Significant Legislative Rule Analysis

Chapter 246-680 WAC
a Rule Concerning Prenatal Tests – Congenital and Heritable Disorders

November 2020

SECTON 1:

Describe the proposed rule, including a brief history of the issue, and explain why the proposed rule is needed.

There have been many advances in prenatal screening over the years. These newer procedures offer better detection rates for birth defects or genetic conditions, as well as lower false positive rates. The purpose of the proposed rule is to continue to ensure equity for accessing prenatal screening and diagnostic services for women that choose them and to bring the rule into alignment with national standards of care and current best practices.

In 1988, the Washington State Legislature passed legislation that (1) required healthcare providers treating pregnant women to inform them about the availability of prenatal screening and testing options (RCW 70.54.220); (2) required multiple payers to cover such services (RCW 48.21.244, 48.44.344 and 48.46.375); and (3) placed limitations on certain payers to ensure they did not cancel, reduce, or alter coverage provided solely based on results of a prenatal test (RCW 48.42.090). The State Board of Health (Board) has the authority to establish standards in rule for screening and diagnostic procedures during pregnancy when those services are determined to be medically necessary. The regulations were written to eliminate the coercive and unethical practices of some payers who offered to cover the costs of prenatal screening and diagnostic procedures only if patients signed an agreement that they would terminate the pregnancy if an abnormality was found.

All pregnancies have a 3-5% risk for a birth defect and may be at an additional risk for genetic disorders. Prenatal tests are available to provide information about some of these risks and can help improve health outcomes. Prenatal screening and diagnostic testing can have a significant impact on pregnancies at risk for a genetic condition or birth defect by:

- Enabling early diagnosis or preventative approaches to reduce the amount of resources needed for postnatal diagnosis of symptomatic children;
- 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
- Informing couples about health risks to current and future pregnancies to empower them to make informed pregnancy related health decisions.

The proposed rule modernizes the pre-natal screening and diagnostic tests for congenital and heritable disorders that are required to be covered by insurers covered by this rule. The proposed rule includes new test requirements as well as eliminates or updates criteria for coverage of certain tests, for example age.

SECTION 2:

Is a Significant Analysis required for this rule?

Yes, the Board and the Department evaluated the rule and determined it is a significant legislative rule as defined in RCW 34.05.328 and requires a significant analysis that includes a cost/benefit analysis.

However, it has been determined that no significant analysis is required for the following portions of the rule.

Table 1: Non-Significant Rule Identification

WAC Section	Section Title	Reason
WAC 246-680-010	Definitions	Most changes clarify existing definitions and do not impact existing rule requirements. Definitions were alphabetized and policy requirements such as screening timelines were moved from the definitions into the body of the rule. Removed the term "group B strep screening" from the definitions because the term was not used in the body of the rule. Language from that definition was moved to section WAC 246-680-020. Removed the definition for "Department" because it was not used in the rule. Changed "prenatal carrier testing" to "carrier screening" and expanded the definition to include X-linked and recessive conditions. Added a new definition for "prenatal cell free DNA screening". The impact of adding or modifying screening tests are analyzed in Section 5.
WAC 246-680-020	Board of health standards for screening and diagnostic tests during pregnancy	1(b) – no edits. 1(c) – Group B strep screening: non-significant edit made to incorporate the timeframe for testing (35-37 weeks of gestation) that was previously in the definition.

SECTION 3:

Clearly state in detail the general goals and specific objectives of the statute that the rule implements.

RCW 70.54.220 requires all licensed or certified prenatal healthcare providers to inform their pregnant clients about the availability of prenatal tests. RCW 48.21.244 (group disability insurance contracts), RCW 48.44.344 (group healthcare services contracts), and RCW 48.46.375 (group health maintenance organizations (HMO) agreements) require coverage of pregnancy-related services and benefits for prenatal diagnosis of congenital disorders in accordance with standards established by the Board. The goal of this rule is to ensure pregnant women covered under certain plans have access to accurate information about prenatal screening and testing procedures and will be able to choose with confidence whether to undergo prenatal testing based on their personal beliefs without coercion.

SECTION 4:

Explain how the Board determined that the rule is needed to achieve these general goals and specific objectives. Analyze alternatives to rulemaking and the consequences of not adopting the rule.

