conditions to recommended practice, analyze existing literature, vet their recommendations with members and typically publish their recommendations in the form of practice guidelines or policies.

*Is there a national group that has worked on prenatal carrier testing? Both ACOG and ACMG have evaluated prenatal carrier screening. Recommended Carrier Testing Based on Ethnicity:*

	African American	Caucasian	Ashkenazi Jewish	Asian	French Canadian/Cajun	Hispanic	Mediterranean
Sickle Cell Disease	ACOG AAFP						
Tay-Sachs Disease			ACMG ACOG		ACOG AAFP		
Canavan disease			ACMG ACOG AAFP				
Cystic fibrosis	ACMG ACOG	ACMG ACOG AAFP	ACMG ACOG AAFP	ACMG ACOG	ACMG ACOG AAFP	ACMG ACOG	ACMG ACOG
Familial Dysautonomia			ACMG ACOG				
Niemann-Pick disease type A			ACMG				
Fanconi Anemia group			ACMG				
Mucopolidosis IV			ACMG				
Bloom syndrome			ACMG				
Gaucher disease			ACMG				
Alpha-thalassemia	AAFP			ACOG AAFP		AAFP	AAFP
Beta-thalassemia	AAFP			AAFP		AAFP	ACOG AAFP
Spinal Muscular Atrophy	ACMG	ACMG	ACMG	ACMG	ACMG	ACMG	ACMG

*If so, can you tell us more about the review process, the recommendations, and acceptance of those recommendations?* The professional organizations consider a variety of factors on which to base their recommendations. The prevalence of the disease, detection rate, natural history of disease and technical standards are all considered. The groups revisit recommendations every few years as new technology and information emerge. In addition, laboratory technical standards and guidelines are reviewed and revised by subcommittees in order to address issues surrounding non-validated commercially available tests.

*Is there a national group that sets the standard for what is to be included in a prenatal screening test made available to all pregnant women? Beyond the professional standards already*

described, there is no single national entity that sets standards. However, Chapter 246-680-020 WAC provides the SBOH an opportunity to initiate its own robust review in setting the standards for Washington State.

*Many tests are available commercially and unless there is a standard set for which tests are to be included, where do we begin?* Routine prenatal screening is already part of routine prenatal care and has been since the 1970's. Until two years ago, the "typical" pregnant women would be offered or receive:

- Review of family health history to determine if additional diagnostic testing may be indicated.
- Carrier screening based on ethnicity (see table below).
- First trimester ultrasound to establish dating of the pregnancy, if it is a singleton or multiple fetuses, and measure the thickness of the fetal neck (nuchal translucency) a risk factor for Down syndrome and cystic hygroma.
- Chorionic villus sampling to diagnose a chromosomal aneuploidy or other genetic condition if indicated by the family history.
- Second trimester ultrasound to evaluate fetal morphology.
- Maternal serum marker screening to identify neural tube defects, chromosomal aneuploidy or increased risk for pregnancy complications.
- Amniocentesis (assuming no CVS performed) to diagnose a neural tube defect, chromosomal aneuploidy or other genetic condition if indicated by the family history.

Since 2012, however, cell free fetal DNA (also known as Non-Invasive Prenatal Screening) became available allowing women to detect chromosomal aneuploidy through a blood test early in the pregnancy. This testing has a significantly higher sensitivity and equal specificity to the maternal serum marker screening. In addition, there are increasing data that suggest that the targeted carrier screening based on ethnicity is not as effective at identifying at-risk carriers as "pan-ethnic" carrier screening. (i.e., screening everyone for multiple conditions irrespective of their race). Should the SBOH choose to review the prenatal diagnosis rule, we would encourage that these two tests be considered.

*Are there adequate systems in place that provide follow up for affected folks, false positives and carriers?* Yes, the same system that is in place currently and includes maternal-fetal medicine clinics, regional genetics clinics, perinatologists, obstetricians and midwives across the state.

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We agree it is important to ensure a system is in place that can support patients through the prenatal screening process to avoid health disparities. Current wait times in prenatal clinics is typically under two weeks and patients with a positive test result receive priority scheduling to mitigate any anxiety the parents may feel. I have not heard a complaint from a Washington resident in over 15 years regarding any prenatal service so I have no reason to believe the system is inadequate to provide the necessary follow-up services needed because of prenatal screening.

I hope this information is useful to you and your committee. I look forward to working with you in the near future to update the Chapter 246-680 WAC: Prenatal Tests- Congenital and Heritable Disorders.

Sincerely,



Allene Mares, RN, MPH  
Assistant Secretary