Rule Comments

Documents and Comments

Document Title	File Name	Document Description	WSR#	Author	Author Organization	Author Phone	Deadline Date
Chapter 246-680 WAC State Board of Health Prenatal Test Congenital and Heritable Disorders.							
1 Comments	2024119sbohprenataltestscongenitaldiseases102final.pdf	Proposes to align the prenatal screening and diagnostic tests that considered medically necessary, and align with current national standards of care and current best practices. The screening and diagnostic tests in this rule are required to be included by certain insurers when authorizing requests or claims for prenatal screening or diagnosis without the requirement of case by case determination.	20-24-119	Samantha L Pskowski	SBOH - STATE BRD OF HEALTH	360-789-2358	01/06/2021
Oppose (1)	**Commenter**	**Commenter Phone**	**Commenter Email**		**Commenter Address**		
Oppose	Taryn Couture	8029221090	taryn.couture@powerslaw.com				

Dear Washington State Board of Health,

The Access for Expanded Carrier Screening Coalition (AECS) appreciates the opportunity to comment on the proposed revision to Section 246-680 of

the Washington Administrative Code on Prenatal Tests-Congenital and Heritable Disorders. AECS is comprised of a multi-disciplinary group of stakeholders with a mission to promote and improve access to carrier screening. The Coalition provides education on the utility of carrier screening for families, clinicians, and payers and works to ensure that all women of reproductive age have access to appropriate and medically necessary carrier screening.

Washington's Current Prenatal Screening Recommendations --
In Washington State Board of Health's (the Board's) reasons for supporting an update to their prenatal screening coverage they state "prenatal screening and diagnostic testing can have a significant impact on pregnancies at risk for a genetic condition or birth defect by: (a) Enabling early diagnosis or preventative approaches to reduce the amount of resources needed for postnatal diagnosis of symptomatic children; (b) Providing an opportunity to initiate appropriate health care services and interventions as soon as possible to improve the health of children and their families; and (c) Informing couples about health risks to current and future pregnancies to empower them to make informed pregnancy related health decisions." We could not agree more, identifying conditions early in pregnancy with carrier screening can avoid potential disease complications by initiating prompt treatment after birth, which can prevent permanent brain and/or organ damage. Furthermore, at-risk couples identified prior to pregnancy have additional options to prevent the birth of an affected child, such as adoption or gamete-donation.

As families and communities become more diverse, with the rate of interracial marriage increasing by 60% since 1980, it has become more challenging for Americans to accurately predict their ethnicity, making access to carrier screening available to everyone regardless of racial or ethnic background even more important.

Current Clinical Guidelines --
The American College of Obstetricians and Gynecologists is the premier professional organization of physicians that specialize in obstetrics and gynecology which puts forward prenatal practice guidance. In ACOG's most recent medical guidance they recommend that all pregnant and preconception patients should be offered carrier screening and that pan-ethnic carrier screening is an acceptable screening strategy. Furthermore, in 2017, ACOG released a committee opinion (#691) on carrier screening, recommending that all couples during pregnancy and preconception should be offered carrier screening, and includes a list of 18 conditions.

Recommendations for Updating Sec. 246-680-020(j) --
To ensure alignment with current standards of care as outlined in ACOG's Committee Opinion 691 (ACOG CO 691), the diversifying population, and the rationale for updating the Board's existing rules on prenatal and carrier screening, we recommend that you 1.) add five additional conditions in section

(j)(ii) to be screened for, as included in ACOG CO 691, in all populations irrespective of their racial/ethnic background or family history and 2.) Add Fragile X syndrome (FMR1) to the proposed list (j)(ii) for carrier screening irrespective of family history.

Recommendation 1. add five additional conditions in section (j)(ii) to be screened for, as included in ACOG CO 691, in all populations irrespective of their racial/ethnic background or family history:
1) familial hyperinsulinism (ACCB8),
2) glycogen storage disease 1A (G6PC) and 1B (SLC37A4),
3) Joubert syndrome type 2 (TMEM216),
4) Maple syrup urine disease type 1A (BCKDHA) and type 1B (BCKDHB), and
5) Usher syndrome type 1F (PCDH15) and type 3 (CLRN1).