The current rule outlines screening and diagnostic tests that are considered to be medically necessary and are required to be included in benefits packages provided by certain insurers, health care service contractors, and HMOs. Prenatal screening and diagnostic testing can have a significant impact on pregnancies at risk for birth defects or a genetic condition by enabling early diagnosis, providing an opportunity for appropriate health care, and empowering women and couples to make informed pregnancy-related health decisions. Since the rule was last updated in 2003, cell free fetal DNA (cfDNA) or non-invasive prenatal screening has become common practice, along with expanded carrier screening and prenatal chromosomal microarray analysis.

Unless the rule is updated, health disparities may increase. Some women may not have the resources to pay for the new prenatal screening and testing options not currently covered by this WAC, whereas women with greater resources may be able to pay for these tests without insurance coverage.
This rule will update existing rules to reflect current standard of medical practice and modern technology. Failure to update the rule would mean certain insurers would continue to be only required to offer coverage for older standards of care, and Washington standards will continue to be out of alignment with national guidelines.

SECTION 5:

Explain how the Board determined that the probable benefits of the rule are greater than the probable costs, taking into account both the qualitative and quantitative benefits and costs and the specific directives of the statute being implemented.

As identified in Section 1, updates to the proposed rule bring the rule into alignment with national standards of care and current best practices and improve the clarity and usability of the rule. The Board and the Department determined the proposed revisions include some significant legislative rule changes that are subject to the requirements of RCW 34.05.328(5).

The proposed rule makes significant changes by amending requirements for several existing prenatal screening tests and the inclusion of new screening tests that meet current clinical recommendations.

WAC 246-680-020(1)(a), Expanding maternal serum marker screening: The proposed change expands maternal serum marker screening for pregnant individuals up until the 22nd week of gestation. Currently, the rule allows for coverage up until the 20th week of gestation.

Maternal serum marker screening remains an essential tool in the evaluation of pregnancy for trisomy 21, trisomy 18, and open neural tube defects. Additionally, abnormal analyte levels may be predictive of other high-risk conditions such as fetal Smith-Lemli-Opitz syndrome, X-linked sterol sulfatase deficiency, fetal growth restriction, poor pregnancy outcome, and maternal preeclampsia. Maternal serum marker screening should be available to those with a priori low risk for the primary conditions for which it screens, and for whom cell-free DNA (cfDNA) screening is not recommended or feasible such as a vanishing-twin gestation or in the case of recurrent failed cfDNA screening.

Benefits: In some cases, a pregnancy remains unrecognized until a later gestational age, or other medical information or screening results are not available until the mid-second trimester, making this a medically necessary option throughout the validated time frame of the screening from 15 to 22 weeks gestation. Allowing for individuals to receive this screening for an additional two weeks allows for screening to take place in the event of an initial error in estimating gestational age, which can often be off by two weeks. Additionally,

Costs: Maternal serum markers screening costs approximately $100-$200[1]. The proposed changes are anticipated to impact a small number of individuals, as data suggests that 35-38% of women choose to receive maternal serum marker screening and data in Washington suggests that less than 5% of those screenings take place after the 20th week of gestation[2,3,4].

[1] https://www.oregon.gov/oha/PH/DISEASESCONDITIONS/GENETICCONDITIONS/Documents/PPTserum.ppt
[2] https://www.cfp.ca/content/cfp/46/3/614.full.pdf
[3] https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.1090
[4] Provider data, University of Washington Prenatal Care Clinic, November 2020.

WAC 246-680-020(1)(d)-(e), Expanding prenatal ultrasonography: The proposed change expands prenatal ultrasonography to all women to establish viability and gestational age in first trimester and to establish fetal morphology in second trimester. Further allowing for additional prenatal ultrasonography at any time during the pregnancy under the existing list of circumstances, with edits for current standards of care and best practice including instances where there is a personal or family history of a congenital abnormality potentially detectable by ultrasonography.

This proposed change establishes prenatal ultrasonography as a standard of care exam during first and second trimesters in accordance with 2016 guidelines from the American College of Obstetricians and Gynecologists (ACOG)[5]. As explained by ACOG, ultrasonography is used by healthcare professionals to view the fetus and check on fetal health during pregnancy. It can detect certain congenital anomalies and may be diagnostic of a particular structural abnormality[6]. There are three types of prenatal ultrasound exams: 1) standard, 2) limited, and 3) specialized.

