COLORADO
Department of Public Health & Environment

Dedicated to protecting and improving the health and environment of the people of Colorado

To: Members of the State Board of Health

From: Dan Wright, Newborn Screening Program Manager, Laboratory Services Division

Through: Tista Ghosh, Deputy Chief Medical Officer TG

Date: February 1, 2016

Subject: **Request for Rulemaking Hearing**
Proposed Amendments to 5 CCR 1005-4, Newborn Screening and Second Newborn Screening, with a request for the rulemaking hearing to occur in April 2016.

The Colorado Department of Public Health and Environment (CDPHE) is requesting that the Board of Health amend 5 CCR 1005-4 to delay the initiation of Pompe disease screening. After the September 2015 rule making hearing, the Laboratory Services Division (LSD) fiscal services staff performed further analysis of the Newborn Genetics Fund expenditures and revenues. The Division determined the costs to screen for Pompe disease beginning in July 2016 were greater than originally projected, and therefore would result in the Newborn Genetics Cash Fund balance being depleted within the first two years of implementation.

Current projections show that without a fee increase, the cost of adding the Pompe test to the existing panel is not sustainable long-term. Moreover, because increased fee revenue is subject to TABOR restrictions, any additional cash fund revenues generated would result in increased TABOR refunds in subsequent years to the detriment of the State General Fund. Therefore, although the Department is in support of adding the Pompe Disease test to the Newborn Screening panel, the Department is requesting a rule-making hearing to delay the date of initiating the test.

It is the Department's intent to begin testing for Pompe Disease beginning in FY 2019-20 (effective July 1, 2019). This will require the Department to increase the fee by approximately $10-13 per test. Because of the existing TABOR implications of generating additional fee revenue, the Department will be required to seek approval from the Governor's Office prior to implementation of a fee increase. In addition, the Joint Budget Committee will need to approve an increase of spending authority, which will authorize Department to initiate Pompe testing.

Because of the time and financial investment needed, the Department's practice is to request that a condition be added but delay the effective date so the Department can obtain the necessary equipment, develop the necessary screening protocols and if needed, adjust the fee to meet the increased costs. This practice ensures that the Department is allocating resources appropriately and in alignment with the direction of the Board. The Department is keeping with that practice here. Including Pompe disease on the newborn screening panel but extending the effective date from July 1, 2016 to July 1, 2019 gives the Department the direction and the time needed for implementation.

STATEMENT OF BASIS AND PURPOSE
AND SPECIFIC STATUTORY AUTHORITY
for Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening

Basis and Purpose:
Under Section §25-4-803 and §25-4-1004(1)(b), C.R.S., the Board of Health has the authority to add conditions to the newborn screening panel.

In the fall of 2015, the Department recommended that the board add Pompe disease to the Newborn Screening Panel. The Department's recommendation aligned with the national Recommended Uniform Screening Panel (RUSP)(Pompe disease was added in March 2015) and the recommendation of the Colorado Newborn Screening Advisory Committee.

The September 2015 rulemaking packet reviewed the Colorado Newborn Screening and Genetic Counseling and Education Act, §25-4-1001 to 1006 C.R.S., criteria for the inclusion of disorders for newborn screening. The criteria and the analysis of the criteria provided to the Board in September 2015 are delineated below. To assist the reader in distinguishing the historical and current information, the information provided in September 2015 is italicized. The footnote numbers have changed to align with this document.

