An Online Software Suite to Improve Accuracy of Newborn Screening
What is Newborn Screening (NBS)

- One of the most successful public health programs to detect certain genetic, endocrine, hemoglobinopathy, immunology, and metabolic conditions shortly after birth
- 20,000 babies identified each year in the United States (~out of 4M life births)
- Blood spot screening

Limitations
- One size fits all - same analytics for each baby
- Only a few out of all screening markers are analyzed
- Only a few metabolites used for each disorder
- Thousands of false alarms - high false positive rate
- Slow Turnaround time of screen-positive cases
Why RUSPtools

- Efficacy: Provide better and more accurate tools to Health care providers
- Democratize access to health care data and information
- Speed up screening process and diagnostic workup
- Reduce burden on families by reducing false alarms
- Reduce cost and turnaround time of screening
What is the RUSPtools

- Platform (multi-product) Software Suite and mobile app for rapid and convenient access to NBS knowledgebase to analyze and interpret an individual’s and population-level newborn screening data.
Functions of Software Suite

01

Influence of clinical variables on baby’s metabolic level

• Birth weight, gestational age, ethnicity
Functions of Software Suite

01
Influence of clinical variables on baby’s metabolic level
- Birth weight, gestational age, ethnicity

02
Use data mining (AI) to improve NBS accuracy by integrating all metabolic data collected at birth and clinical variables
## Beta Version of Software Suite

<table>
<thead>
<tr>
<th>Feature</th>
<th>RUSPtools</th>
<th>CLIR*</th>
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<tbody>
<tr>
<td>Online</td>
<td>✔</td>
<td>✔</td>
</tr>
<tr>
<td>Easy to use</td>
<td>✔</td>
<td>✗</td>
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<tr>
<td>Multiple functions</td>
<td>✔</td>
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<tr>
<td>Collecting private data</td>
<td>✔</td>
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*academic competitor

[https://rusptools.shinyapps.io/RandomForest/](https://rusptools.shinyapps.io/RandomForest/)
[https://rusptools.shinyapps.io/MetaDB/](https://rusptools.shinyapps.io/MetaDB/)
Market and Making Money

- Service to State Health Departments, CDC, Health care providers (Hospital and Clinics)
- Companies providing NBS technology for State laboratories (f.e., Perkin Elmer Genetics)
- Private companies providing supplemental screening (Mayo, Sema4)
- International NBS stakeholders
  - China: only 2-5 metabolic disorders are currently screened, private NBS program, rapidly expanding
  - India: No universal screening, private NBS program
- Free for academic use, license for commercial use
- Potential for DTC service
Accomplishments with Blavatnik Funding?

• Develop more functions in the online software suite

• Find more collaborators within US and across the world.