A standard ultrasound exam checks the fetus's physical development, screens for major congenital anomalies, and estimates gestational age**.** A standard ultrasound exam also can provide information about the following: the fetus's position, movement, breathing, and heart rate, estimate of the fetus's size and weight, the amount of amniotic fluid in the uterus, the location of the placenta, the number of fetuses, and if the fetus is in a good position it may be possible to tell the sex. A limited ultrasound exam is done to answer a specific question, such as fetal position in the uterus during labor. A specialized ultrasound exam is performed if a problem is suspected based on risk factors or other tests. For example, if there are signs that the fetus is not growing well, the fetus's growth rate can be tracked throughout pregnancy with specialized ultrasound exams. Depending on what the suspected problem might be, specialized techniques may be used, such as Doppler ultrasonography and 3-D ultrasonography[7].

Benefits: Prenatal ultrasonography is an accessible, non-invasive method of diagnosing pregnancy status, number of fetuses, abnormalities in amniotic fluid volume, fetal anatomy, fetal and placental position and level of fetal activity. The ACOG guideline indicates that ultrasonography can be used to further assess the risk identified by other factors (age or serum screening). Ultrasound screening may detect the physical attributes associated with trisomies 13 and 18, conditions like "Down syndrome [are] more elusive," and may require other testing methods to reach diagnosis.[8]

Cost: The average cost of a prenatal ultrasonography exam was estimated to be $336 using Washington state agency utilization data[9]. This price may vary for individuals dependent on the provider and the contracted rate for an individual patient's insurance coverage.

WAC 246-680-020(1)(f), Expanding amniocentesis: The proposed rule change removes specific criteria for coverage of amniocentesis and requires coverage for all women after fourteen weeks of gestation. The proposed rule change would align the rule with existing standard of care practice followed in prenatal settings and with the current guidelines for amniocentesis. ACOG and the Society for Maternal

[5] http://unmobgyn.pbworks.com/w/file/fetch/114688045/ACOGPracticeBulletin175UltrasoundInPregnancy.pdf
[6] Ibid.
[7] https://www.acog.org/Patients/FAQs/Ultrasound-Exams
[8] https://prenatalinformation.org/2016/04/29/acog-issues-new-prenatal-testing-guidelines/
[9] https://www.hca.wa.gov/assets/program/findings_decision_us_121010.pdf

Fetal Medicine (SMFM) 2016 recommend, as described in Practice Bulletins No. 77 and 88, that all women should be offered diagnostic testing regardless of maternal age or other risk factors[10],[11]. Amniocentesis is performed between 15 and 20 weeks; it is not recommended to be performed earlier than 15 weeks. The estimated procedure-related loss rate is approximately 0.11%. Loss rates for both amniocentesis and chorionic villus sampling (CVS), though, are found in high-volume, experienced centers and ACOG notes the low loss rates "may not apply to other situations," i.e. "among health care providers with less cumulative experience." Additionally, the American College of Medical Genetics (ACMG) recommends that amniocentesis be offered to all pregnant women as a diagnostic test.

Benefit: Amniocentesis aids in prenatal detection of chromosomal abnormalities and neural tube defects in the developing fetus. For a cohort of 100,000 pregnant women (1988 US natality cohort) 35 years of age at the expected date of delivery and without considering intangible benefits, amniocentesis and (CVS) each resulted in 485 births averted when the fetus had an abnormality compared with no prenatal testing. Incremental quality- adjusted outcomes relative to no prenatal testing were 192 for amniocentesis.

Cost: A 2012 Kaiser Health News report indicated the typical cost of amniocentesis was $2,500[12]. Examination of two Washington State hospital chargemasters showed costs of $1,423 and $1,710 for diagnostic amniocentesis[13].

WAC 246-680-020(1)(g) Expanding chorionic villus sampling: The proposed rule eliminates the existing criteria for coverage of CVS and requires coverage for all women between ten and fourteen weeks of gestation. The proposed change would align the rule with the existing standard of care practice followed in prenatal settings, and with the current guidelines for CVS. ACOG retains the recommendation from Practice Bulletins No. 77 and 88 that all women should be offered diagnostic testing regardless of maternal age or other risk factors. CVS is performed between 10 and 13 weeks.