Rationale --
• While the current proposal includes all the other diseases ACOG recommends, these five diseases are excluded without justification. The conditions listed above are as clinically significant and have similar carrier frequencies as the diseases included in the Board of Health's proposed list for coverage and fit with ACOG CO 691.
• Published empiric data from over 93,000 tests show the proportion of identified carriers of these additional five diseases is similar between patients who reported Jewish ancestry and those who did not.
• To ensure alignment with both ACOG CO 691 and to fit with the Board's carrier screening recommendations, we recommend these five conditions be added for coverage.

Recommendation 2. Add Fragile X syndrome (FMR1) to the proposed list (j)(ii) for carrier screening irrespective of family history.

Rationale --
• Fragile X syndrome is the most common hereditary cause of intellectual disability and the leading single-gene cause of autism. The carrier frequency of Fragile X syndrome is 1/260 and since it is inherited in an X-linked manner, female carriers have up to a 50% chance to have an affected child. Fragile X syndrome can occur in any ethnic group.
• After the conditions listed above, Fragile X syndrome is only disease listed in the ACOG guideline on carrier screening that is not explicitly included in the proposed state rule on carrier screening. While the ACOG recommendation is to offer screening for Fragile X based on a family history of intellectual disability, we recommend making this screening available to all patients given its severity and prevalence. Female carriers are typically

asymptomatic and in 30% of cases, there is no significant family history. Even ACOG acknowledges that restricting testing to those with a family history will miss a significant number of carriers. Furthermore, as the average age to be diagnosed with Fragile X syndrome is 3 years, 25% of families will have a second affected child before the first is diagnosed.

Conclusion --
We respectfully request that Washington State Board of Health include the above additional six conditions in their recommendations for carrier screening coverage to better align with ACOG's CO 691. Carrier screening is a powerful tool for supporting health reproductive decision making. Couples use the information provided for preconception decision-making or support further pregnancy screening and birth plans, and we want to be sure all genetically significant conditions are covered in the State of Washington. We appreciate your consideration and are ready to answer any questions or further information. For additional information on AECS visit accesstoecs.com.

Sincerely,
Access to Expanded Carrier Screening Coalition

3 Comments	Rules_9172_SA_SBOHPrenatalTest.pdf	Significant Legislative Analysis	20-24-119	Samantha L Pskowski	SBOH - STATE BRD OF HEALTH	360-789-2358	01/06/2021
Oppose (1)	**Commenter**	**Commenter Phone**	**Commenter Email**		**Commenter Address**		
Oppose	**Rachel Fidino**	**(509) 540-4308**	**rachel@newuwomensclinic.com**		**35 S Louisiana Street Suite A120, Kennewick WA 993**		

January 6, 2021

Washington State Board of Health
P.O. Box 47990
Olympia, WA 98504-7990

RE: Proposed Rule for Washington State Board of Health Chapter 246-680 WAC, Prenatal Tests – Congenital and Heritable Disorders

Dear Washington State Board of Health,

I am writing in response to the Washington State Board of Health's proposed rule for Chapter 246-680 WAC, Prenatal Tests – Congenital and Heritable Disorders to commend the Board on updating this rule to align with current prenatal care standards and to request the Board consider updating Section (j) to more completely reflect the most up-to-date carrier screening guidance.

I would like to introduce myself and position regarding prenatal genetics. I am double board certified in women's health and advanced genetics and am the Chief Executive Officer of a medical practice in Eastern Washington. I serve on the Board of Directors for Nurse Practitioners in Women's Health and on the Expanded Carrier Screening Coalition for the United States. I also serve on multiple other committees including ACOG and the CDC. I am a Key Opinion Leader for genetics as it relates to hereditary cancer syndromes, carrier screening, and non-invasive prenatal screening. I have provided my expertise on multiple national position statements as it relates to inherited conditions.

