A novel visualization of state-by-state variability in newborn screening (NBS) highlights underrepresentation of lysosomal storage disorders

Daniel G. Piqué, M.D., Ph.D., Erica Wright, M.S., CGC, & Shawn E. McCandless, M.D.

Contact info | Website/mailing list at dcape.rbind.io/about Email: dcape1@gmail.com

Introduction

- NBS is a program run by each state that detects a variety of treatable infantile-onset metabolic and genetic disorders
- The conditions screened for on NBS vary state-by-state, though the extent of variation has not previously been assessed visually.
- The RUSP is a list of 35 minimum recommended screening tests for states to include on NBS
- Most LSDs listed have enzyme replacement therapies available; other disorders are treated with diet, nutrient supplementation, & by avoiding fasting.

Methods

Data on newborns screening offerings by state was manually downloaded from newsteps.org in December 2021. State-level data was downloaded from Wikipedia using the rvst API (version 0.3.6) in December 2021. Visualization created using the R programming language (version 4.0.3).

Conclusions

- Listing of AA, FAO, and OA disorders is highly variable between states, despite the use of standard MS/MS biomarkers (AA, ACP) that could flag these disorders
- Reporting of NBS offerings by analyte or assay type could provide a more consistent state-by-state comparison
- Could be used to help advocate for expanding NBS options, particularly between neighboring states in regions (e.g. the South) with wide discrepancies in NBS offerings

There are 32 conditions screened for in all 50 states

Tennessee, one of 3 states offering the most comprehensive NBS, screens for twice as many conditions as Arkansas, a geographic neighbor.

In general, states in the Northeast offer more NBS options. Most have adopted RUSP guidelines for LSDs.

States in the South have the greatest variability in NBS offerings, including LSDs.

States in the West with smaller populations offer fewer NBS options, and most have not adopted RUSP guidelines for LSDs.

Information Management Systems (IMS)

- Internally Developed
- StarLims
- PerkinElmer
- Neometrics
- Natus
- StarLims
- Internally Developed

Political affiliation

There's no clear relationship between the political leaning of each state and amount of LSD screening offered.