Benefit: For a cohort of 100,000 pregnant women (1988 US natality cohort) 35 years of age at the expected date of delivery and without considering intangible benefits, amniocentesis and chorionic villus sampling each resulted in 485 births averted when the fetus had an abnormality compared with no prenatal testing[14]. Incremental quality- adjusted outcomes relative to no prenatal testing was 70 for chorionic villus sampling[15]. Its calculated procedure-related loss rate is 0.22%. While earlier studies had suggested an association with limb-reduction defects, the risk is not significantly greater than the general population, provided CVS is performed at or after 10 weeks of gestation[16,17].

Cost: $1,300-$4,800[18].

WAC 246-680-020(1)(h),_Inclusion of fetal diagnostic testing: The proposed rule creates a new requirement to cover fetal diagnostic testing including, "(i) Cytogenetic studies on fetal cells including

[10] https://pubmed.ncbi.nlm.nih.gov/17197615/
[11] https://pubmed.ncbi.nlm.nih.gov/18055749/
[12] https://khn.org/news/prenatal-blood-tests/
[13] University of Washington Medical Center; Multicare Puget Sound Hospitals, 2019
[14] https://www.sciencedirect.com/science/article/pii/S0015028212004384
[15] Ibid.
[16] https://pubmed.ncbi.nlm.nih.gov/17197615/
[17] https://pubmed.ncbi.nlm.nih.gov/18055749/
[18] https://www.valuepenguin.com/costs-common-prenatal-tests#cvs

chromosome analysis (karyotype testing), cytogenomic microarray analysis (CMA), and fluorescent in-situ hybridization (FISH) for any woman undergoing amniocentesis or chorionic villus sampling; and (ii) DNA testing, cytogenomic microarray analysis, biochemical testing, or testing for infectious diseases if medically indicated because of an abnormal ultrasound finding, intrauterine fetal demise, or known family history." The proposed change would align the rule with standard of care practice followed in prenatal settings, and with the current guidelines for these tests.

ACOG recommends, in Practice Bulletins No. 77 and 88 that all women should be offered diagnostic testing regardless of maternal age or other risk factors[19,20]. FISH should be considered a screening test due to false-positive and false-negative results having been reported. Any clinical decision should not be based solely on FISH, but after confirmatory diagnostic results or consistent clinical information, e.g. abnormal ultrasound findings or a positive screening result for Down syndrome or Trisomy 18. CMA should be made available to any patient choosing to undergo invasive diagnostic testing. In the case of an ultrasound finding of fetal structural abnormality, CMA is recommended as a primary test, unless the abnormality is "strongly suggestive" of a particular aneuploidy, in which case karyotype may be offered before CMA[21].

Benefit: Cytogenetic testing is an important tool to identify chromosomal abnormalities in the fetus[22]. Access to cytogenetic testing can allow pregnant women and their partners to make informed decisions about their care by providing additional information about genetic risk for certain abnormalities. For instance a negative test may result in not needing to receive other health care services while a positive test can give a patient information about whether to perform more invasive diagnostic testing.

Cost: The University of Utah Medical center cites a cost of $1,200 - $1,500 for the karyotype diagnostic test, $1,500 for FISH, and $4,800 for chromosomal microarray[23].

WAC 246-680-020(1)(i), Inclusion of prenatal cell free DNA testing: The proposed change creates a new requirement to cover, "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." The proposed rule change aligns the law with existing standard of care practice followed in most prenatal settings, and with the current guidelines for cell-free DNA testing (cfDNA). ACOG Practice Bulletin 162 recommends cell-free DNA screening may be offered anytime from 10 weeks gestation through the duration of the pregnancy.