Current Guidelines:

As a clinician that routinely provides prenatal care to patients in the state of Washington, I strive to ensure that I align with the current standards as outlined by the American College of Obstetricians and Gynecologists (ACOG), which is the leading professional organization for clinicians that specialize in obstetrics and gynecology. As the Board is aware, carrier screening is a powerful tool that provides my patients important information on their risks to pass on genetic conditions. My patients utilize this information in a variety of ways including enabling early diagnosis and treatment to optimize clinical outcomes as well as allowing patients to make informed decisions regarding pregnancy planning and management.

ACOG has issued two committee opinions that outline carrier screening recommendations (ACOG Committee Opinion 690 and ACOG Committee Opinion 691. ACOG Committee Opinion 691(ACOG CO 691) outlines 18 specific conditions to be considered as part of routine carrier screening given their disease severity and prevalence. ACOG Committee Opinion 690 (ACOG CO 690) acknowledges that while traditionally carrier screening was targeted by ethnic background, given the increasing diversification of the United States' population, pan-ethnic screening is an acceptable approach as

it is becoming more difficult for Americans to accurately report their ethnicity.

Recommendation and Rationale:

In the Board's current proposed rule, section (j) (ii) outlines 12 conditions which can be screened for at any time during pregnancy, irrespective of family history. It is unclear why the remaining 6 conditions outlined in ACOG CO 691 are not included. In congruence with the recommendations from ACOG CO 691, I suggest that the rule be expanded to include the following 6 disorders:

•Fragile X syndrome
•Familial hyperinsulinism (ACCB8)
•Glycogen storage disease type IA (G6PC) and type IB(SLC37A4)
•Joubert syndrome type 2 (TMEM216)
•Maple Syrup Urine Disease type IA (BCKDHA) and type IB (BCKDHB)
•Usher syndrome type 1F (PCDH15) and type 3 (CLRN1).

These disorders share the same disease severity as the conditions currently included in section (j) (ii), many offer the opportunity to attenuate or prevent an adverse clinical outcome with early interventions, and/or the prevalence of carriers in WA is equal to or greater than the conditions proposed.

The following 6 conditions are described in greater detail as it relates to condition, clinical summary, and frequency of carriers in Washington State (2016-2020):

Fragile X Syndrome: Per the CDC, the most common known cause of inherited intellectual disability. Early intervention can improve outcomes. Carrier frequency: 1 in 222 individuals in WA state. More common than 8 of the conditions currently proposed.

Maple Syrup Urine Disease (MSUD): Inborn error of metabolism where life-threatening complications can arise in 7-10 days of life in the absence of proper treatment. Carrier Frequency: 1 in 255 in WA state. More common than 2 of the conditions currently proposed.

Glycogen Storage Disease type 1(a&b): Inborn error of metabolism that if left untreated can lead to crisis characterized by extremely low blood sugars.

Frequent feeding with a special diet is critical to survival. Carrier Frequency: 1 in 254 in WA state. More common than 8 of the conditions currently proposed.

Familial hyperinsulinism: Inborn error of metabolism impacting glucose metabolism. Affected infants require immediate infusions of glucose to prevent seizures and protect against brain damage. Carrier Frequency: 1 in 374 in WA state. More common than 5 of the conditions currently proposed.

Joubert syndrome: Impacts the development of structures in the brain leading to developmental delay and impaired muscle coordination. Carrier Frequency: Most common in the Ashkenazi Jewish population.
For AJ in WA, Joubert is just as common as Bloom Syndrome (1 in 128).

Usher syndrome: Hearing and vision loss of varying severity depending on subtype. Carrier Frequency: 1 in 523 in WA state.
More common than 4 conditions currently proposed.

