SCID Newborn Screening: Building the Clinical Network

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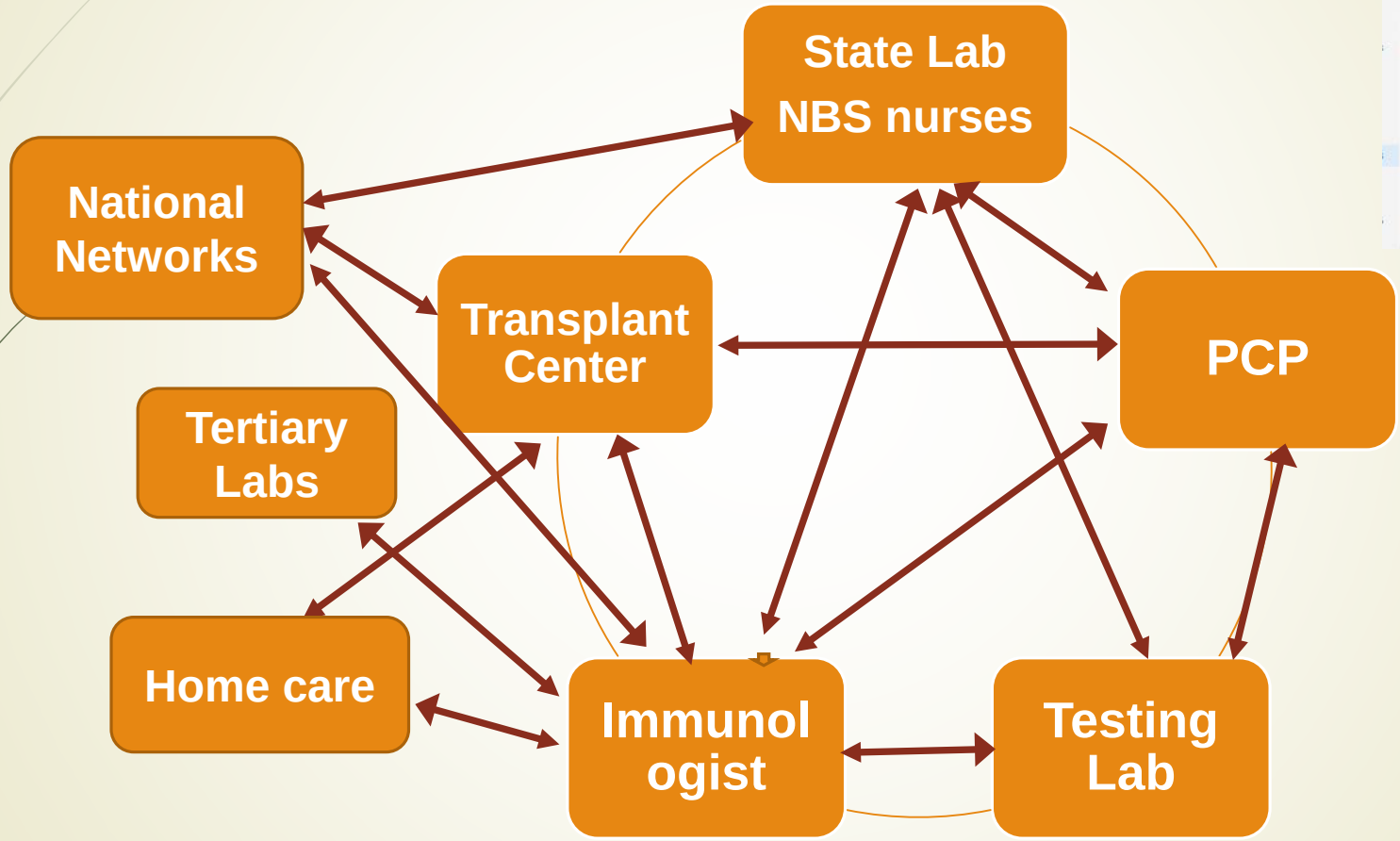
Disclosures

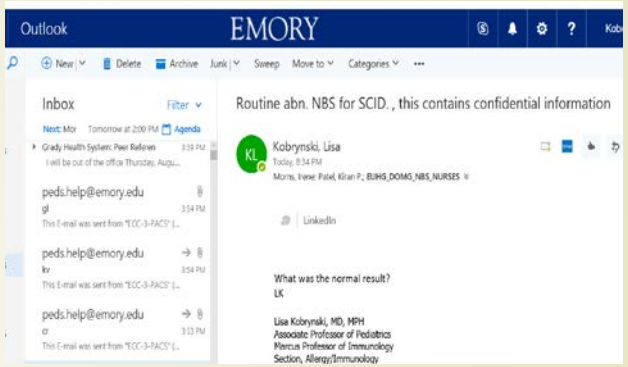
- Sponsored research for Baxalta-Shire
- Advisory Board CSL Behring