Benefit: In a large, routine prenatal-screening population of 15,841 pregnant women, cfDNA testing for trisomy 21 had higher sensitivity, a lower false positive rate, and higher positive predictive value than standard screening with the measurement of nuchal translucency and biochemical analytes[24]. Additionally, a 2014 Canadian study demonstrates that introducing contingent cfDNA testing improves performance by increasing the number of cases of Down syndrome detected prenatally, and reducing

[19] https://pubmed.ncbi.nlm.nih.gov/17197615/
[20] https://pubmed.ncbi.nlm.nih.gov/18055749/
[21] https://prenatalinformation.org/2016/04/29/acog-issues-new-prenatal-testing-guidelines/
[22] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3282538/
[23] https://physicians.utah.edu/echo/pdfs/2017-12-01-genetic-screening-in-pregnancy.pdf
[24] https://www.nejm.org/doi/full/10.1056/NEJMoa1407349

the number of amniocenteses performed and concomitant iatrogenic pregnancy loss of pregnancies not affected by Down syndrome[25]. Costs are modestly increased, although the cost per case of DS detected is decreased with contingent cfDNA testing.

Cost: Washington claims data in 2018 show an average cost ranging $482 - $553[26]. Limitations of this dataset exist in that the claims are only representative of Medicaid Managed Care Organization and Public Employee Benefits Board claims.

WAC 246-680-020(1)(j), Inclusion of carrier screening: The proposed rule creates a new requirement to cover, "Carrier screening at any time during the pregnancy for:

(i) Recessive or X-linked conditions if indicated by a positive family history; and

(ii) Any of the following conditions irrespective of family history:

(A) Alpha-thalassemia (HBA1/HBA2);
(B) Beta-thalassemia;
(C) Bloom syndrome;
(D) Canavan disease;
(E) Cystic fibrosis;
(F) Familial dysautonomia (IKBKAP);(G) Fanconi anemia type C (FANCC);
(H) Gaucher disease (GBA);
(I) Mucolipidosis IV (MCOLN1); or
(J) Niemann-Pick disease (SMPD1);
(K) Sickle cell disease;
(L) Spinal muscular atrophy (SMN1);
(M) Tay-Sachs disease (HEXA).
(N) Fragile X Syndrome;

The proposed change would partially align the rule with existing standard of guidelines. Carrier screening is recommended by ACOG for 7 of the 14 genetic conditions included in this proposed revision (Alpha-thalassemia (HBA1/HBA2); Beta-thalassemia; Cystic fibrosis; Sickle cell disease; Spinal muscular atrophy (SMN1); Tay-Sachs disease (HEXA); and Fragile X Syndrome)[27]. Recent demographic changes in the US have resulted in the increased likelihood of diseases and conditions once found almost exclusively among people of specific ethnic backgrounds occurring in non-targeted groups. For example, up to 12% of infants diagnosed with a beta-hemoglobinopathy via newborn blood-spot analysis in California during the early 1990s were outside of the groups included in ACOG's carrier screening guideline[28]. The 2010 Census shows substantial increases in individuals reporting mixed racial ancestry,

[25] https://obgyn.onlinelibrary.wiley.com/doi/pdf/10.1002/pd.4311
[26] Data set includes Washington claims from Medicaid and Public Employee Benefit Board enrollees. https://www.hca.wa.gov/assets/program/cfdna-final-report-20191213.pdf
[27] https://www.acog.org/-/media/project/acog/acogorg/clinical/files/committee-opinion/articles/2017/03/carrier-screening-for-genetic-conditions.pdf
[28] F.E. Shafer, F. Lorey, G.C. Cunningham, *et al.* Newborn screening for sickle cell disease: 4 years of experience from California's newborn screening program.

especially among those of reproductive age and younger. Similarly, the Jewish intermarriage rate is currently 48%, assuring that diseases currently screened in the Ashkenazi Jewish population will persist in other groups, as has occurred with Tay–Sachs disease. The shift to pan-ethnic offering of any disorder screened can be summarized most simply as an equitable, effective model for an evolving population. In addition to removing ethnicity considerations, the ECS model also proposes expanding the list of diseases identified in routine carrier screening. For instance, data from a large multi-ethnic population showed that the risk of a collective group of 89 diseases exceeded that of open neural tube defects or trisomy 21 pregnancy for a 20-year-old woman[29].

