A proposal to efficiently improve diagnostic clarity, therapeutic and clinical referrals, disease and therapeutic understanding, and quality of life in the newborn screening ecosystem while reducing cost and overhead

Dean Suhr, MLD Foundation

WorldSymposium 2022, Poster #296

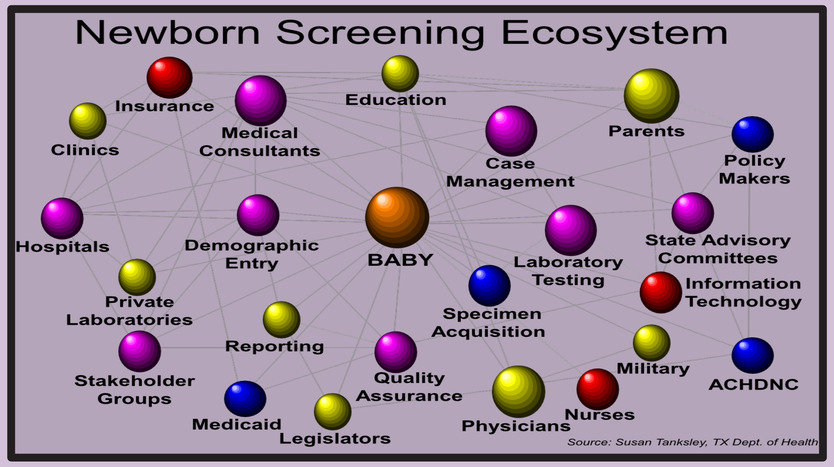
Newborn Screening

"Newborn Screening is one of the country's greatest public-health successes, but the process of adding new conditions to the program happens too slowly to keep pace with an anticipated influx of cell- and gene-based therapies — at least 60 in the next decade."[1] – Don Bailey

Newborn screening is key to the timely and efficient diagnosis of many progressive disorders, including many lysosomal diseases, and enables families to have the broadest and best therapeutic and clinical choices pre-symptomatically when therapies are most effective. The multi-stakeholder newborn screening ecosystem is not optimally efficient at providing accurate, comprehensive, and timely information to parents and their infant's treating physicians to inform decision making as parents seek to optimize the baby's Quality of Life (QoL).

Existing NBS Ecosystem

The existing ecosystem is very complex with highly qualified and interested participants each having broad interests but only responsible for a small part of the process. Further, each party has differing constraints, limitations, goals, and measures of success. This often results in reduced and slower information exchange and communication than is optimal to optimize the QoL of the newborn.



What If The Baby's QoL Was The Focus?

The end goal is not to improve NBS, or to get a therapy developed and approved – it's to improve the baby's QoL. Of course, the NBS, clinical, and therapeutic "tools" are all key to improving QoL.

Better communication, data, feedback, learning, and continual improvement *from the baby's ecosystem* are key elements to improving QoL. None of these elements are static … they need to continue and improve over time, both for the benefit of that baby and also for the benefit of babies to follow with that same disease.

The diagram to the right shows the traditional data and communication flow in black, with the places that need improvement in red. And also note the added key ecosystem partner, "Domain Experts". Generally this is patient advocacy who is unique to have an effective direct line of trusted bi-directional communication with the families, a very thorough understanding of QoL factors, and strong clinician, specialist, biopharma, and policy connections.

And Now We've Crossed the Line

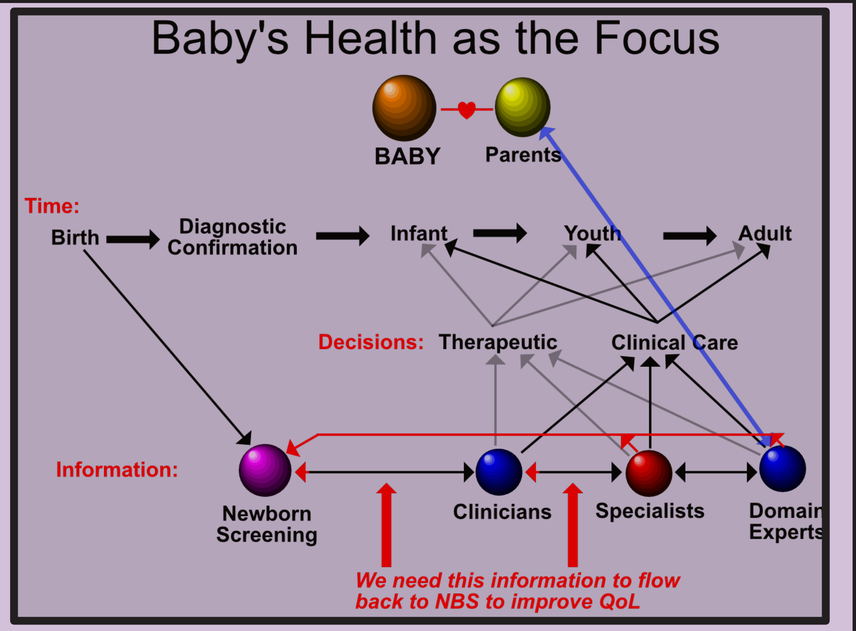
In order to share data to improve learning that leads to improved baby and societal QoL, we need consent, funding, trust, participation, and a willingness to share. Our current system marches on in time with limited feedback. A better system is always observing, learning, and sharing to help those earlier on the diagnostic, clinical and therapeutic journey.

But what about consent, privacy, ethics, and choice? Many see these as unchangeable walls made of steel and concrete. We believe instead that they are made of brick – solid and firm, but each brick can be adjusted and re-mortared while still retaining the structural integrity.

Getting There – From the Inside or Externally?

"Don't think outside the box, think as if there was no box" -Amazon

Forty experts published a paper *Expert Evaluation of Strategies to Modernize Newborn Screening in the United States*[2] with a lot of support for ideas but high uncertainty on how to get there. Slightly more than half (22 [55%]) preferred to retain the current system and make all changes within it, while developing a small number of new components. Slightly less than half (18 [45%]) felt that more substantial changes were needed, developing many new components or an entirely new system. We can't stay on the fence, we must address the concerns and get moving. Lack of timely resolution leans us, like many of the above experts, towards an external solution.



Long term follow-up (engaging clinicians and consented families in the process), improving diagnostic-driven decision making (polygenic risk scores, biomarkers), leveraging Domain Experts to improve QoL, capturing and managing the data … these issues need to be addressed now.

1. Dattaro, L. Reimagining the newborn screening system: Q&A with Don Bailey (Jan. 2022) https://doi.org/10.53053/LRNS7670

2. Bailey DB, Porter KA, Andrews SM, Raspa M, Gwaltney AY, Peay HL. Expert Evaluation of Strategies to Modernize Newborn Screening in the United States. JAMA Netw Open. 2021;4(12): e2140998. doi:10.1001/jamanetworkopen.2021.40998) https://jamanetwork.com/journals/jamanetworkopen/fullarticle/2787589