Clinical Network Members

- State Screening Lab
- Newborn screening nurses
- Primary care physician
- Immunologist/hematologist/infectious diseases specialists affiliated with the screening program
- Local or referral laboratory
- Hospital referral center
- Treating physicians for transplant or other care
- Home care nursing
- National Networks

Good Communication is Key





Clinical Network Members

- State laboratory and screening nurses: performing testing and reporting results – clinicians need to be included in the results reporting
  - Can Clinicians access NBS results directly
  - What is the timing for NBS nurses to communicate with clinicians to determine need for follow up testing and for clinician to interpret test results
  - How does the clinician determine need for follow up visits and how is this handled? Do they communicate this with PCP and patient directly?
  - Are the appropriate laboratories for performing confirmatory testing local or a referral laboratory?
  - Is the clinician for the SCID NBS team already familiar with diagnosing and treating patients with primary immunodeficiencies?
  - Is there a BMT team familiar with transplant in primary immunodeficiencies at their center?

Clinical Network Members

- Issues:
  - Unfamiliarity with TREC RT-PCR and interpretation of results
  - Unfamiliar with algorithms for testing and evaluation of patient: delay in initiating testing and evaluation
  - Lack of resources for specialized testing at local medical center (e.g. oncology labs performing lymphoma/leukemia flow cytometry, lack of availability of flow for CD45Ra/Ro testing, labs requiring large amounts of blood)
  - How to arrange for patient to go out of state for diagnosis or treatment?
  - Insurance refusal to cover costs of confirmatory testing – no case manager/social worker

Clinical Network Members

- Do you have the resources for specialized diagnostic testing and starting therapy?
  - Specialized immunological testing: may be local labs or referral laboratories (in or out of state)
  - Genetic testing – SCID panels, microarray for 22q11 deletion, Jacobsen syndrome, sequencing of CHD7 when appropriate – requires clinical expertise and laboratory support
    - If local genetic testing is not availaible, referral genetic labs (e.g. genedx, fulgent, ARUP, Mayo or other medical centers)
  - Is there a case manager or nurse to help with insurance approval for specialized testing
  - For patients that are far away from a medical center or live out of state – they may need additional assistance

Clinical Network

- Issues:
  - Overcoming barriers to insurance coverage for specialized tests
  - Identification of labs capable of performing specialized tests (and arranging to get samples to them)
  - Assistance for families who have to travel long distances for evaluation and treatment

Clinical Network

- Newly diagnosed patients -
  - How to arrange for careful follow up and coordination with PCP and local doctors
  - Working with the transplant team – discuss treatment, timing of treatment, if needed coordinate HLA typing, BMT team usually identifies potential donors, and monitor labs as needed for pre-hematopoietic cell transplant
  - Work with home care companies for infants on gamma globulin replacement and for any other therapies (e.g. G-tube feeding)
  - Work with the treating institution regarding admission, approval for treatment
  - Communicate disposition of baby to NBS program and PCP

Clinical Network

- Through treatment
  - Continued communication between transplant team, immunologist and PCP
  - After discharge transplant team is primarily responsible, but at some point immunologist takes over the care
  - Involvement in reporting long term follow up to national networks such as Primary Immune Deficiency Treatment Consortium (for BMT), USIDNet (for all cases of T cell lymphopenia), maybe others in future (e.g. regional networks, NBSTRN)

Examples

- State A: State Lab, NBS team identified a clinical immunologist at a single site, this site also serves as the referral center for treatment for the entire state – “one stop shop”
- State B: There are one or two State Labs performing screening within the state, each lab team identifies one or more clinicians to review abnormal results, these clinicians may refer patients to one or more hospitals for diagnosis and treatment – “shopping around”
- State C: Their NBS testing is done by another state, they usually identify a clinician to assist in reviewing abnormal screening and ordering confirmatory testing, once identified babies are usually referred to regional centers with expertise in SCID for diagnosis and treatment – “team work”

National Networks

- Purpose is to gather data on infants identified with SCID as newborns
  - Epidemiology: genetic types, race, gender, age at diagnosis, family history, etc….
  - Treatment outcomes: age at transplant, type of transplant, complications, disability, immune reconstitution and survival
  - Publication of data – advancing our collective knowledge on best practices for management of SCID and other T cell lymphopenias, and potentially assisting programs in refinement of their screening
- Examples
  - PIDTC – gathers data on infants receiving HCT for primary immune deficiencies, consortium of multiple centers nationwide
  - USIDNET – database of all PI patients reported by physicians and patients, gathers data on disease, and some outcomes
- Limitations
  - Voluntary reporting, does not capture all cases nationwide
  - USIDNET does not require regular updating of information

Summary

- Building a strong clinical network requires multiple partners
- Each partner needs to be fully involved in the process
- Communication between partners is key!
- Don’t hesitate to call upon outside resources – other labs, clinicians, APHL, CDC, NBSTRN and others…..

TEAM WORK!