1. *"Condition for which the test is designed presents a significant danger to the health of the infant or his family." Pompe disease is an inborn errors of metabolism in which the deficiency of the enzyme acid alpha-glucosidase (GAA) results in accumulation of excess glycogen. This accumulation of glycogen leads to progressive weakness/damage of muscles and cardiomyopathy. There is a broad clinical spectrum of the disease. Patients presenting with symptoms within the first year of life are classified as infantile-onset Pompe disease. Infantile-onset Pompe disease accounts for approximately 30% of patients. Patients presenting after the first year of life are classified as late-onset. Those with infantile-onset Pompe disease begin to show symptoms within the first days of life with cardiac, musculoskeletal, respiratory, and gastrointestinal involvement. These symptoms include massive cardiomegaly and cardiomyopathy, progressive muscle weakness with muscle wasting, secondary respiratory insufficiency and recurrent infections, poor feeding, and poor growth. Patients with infantile-onset are cognitively normal. If untreated, patients with infantile-onset Pompe disease will often die within the first year of life from cardiac failure or secondary respiratory disease.[1] Patients with late-onset Pompe disease may or may not have cardiac involvement. They have progressive skeletal muscle symptoms and respiratory involvement but with slower progression. Often the muscle weakness can go unrecognized in childhood. Many seek care in adulthood but diagnosis is often delayed by 8-10 years. If unrecognized and untreated, lifespan is shortened.[2]*

   "Condition is amenable to treatment." Patients with Pompe disease are treated with enzyme replacement therapy (ERT) administered once every two weeks by

[1] Kishnani PS, Hwu WL, Mandel H, Nicolino M, Yong F, Corzo D. A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. J Pediatr. 2006 May;148(5):671-676.

[2] Winkel LP, Hagemans ML, van Doorn PA, Loonen MC, Hop WJ, Reuser AJ, van der Ploeg AT. The natural course of non-classic Pompe's disease: a review of 225 published cases. J Neurol. 2005 Aug;252(8):875-84.

intravenous infusion. ERT is an effective treatment, but not curative. Data shows improved survival if treatment is initiated within first 6 months of life with normalization of cardiac size.[3] However, ERT does not reverse skeletal muscle damage.[4] Therefore, initiation of treatment within first weeks of life would result in best outcome.[5]

Outcome of treatment is also influenced by CRIM status (cross reacting immunologic material). CRIM+ patients (75%) have some residual enzyme activity. Therefore, CRIM+ patients do not typically produce significant antibodies to ERT and hence, have a generally better response to ERT. CRIM- patients (25%) have no residual enzyme activity and historically have a poor response to ERT with minimal functional gains. However, new research shows that immune-modulation prior to or simultaneous to ERT can decrease antibody titer and improve response to therapy.[6]

Recent published literature reported on 10 patients (all CRIM+) with infantile-onset Pompe diagnosed by newborn screening that began receiving treatment between 6-34 days of life. After a median treatment time of 63 months (28-90 months), all could walk independently and none required mechanical ventilation (breathing machine). All 10 patients were able to participate in daily activity but did show some muscle weakness, speech issues, and eye findings.[7] Data from the Genzyme Pompe registry showed improved survival and mechanical ventilation free status for patients with infantile-onset Pompe disease that were treated prior to 3 months of age (includes both CRIM+ and CRIM-).[8]

Initiation of ERT in clinically diagnosed late-onset Pompe patients has been proven to improve outcome.[9] However, there is no data evaluating presymptomatic treatment in this population. Since ERT cannot reverse muscle damage, experts in the field

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[3] Chen LR, Chen CA, Chiu SN, Chien YH, Lee NC, Lin MT, Hwu WL, Wang JK, Wu MH. Reversal of cardiac dysfunction after enzyme replacement in patients with infantile-onset Pompe disease. J Pediatr. 2009 Aug;155(2):271-5

[4] Kishnani PS, Corzo D, Leslie ND, Gruskin D, Van der Ploeg A, Clancy JP, Parini R, Morin G, Beck M, Bauer MS, Jokic M, Tsai CE, Tsai BW, Morgan C, O'Meara T, Richards S, Tsao EC, Mandel H. Early treatment with alglucosidase alpha prolongs long-term survival of infants with Pompe disease. Pediatr Res. 2009 Sep;66(3):329-35.

[5] Chien YH, Lee NC, Thurberg BL, Chiang SC, Zhang XK, Keutzer J, Huang AC, Wu MH, Huang PH, Tsai FJ, Chen YT, Hwu WL. Pompe disease in infants: improving the prognosis by newborn screening and early treatment. Pediatrics. 2009 Dec;124(6):e1116-25

[6] Messinger YH, Mendelsohn NJ, Rhead W, Dimmock D, Hershkovitz E, Champion M, Jones SA, Olson R, White A, Wells C, Bali D, Case LE, Young SP, Rosenberg AS, Kishnani PS. Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genet Med. 2012 Jan;14(1):135-42.

