Chapter 246-680 WAC– Prenatal Tests – Congenital and Heritable Disorders
Written Comments and Staff Recommendations (WSR 20-24-119)
Public Hearing, January 13, 2021

Comments on the Proposed Rule		
Commenters	**Summary of Comments**	**Recommendations**
Obstetrics and Genetics Clinician	The science and clinical applications of NIPS, cell free DNA analysis for aneuploidy, are so convincing, limited access due to location, ethnicity, or financial considerations should not occur. This is an issue of equity.	**Recommendation: Adopt as proposed.** Comments are consistent with the proposed rule and the significant analysis prepared by the Board of Health and the Department of Health.

Coalition for Access to Prenatal Screening (CAPS)	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. Updated guidelines through Practice Bulletin state that screening and diagnostic testing should be discussed and offered to all patients in pregnancy regardless of maternal age or baseline 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.	**Recommendation: Adopt as proposed.** Comments are consistent with the proposed rule and the significant analysis prepared by the Board of Health and the Department of Health.

Obstetrics clinician	As a practitioner, strive to align with the current standards as outlined by American College of Obstetricians and Gynecologists (ACOG). Carrier screening is a powerful tool that patients rely on for information regarding their risk of passing on genetic conditions. ACOG has issued two committee opinions outlining recommendations for carrier screening. The Board's proposed rule includes 13 conditions which can be screened for at any time during pregnancy. Suggest the rule be expanded to include the following 5 disorders as well: - Familial hyperinsulinism (ACCB8) - Glyocgen 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 included in the proposed rule, WAC 246-680-020(1)(j)(ii). Request that the Board establish a regular review schedule for this rule given that standards of care evolve as new clinical data becomes available.	**Recommendation: Adopt as proposed.** The proposed additional disorders to include in carrier screening are X-linked disorders that are covered under WAC 246-680-020(1)(j)(i). The proposed rule currently incorporates those conditions most routinely utilized because of race/ethnicity and certain disorders included in ACOG's recommendations outlined in Practice Bulletin 691.

Access to Expanded Carrier Screening Coalition	Agree with the inclusion of carrier screening as identifying conditions early in pregnancy with carrier screening can avoid potential disease complications by initiating prompt treatment after birth.
	
	Recommendations for updating WAC 246-680-020(j):
	
	- Add five additional conditions in section (j)(ii) to be screened for as included in ACOG 691 in all populations irrespective of their racial/ethnic background or family history. These five additional conditions are:
	  o Familial hyperinsulinism (ACCB8)
	  o Glyocgen storage disease type IA (G6PC) and type IB (SLC37A4)
	  o Joubert syndrome type 2 (TMEM216)
	  o Maple Syrup Urine Disease type IA (BCKDHA) and type IB (BCKDHB)
	  o Usher syndrome type 1F (PCDH15) and type 3 (CLRN1)
	- Add Fragile X syndrome (FMR1) to the proposed list for carrier screening irrespective of family history.
		Recommendation: Adopt as proposed.
		
		The proposed additional disorders to include in carrier screening are X-linked disorders that are covered under WAC 246-680-020(1)(j)(i). The proposed rule currently incorporates those conditions most routinely utilized because of race/ethnicity and certain disorders included in ACOG's recommendations outlined in Practice Bulletin 691.
		
		Recommendation: Amend WAC 246-680-020(j)(ii) to include Fragile X syndrome.
		
		Inclusion of Fragile X syndrome in the list of disorders to be screened for using carrier screening regardless of family history was incidentally removed in a drafting error. Fragile X was included in the significant analysis produced.