Conclusion:

My patients who elect to pursue carrier screening find tremendous value in the results as it enables them to make informed pregnancy decisions that align with their personal preferences and values. I applaud the Washington State Board of Health for updating this important rule, and I respectfully request that the Board update section (j) (ii) of the rule to fully align with current ACOG standards of care by including the six conditions outlined above. In addition, I would request that the Board establish a regular review schedule for this rule given that standards of care evolve as new clinical data becomes available.

I appreciate your consideration of this request and remain available via phone (509-540-4308) or email (rachel@newuwomensclinic.com) to answer any questions or provide further information.

Sincerely,

Dr. Rachel M. Fidino CEO, MSN, ARNP, WHNP-BC, AGN-BC, DNP

Rule Comments

Documents and Comments

Support (2)	Commenter	Commenter Phone	Commenter Email	Commenter Address
	Amanda Vitale	6025824683	avitale@conafaygroup.com	2200 Pennsylvania Avenue NW, Suite 600W, Washingto

To whom it may concern:

The Coalition for Access to Prenatal Screening (CAPS) seeks to expand coverage of cell-free DNA-based noninvasive prenatal screening (cfDNA NIPS) for all pregnant women, under CPT codes 81420 and 81507, regardless of maternal age or baseline risk. CAPS submitted technical recommendations in 2019 to the draft Chapter 246-680, and we are pleased to do so again.

We applaud the Washington Board of Health for its draft update to Chapter 246-680 which listed “Prenatal cell free DNA testing performed after nine weeks of gestation for the detection of aneuploidy including trisomy 21, 18, 13, or the sex chromosomes” as a medical necessity.
We believe this change would better align with recent new guidelines from leading professional organizations in prenatal care.

In mid-August 2020, the American College of Obstetricians and Gynecology (ACOG) and the Society of Maternal and Fetal Medicine (SMFM) released Practice Bulletin 226: Screening for Fetal Chromosomal Abnormalities, to in part “further clarify methods of screening for fetal chromosomal abnormalities, including expanded information regarding the use of cell-free DNA in all patients regardless of maternal age or baseline risk.”

The Practice Bulletin states:
• Screening (serum screening with or without NT ultra-sound or cell-free DNA screening) and diagnostic testing (CVS or amniocentesis) for

chromosomal abnormalities should be discussed and offered to all patients early in pregnancy regardless of maternal age or baseline risk (Level A recommendation, based on good and consistent scientific evidence).
• A patient's baseline risk for chromosomal abnormalities should not limit testing options.
• Cell-free DNA is the most sensitive and specific screening test for the common fetal aneuploidies (Level A recommendation).
• [G]iven the personal nature of prenatal testing decision making as well as the inefficiency of offering testing only to patients at high risk, the current recommendation is that all patients should be offered both screening and diagnostic testing options.

Policies that cover cfDNA NIPS for only high-risk women are incongruous with guidelines and create disparities in care and unequal access for women without risk factors or advanced maternal age. We believe Practice Bulletin (PB) 226 eliminates the need for prior authorization since maternal age and baseline risk are no longer considered appropriate determinants of who should access cfDNA screening.

Since the Washington Health Clinical Committee voted to cover cfDNA NIPs without conditions in May 2020, we believe this rulemaking process will further expand access to this important prenatal screening tool for women in Washington.

We look forward to the January 2021 hearing.

Thank you,

Amanda Vitale

	Robert Nathan Slotnick	**7752196179**	**robert.slotnick@invitae.com**	**1605 Ashland Bluff Way**

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Document Title	File Name	Document Description	WSR#	Author	Author Organization	Author Phone	Deadline Date
I'd like to comment on the utility of NIPS (cell free DNA analysis for aneuploidy) in all pregnancies I am a high risk obstetrician and medical geneticist. My career and research has been shaped by the development of obstetric screening and diagnostic testing for fetal abnormalities including chromosome defects. I'd like to speak to the science and clinical utility of NIPS for all, but also comment on the goal of providing access to all populations for this technology. Because the science and clinical applications are so convincing, limited access due to location, ethnicity or financial considerations should not occur. This question has become one of justice and equality.							