[7] Chien YH, Lee NC, Chen CA, Tsai FJ, Tsai WH, Shieh JY, Huang HJ, Hsu WC, Tsai TH, Hwu WL. Long-term prognosis of patients with infantile-onset Pompe disease diagnosed by newborn screening and treated since birth. J Pediatr. 2015 Apr;166(4):985-91. *(Document included within.)*

[8] Kemper, A.R. *et al*. Evidence Report: Newborn Screening for Pompe Disease 2013 June 3.

[9] Anderson LJ, Henley W, Wyatt KM, Nikolaou V, Waldek S, Hughes DA, Lachmann RH, Logan S. Effectiveness of enzyme replacement therapy in adults with late-onset Pompe disease: results from the NCS-LSD cohort study. J Inherit Metab Dis. 2014 Nov;37(6):945-52

predict that presymptomatic treatment in late-onset patients will have benefit while others recommend close monitoring for subclinical muscle findings.[10]

2. *"Incidence of the condition is sufficiently high to warrant screening." The incidence of Pompe disease was historically reported as 1 in 40,000. However, recent anonymous screening of dried blood spots followed by confirmatory DNA studies in Washington state found an incidence of 1 in 28,000.*[11] *Approximately 30% of clinically diagnosed patients have infantile-onset Pompe disease.*[12]

3. *"The test to detect the condition meets commonly accepted standards of reliability." There are three techniques to perform newborn screening for Pompe: enzymatic, digital microfluidics, and tandem mass spectrometry (MS/MS). Recent data suggests that the MS/MS technique limits the number of patients with pseudodeficiencies that screen positive. Patients with pseudodeficiency have a lower enzyme activity than the general population, but do not have disease. Specifically, New York state reports that after screening 257,546 patients since October 2014 using MS/MS, only one patient was incidentally found to have a pseudodeficiency.*[13] *The CDPHE lab has been testing for other disorder using MS/MS technology and is very familiar and comfortable with this technology.*

4. *"Cost-benefit consequences of screening are acceptable." Cost per test is estimated to be $7.00 per newborn including follow-up. This cost benefit is comparable to the other disorders currently screened for in Colorado. Although cost of treatment with enzyme replacement therapy is high at $100,000-300,000 per year, there would likely be a net savings since early diagnosis provides opportunity for appropriate disease-specific intervention. Accurate and timely diagnosis is proven to avoid medical complications such as recurrent respiratory illnesses, mechanical ventilation, cardiac failure, and loss of ambulation, thereby eliminating unrelated testing and significantly reducing the number and average length of hospital stays, emergency room care and costly intensive care services.*[14]

After the Board added Pompe disease to the panel, the Department began implementation. At this point, the Department received new information about the costs to add Pompe disease to the panel. The analysis for criterion 1, 2 and 3 is unchanged. The analysis for criterion 4 has changed.

Based upon this new information, the anticipated average cost to add Pompe disease is over $700,000. It is anticipated that costs will range from $667,000 to $733,000 over a five year

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[10] Echaniz-Laguna A, Carlier RY, Laloui K, Carlier P, Salort-Campana E, Pouget J, Laforet P. SHOULD patients with asymptomatic pompe disease be treated? A nationwide study in france. Muscle Nerve. 2015 Jun;51(6):884-9. *See also footnote 7.*

[11] Scott CR, Elliot S, Buroker N, Thomas LI, Keutser J, Glass M, et al. Identification of Infants at Risk for Developing Fabry, Pompe, or Mucopolysaccharidosis-I from Newborn Blood Spots by Tandem Mass Spectrometry. J Pediatr.2013.

[12] *See footnote 4.*

[13] June 22, 2015 email correspondence with Beth Vogel, CGC, scientist for NYS Newborn Screening Program.