The prevalence of Trisomy 21 and open neural tube defects has been used to justify universal screening for these disorders. Likewise, the rare nature of recessive diseases are also typically cited as an important criterion for population based screening. Expanded carrier screening incorporates both justifications. ACOG recommends that all women be provided counseling about carrier screening and that such counseling and subsequent screening should be performed prior to pregnancy. If an individual is found to be a carrier, the individual's reproductive partner should be offered testing as well to provide the most robust information about potential reproductive outcomes. Family history from the patient and partner should be obtained in order to screen for inherited risk. Carrier screening for a particular condition should generally only be performed once in a person's lifetime and records maintained[30].

Cost: One provider for carrier screening advertises a test for self-pay patients at $250[31].

Table 1: Carrier Screening Conditions in Proposed Rule by Prevalence

#	Condition Name	Condition Prevalence
(A)	Alpha-thalassemia (HBA1/HBA2);	~ 1: 11,000 (California population)
(B)	Beta-thalassemia;	~ 1000 (US)
(C)	Bloom syndrome;	~ 275 (Ashkenazi Jew)
(D)	Canavan disease;	1:6400 (Ashkenazi Jew)
(E)	Cystic fibrosis;	1:3900 (US)
(F)	Familial dysautonomia (IKBKAP);	1:3700 (Ashkenazi Jew)
(G)	Fanconi anemia type C (FANCC);	1:136,000 (US)

[29] I.S. Haque, G.A. Lazarin, M. Raia, H. Bellerose, E.A. Evans, J. Goldberg. Expanded carrier screening of 322,484 individuals: the case for going beyond cystic fibrosis Eur J Hum Genet, 23 (2015) - https://cs.stanford.edu/people/ihaque/posters/haque2015eshg.pdf

[30] https://www.acog.org/clinical/clinical-guidance/committee-opinion/articles/2017/03/carrier-screening-for-genetic-conditions

[31] https://www.invitae.com/en/individuals/reproductive-genetic-testing/carrier-screening/

(H)	Gaucher disease (GBA);	1:6000 (US) and 1:450 (Ashkenazi Jew)
(I)	Mucolipidosis IV (MCOLN1);	1:40,000 (US)
(J)	Niemann-Pick disease (SMPD1);	1:100,000 (US)
(K)	Sickle cell disease;	~100,000 (US)
(L)	Spinal muscular atrophy (SMN1).	1:10,000 (US)
(M)	Tay-Sachs disease (HEXA);	1:3,600 (Ashkenazi Jew)
(N)	Fragile X Syndrome;	1:4000 (males) and 1:6000-8000 (females)

A. https://thalassemia.com/documents/PHRESH-thalassemia-fact-sheet.pdf
B. https://www.cdc.gov/features/international-thalassemia/index.html
C. https://rarediseases.org/rare-diseases/bloom-syndrome/ (1:100 Ashkenazi Jew is a carrier)
D. https://rarediseases.org/rare-diseases/canavan-disease/
E. https://rarediseases.org/rare-diseases/cystic-fibrosis/
F. https://ghr.nlm.nih.gov/condition/familial-dysautonomia#statistics
G. https://rarediseases.org/rare-diseases/fanconi-anemia/
H. https://rarediseases.org/rare-diseases/gaucher-disease/
I. https://rarediseases.org/rare-diseases/mucolipidosis-iv/
J. https://rarediseases.org/rare-diseases/niemann-pick-disease-type-c/
K. https://rarediseases.org/rare-diseases/sickle-cell-disease/
L. https://rarediseases.org/rare-diseases/spinal-muscular-atrophy/
M. https://rarediseases.org/rare-diseases/tay-sachs-disease/
N. https://rarediseases.org/rare-diseases/fragile-x-syndrome/

WAC 246-680-020(1)(k), Inclusion of parental testing: The proposed rule creates a new requirement to cover, "Molecular genetic or cytogenetic testing of parents to allow for definitive fetal testing, or parental testing to better inform results when fetal testing results yield uncertain significance." Parental testing in certain situations can provide definitive information when fetal diagnostic testing is inconclusive. This proposed change is in line with current clinical recommendations to offer counseling and screening for all patients.

Benefit: Parental testing is sometimes necessary to interpret genetic testing results. Should a fetal molecular, microarray or karyotype result yield ambiguous results (e.g. a variant of unknown significance) comparing that result to the parental results can help determine if the variant is familial and not pathologic or if it is new and therefore more likely to explain the condition.

