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Transmitted via email to taguilar@pa.gov

Re: Rare Disease Advisory Council Consideration of Newborn Screening Policy

Dear Members of the Pennsylvania Rare Disease Advisory Council:

This letter is written as a follow-up to my discussions with individual members of the Rare Disease Advisory Council regarding its examination of Pennsylvania's newborn screening (NBS) program. In these discussions, it was brought to my attention that the Council is seeking to evaluate the State's implementation of NBS based in part on NORD's 2016/17 State Policy Report Card. In order to support the Council's work on this matter, I wish to clarify the scope of NORD's State Report regarding NBS and how we intend to evaluate Pennsylvania and other states moving forward.

NORD's 2016/17 State Report Card (accessible here: http://rareaction.org/resources-for-advocates/state-progress-report/) evaluated state NBS programs solely on their adoption of the Federal Recommended Uniform Newborn Screening Panel (RUSP) as developed by the Advisory Committee on Heritable Disorders in Newborns and Children. Specifically, we examined whether states mandate screening for RUSP Core and Secondary Conditions for all populations. Our analysis of Pennsylvania's NBS program found that while it mandates screening for nearly all Core Conditions (earning an "A" grade for this category of NORD's report), it only mandates screening for many Secondary Conditions for certain populations (earning a "D" grade for this category of NORD's report). By contrast, several other states we evaluated mandate the detection of the RUSP Secondary Conditions for all populations.

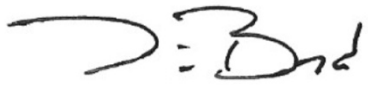
Based on this evaluation, NORD continues to advocate for Pennsylvania's program to mandate the detection of applicable Secondary Conditions for all populations. However, it is not our intention to say that Pennsylvania should model itself on other NBS programs beyond the list of screened conditions. To that end, we recognize that the strength of any state's NBS program is not limited to the number of conditions detected. For example, state government support for its NBS labs (including funding for personnel and new tests) is critically important, as is the process by which Pennsylvania adds new conditions to its program. These issues can distinguish states

from others above and beyond whether specific conditions are on its NBS panel (and whether every condition is mandated for all populations).

NORD intends to expand its NBS analysis in its forthcoming 2018 Report Card to better capture these issues and compare state programs. We also intend to better contextualize the list of conditions on state NBS panels, such as to make it clearer that RUSP Secondary Conditions can only be detected as part of the tests identifying certain Core Conditions. With these changes in mind, our initial analysis for 2018 suggests that Pennsylvania's program is structured quite well compared to other states and in certain respects should be a model for others to follow. NORD will still be advocating for all states to expand their NBS programs to adhere to the RUSP recommendations (and go beyond the RUSP when the scientific evidence supports doing so), but we hope that the expansion of our analysis will better inform the rare disease community about screening policies in their state.

Thank you for your time and consideration of this letter. I hope this helps clarify NORD's analysis of NBS and strengthen the Council's work on this issue. If I can be of further assistance, please do not hesitate to contact me.

Sincerely,



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