[14] *See footnotes 3, 5, 7, and 8.*

period. This is an increase from the Department's original projection of $471,000. The Department anticipates that the cost will be $10-13 per screening rather than $7.00. During the September 2015 rulemaking, the Department indicated that the cost could largely be absorbed by the Newborn Genetics Fund. The increased cost cannot be absorbed by the fund. Including Pompe disease in the newborn screening panel without a fee increase, is not sustainable in the long-term and will jeopardize existing day-to-day operations.

Though the cost has increased the Department continues to recommend that Pompe disease be included on Colorado's newborn screening panel. The Department is requesting that it be added effective July 1, 2019 as this gives the Department the time needed to pursue adjusting the fee, requesting the necessary spending authority, procuring the needed equipment and developing the screening protocols.

Though this rulemaking is specific to the implementation date for Pompe disease, the revenue and expenditures for all activities occurring under the Newborn Genetics Cash fund fluctuate. The Department monitors the funds and adjusts the fee and activities as needed to properly manage costs and execute its responsibilities. Though the cost for adding Pompe disease is $10-13, other variables can increase or reduce newborn screening costs. When assessing newborn screening costs as a whole, there are preliminary indicators that the fee may need to increase by $10-20 to meet the current need; however, the Department wants to exhaust every opportunity to leverage and full utilize current funds before requesting authorization to increase the fee. The Department appreciates increased fee revenue is subject to TABOR restrictions, any additional cash fund revenues generated would result in increased TABOR refunds in subsequent years to the detriment of the State General Fund.

Specific Statutory Authority: These rules are promulgated pursuant to the following statutes:
Sections 25-4-801 through 25-4-804 and 25-4-1004, C.R.S

SUPPLEMENTAL QUESTIONS

Is this rulemaking due to a change in state statute?

_____ Yes, the bill number is _____; rules are ___ authorized ___ required.
__X___ No

Is this rulemaking due to a federal statutory or regulatory change?

_____ Yes
__X___ No

Does this rule incorporate materials by reference?

_____ Yes
___X__ No

Does this rule create or modify fines or fees?

_____ Yes
__X _ No

REGULATORY ANALYSIS
for Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening

1. A description of the classes of persons who will be affected by the proposed rule, including classes that will bear the costs of the proposed rule and classes that will benefit from the proposed rule.

Colorado parents and their medical insurers will benefit from early detection of Pompe disease.

The costs for implementation of screening for Pompe will be borne by the Newborn Screening fund balance and a fee increase to account for the extra expenditure. To support screening of Pompe disease, the fee is projected to increase $10-$13. Birthing facilities will bear the increased cost of the newborn screen.

2. To the extent practicable, a description of the probable quantitative and qualitative impact of the proposed rule, economic or otherwise, upon affected classes of persons.

Quantitative: An estimated three newborns with a significant treatable defect will be discovered every two years allowing for the initiation of appropriate treatment procedures.[15] One out of these three infants identified will have infantile onset Pompe disease. Untreated infants with infantile onset Pompe disease typically die within the first year of life.[16] Late treated infants with infantile onset Pompe disease will be at high risk for death, mechanical ventilation, and significant irreversible muscle damage.[17] Patients on mechanical ventilation have recurrent respiratory infections requiring frequent hospitalizations including admissions to intensive care units. Patients that have severe muscle weakness will require wheelchairs and other expensive adaptive equipment. Patients with poor outcome will also require additional nursing care due to the complexity of their healthcare needs.

Qualitative: Parents of these newborns affected with the infantile form of the disorder will be spared the suffering of their children through severe health issues and the death of their baby within the first year of life. The family will be relieved of the protracted diagnostic and treatment burden associated with difficult to diagnose disorder. Treatment becomes more efficacious with early diagnosis and the child benefits from better outcome. The Colorado Newborn Screening program will be recognized as meeting current standard of care for newborn screening nationally.

3. The probable costs to the agency and to any other agency of the implementation and enforcement of the proposed rule and any anticipated effect on state revenues.