Cost: The University of Utah Medical center cites a cost of $1,200 - $1,500 for the karyotype diagnostic test and $4,800 for chromosomal microarray. [32]

Cost Benefit: The probable benefits described in this analysis are substantial in improving reproductive health planning. The proposed rule expands access to numerous prenatal tests that can provide individuals with information to make an informed choice regarding reproductive planning. Carrier screening can provide prospective parents with information on the risk of a child having a recessive gene abnormality. This information can therefore allow individuals to make informed decisions around reproductive planning.

In addition to the qualitative benefits of prenatal screening, quantitative studies have shown that prenatal tests have the potential to generate significant cost-savings. The tests range in cost from approximately $336 - $4,600. This cost is spread between the insured and the insurer in most instances, with the amount paid by each varying with each insurance plan. A birth averted with the detection of a fetal abnormality, for example through amniocentesis relative to no prenatal testing and without intangible benefits was $183,299 for amniocentesis. The incremental cost per quality-adjusted outcome was $464,221 for amniocentesis[33]. Using the same parameters, CVS was found to have incremental cost per birth averted when the fetus had an abnormality was $199,381 for CVS. The incremental cost per quality-adjusted outcome was $1,407,563 for CVS[34]. In addition to savings from births averted, prenatal tests, specifically ultrasonography, have the ability to detect instances that may result in pre-term birth and allow medical providers and patients to take appropriate steps. For example, a routine ultrasound has the potential to identify a short cervix and risk of pre-term birth. One study found that universal screening with progesterone gel treatment when indicated resulted in cost-saving of $9,982/QALY when compared to no screening or treating[35]. Additionally, second trimester ultrasound and serum screening are able to detect and diagnose spina bifida myelomeningocele, the most common type of spina bifida, where fetal surgical options exist. One study found both cost-effectiveness and cost-benefit of fetal surgical correct, with an incremental cost-effectiveness ratio of $35,531 per quality of life year gained[36].

Based on this evidence, the Board determined that the benefits of these prenatal tests outweigh the costs.

SECTION 6:

[32] https://physicians.utah.edu/echo/pdfs/2017-12-01-genetic-screening-in-pregnancy.pdf
[33] http://www.crd.york.ac.uk/crdweb/ShowRecord.asp?ID=21995005500
[34] Ibid.
[35] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2878953/
[36] https://obgyn.onlinelibrary.wiley.com/doi/full/10.1002/uog.11176

Identify alternative versions of the rule that were considered, and explain how the Board determined that the rule being adopted is the least burdensome alternative for those required to comply with it that will achieve the general goals and specific objectives state previously.

An alternate to rulemaking is leave this rule as is and allow each carrier with plans subject to this rule to decide what they will cover as new technologies emerge related to prenatal genetic screening and testing. This is suboptimal from a state public health perspective as it serves to increase health care disparities in Washington State, since the quality of care will be determined by access to a given health plan and ability to pay.

The Board considered expanding the instances in which non-invasive prenatal screening (NIPS) would be required to be covered. The proposed rule allows for NIPS be limited to aneuploidy; however, NIPS can also be used in instances of screening for single gene disorders. The Board chose not to pursue including this in the rule, which would have increased costs.

The Board also considered requiring carrier screening for recessive X-linked disorders regardless of family history. This approach would have resulted in increased costs. Instead, the proposed rule requires family history for carrier screening of recessive X-linked disorders.

SECTION 7:

Determine that the rule does not require those to whom it applies to take an action that violates requirements of another federal or state law.

The rule does not require those to whom it applies to take an action that violates requirements of another federal or state law.

SECTION 8:

Determine that the rule does not impose more stringent performance requirements on private entities than on public entities unless required to do so by federal or state law.

The rule does not impose more stringent performance requirements on private entities than on public entities.

SECTION 9:

Determine if the rule differs from any federal regulation or statute applicable to the same activity or subject matter and, if so, determine that the difference is

justified by an explicit state statute or by substantial evidence that the difference is necessary.

The rule does not differ from any applicable federal regulation or statute.

SECTION 10:

Demonstrate that the rule has been coordinated, to the maximum extent practicable, with other federal, state, and local laws applicable to the same activity or subject matter.

There are no other applicable laws.