All costs are borne by the Newborn Genetics Fund. A $10-13 fee increase will be needed to occur to add Pompe Disease. If the proposed rule is approved, the Department will seek the necessary authorizations to increase the fee.

4. A comparison of the probable costs and benefits of the proposed rule to the probable costs and benefits of inaction.

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[15] *See footnote 11.*
[16] *See footnote 4.*
[17] *See footnote 8.*

It is estimated that it will cost over $700,000 annually to implement this rule change. The Newborn Genetics fund balance will be used to cover the initial cost of implementation and the Department will seek the necessary authorizations to increase the fee so the fund is sustainable long-term.

5. A determination of whether there are less costly methods or less intrusive methods for achieving the purpose of the proposed rule.

The alternative is to remove Pompe disease from the newborn screening panel. The Department does not recommend this. Screening benefits Colorado families. Though diagnosis and disease-specific interventions are costly, reducing mechanical ventilation costs, avoiding medical complications and unnecessary testing, reducing the number and average length of hospital stays, and reducing the need for emergency and intensive care services are benefits that exceed these costs.

6. Alternative Rules or Alternatives to Rulemaking Considered and Why Rejected.

Screening is authorized via Board of Health rules. Adding Pompe disease to the panel allows for the Department to monitor prevalence. Though supplemental screening through the private sector, it is more costly and parents may not elect to receive supplemental screening. Though the prevalence is not high, it is comparable to other conditions on the panel and there is a sufficient public health interest to merit uniform screening.

7. To the extent practicable, a quantification of the data used in the analysis; the analysis must take into account both short-term and long-term consequences.

Data was analyzed from the two largest cohorts of patients with infantile onset Pompe disease on treatment with a focus on outcome and age of initiation of enzyme replacement therapy. The Department has also considered the new cost projections.

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Long-Term Prognosis of Patients with Infantile-Onset Pompe Disease Diagnosed by Newborn Screening and Treated since Birth

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Objective To determine the benefit of newborn screening for the long-term prognosis of patients with classic infantile-onset Pompe disease (IOPD).

Study design A cohort of patients with classic IOPD were diagnosed by newborn screening, treated with recom- binant human acid a-glucosidase (rhGAA), and followed prospectively. Outcome measurements included survival, left ventricular mass, serum creatinine kinase, motor function, mental development, and systemic manifestations.

Results Ten patients who presented with left ventricular hypertrophy at diagnosis received rhGAA infusions starting at a median age of 16 days (6-34 days). All patients were cross-reactive immunologic material-positive. After a median treatment time of 63 months (range 28-90 months), all could walk independently, and none required me- chanical ventilation. All patients had motor capability sufficient for participating in daily activities, but muscle weak- ness over the pelvic girdle appeared gradually after 2 years of age. Ptosis was present in one-half of the patients, and speech disorders were common. Anti-rhGAA antibody titers were low (median maximal titer value 1:1600, range: undetectable rv 1:12 800).

Conclusion By studying patients treated since birth who have no significant anti-rhGAA antibody interference, this prospective study demonstrates that the efficacy of rhGAA therapy is high and consistent for the treatment of classic IOPD. This study also exposes limitations of rhGAA treatment. The etiology of the manifestations in these early-treated patients will require further study. (J Pediatr 2015;166:985-91).

Data from the Pompe Disease Registry (unpublished date previously provided to the external Condition Review Workgroup as part of SACHDNC process, obtained permission from Genzyme to reproduce for purpose of rule change):[11]

Findings from the Pompe Disease Registry

We requested that Genzyme query the Pompe disease registry to compare survival and ventilator-free survival comparing those who began ERT before 3 months of age to those who began ERT at three months of age and older for those with classic infantile-onset Pompe disease. The results of this request as directly reported appear in the box below. Genzyme provided two analyses. The first includes all subjects in the database, and the second excludes those managed in Taiwan, many of whom would have been detected by newborn screening and have higher rates of ventilator-free survival. The second table in this analysis describes the expected outcomes for clinically detected cases.

Survival Outcomes for Infantile-Onset Pompe Disease by Age at First ERT: Findings from the Pompe Disease Registry

Summary

Patients from the Pompe Registry with symptom onset ≤12 months of age with evidence of cardiomyopathy who received their first treatment with ERT prior to 3 months of age report better survival and invasive ventilator-free survival at 12 months, 24 months, and 36 months of age than patients who received their first treatment with ERT at 3 months of age or older.

Results

Table 3.4. All Patients with Symptom Onset ≤12 months of age with Evidence of Cardiomyopathy

	Age of First Treatment	
	ERT <3 months	ERT ≥3 months
Survival	(Percent Surviving (95% CI))	
	n=36	n=104
12 months	94.1% (78.5, 98.5)	91.3% (84.0, 95.4)
24 months	84.6% (66.8, 93.3)	73.3% (63.3, 81.0)
36 months	80.9% (62.2, 91.0)	63.5% (52.7, 72.5)
Mechanical Ventilation-Free Survival		
	n=24	n=69
12 months	91.3% (69.5, 97.8)	89.8% (79.8, 95.0)
24 months	81.7% (58.2, 92.7)	66.4% (53.1, 76.8)
36 months	76.2% (51.7, 89.4)	56.5% (42.6, 68.2)

Table 3.5. Patients with Symptom Onset ≤12 months of age with Evidence of Cardiomyopathy, Excluding Patients from Taiwan

	Age of First Treatment	
	ERT <3 months	ERT ≥3 months
Survival	(Percent Surviving (95% CI))	
	n=30	n=96
12 months	92.9% (74.3, 98.2)	90.6% (82.7, 95.0)
24 months	81.0% (60.2, 91.7)	72.1% (61.5, 80.3)
36 months	76.5% (54.8, 88.8)	61.3% (49.9, 70.9)
Mechanical Ventilation-Free Survival		
	n=20	n=65
12 months	89.5% (64.1, 97.3)	89.2% (78.6, 94.6)
24 months	77.5% (50.5, 91.0)	65.9% (52.1, 76.7)
36 months	71.1% (43.6, 86.9)	55.3% (40.9, 67.5)

Discussion

The analysis is descriptive in nature; no adjustments have been made for severity of disease or any potential confounding factors that may influence the time of diagnosis, the time of treatment, length of survival or ventilator-free survival, or variables that may influence censoring (i.e. loss to follow-up in the Registry).

The Pompe Registry does not collect information on newborn screening. Because patients from Taiwan may have been identified by newborn screening (and not clinically diagnosed), all patients from Taiwan were excluded from the analysis presented in Table 4.2.

Data are not presented for patients with symptom onset ≤12 months without evidence of cardiomyopathy. No deaths from this population meeting the study criteria were reported to the Registry.

Methods

All treated patients in the Pompe Registry with symptom onset ≤12 months with a record of treatment with ERT were eligible for analyses. Patients were stratified into those with and without evidence of cardiomyopathy; and data for patients with cardiomyopathy were included.

Kaplan-Meier curves were fitted, stratified by those patients with a record of first infusion <3 months of age or ≥3 months of age, for the population with symptom onset ≤12 months and evidence of cardiomyopathy. Events were defined as (1) death, and (2) use of invasive ventilation therapy or death. The time to the event was derived as time from birth. Only patients at risk for the event were included in the Kaplan-Meier analyses.

The 95% confidence intervals (CIs) of event-free survival are reported from the Kaplan-Meier estimation. The CIs are calculated using a transformation of the log (-logS(t)) function; the limits are then transformed back to the survival function.

Patients with a reporting physician from Taiwan were excluded from the second analysis.

STAKEHOLDER COMMENTS
for Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening

The following individuals and/or entities were included in the development of these proposed rules:

Colorado Newborn Screening Advisory Committee

Committee composed of Neonatology; Pediatrics, Community Provider; Pediatrics, Hospital-based Provider; Rural Provider; Hospital-based Nursery RN; Colorado Hospital Association Representative; State Medicaid Designee; Patient, Parent; Parent. Also contacted were organizations with a vested interest in newborn screening such as March of Dimes, AAP and the Inherited Metabolic Diseases Clinic at Children's Hospital Colorado/University of Colorado Denver

The following individuals and/or entities were notified that this rule-making was proposed for consideration by the Board of Health:

Colorado Hospital Association
Hospitals and midwives contacts were notified via CDPHE newborn screening distribution list. List includes hospital laboratory supervisors and managers and labor and delivery nurses, and other pertinent personnel involved in the newborn screening process.
Hospitals and birthing facilities were notified through the CDPHE Health Facilities Portal

Summarize Major Factual and Policy Issues Encountered and the Stakeholder Feedback Received. If there is a lack of consensus regarding the proposed rule, please also identify the Department's efforts to address stakeholder feedback or why the Department was unable to accommodate the request.

There are no issues at this time. However we will be working with the community and we anticipate that some may question or oppose the fee. The Department is committed to receiving that feedback, educating the community and determining, whether the community as a whole, continues to recommend Pompe disease for the NBS panel.

Please identify health equity and environmental justice (HEEJ) impacts. Does this proposal impact Coloradoans equally or equitably? Does this proposal provide an opportunity to advance HEEJ? Are there other factors that influenced these rules?

Mandated newborn screening for Pompe disease reduces health care disparities by ensuring that newborns receive equal access to timely diagnosis and treatment regardless of race, ethnicity, socioeconomic status, and geography. The current model the Department uses is for all patients screened to bear the cost of those few that will be found. This model is utilized for all conditions on the newborn screening panel.

Department of Public Health and Environment

NEWBORN SCREENING AND SECOND NEWBORN SCREENING

5 CCR 1005-4

NEWBORN SCREENING REGULATIONS

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1.6 List of Conditions for Newborn Screening

1.6.1 The Laboratory shall conduct screening tests for the following conditions:

1.6.1.1	Phenylketonuria
1.6.1.2	Congenital Hypothyroidism
1.6.1.3	Hemoglobinopathies
1.6.1.4	Galactosemia
1.6.1.5	Cystic Fibrosis
1.6.1.6	Biotinidase Deficiency
1.6.1.7	Congenital Adrenal Hyperplasia
1.6.1.8	Medium Chain Acyl-CoA dehydrogenase deficiency
1.6.1.9	Very Long Chain Acyl-CoA dehydrogenase deficiency
1.6.1.10	Long-Chain L-3-Hydroxy Acyl-CoA dehydrogenase deficiency
1.6.1.11	Trifunctional protein deficiency
1.6.1.12	Carnitine Acyl-carnitine translocase deficiency
1.6.1.13	Short Chain Acyl-CoA dehydrogenase deficiency
1.6.1.14	Carnitine palmitoyltransferase II deficiency
1.6.1.15	Glutaric acidemia Type 2
1.6.1.16	Arginosuccinic acidemia
1.6.1.17	Citrullinemia
1.6.1.18	Tyrosinemia
1.6.1.19	Hypermethionemia
1.6.1.20	Maple Syrup urine disease
1.6.1.21	Homocystinuria
1.6.1.22	Isovaleric acidemia

1.6.1.23	Glutaric acidemia Type 1
1.6.1.24	3-hydroxy-3-methylglutaryl-CoA Lyase deficiency
1.6.1.25	Multiple Carboxylase deficiency
1.6.1.26	3-methylcrotonyl-CoA carboxylase deficiency
1.6.1.27	3-methylglutaconic aciduria
1.6.1.28	Methylmalonic acidemias
1.6.1.29	Propionic acidemia
1.6.1.30	beta-Ketothiolase deficiency
1.6.1.31	Carnitine uptake defect
1.6.1.32	Arginase deficiency
1.6.1.33	Malonic acidemia
1.6.1.34	Carnitine palmitoyltransferase deficiency 1A
1.6.1.35	Severe Combined Immunodeficiency
1.6.1.36	BEGINNING JULY 1, 2019, Pompe Disease

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