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APHL

In Our Own Time: Newborn Screening for Pompe Disease

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June 22, 2017

dsworth
ter

45 2.0K 6.3K

Meyer Aviles @meyeravi · 1d
Happens to the best of us

NHP Southern Command @NHPSouthernComm
If the person who drove their car onto a rock leavi
this morning wants their car they can come talk to
Troopers to get it back!!!

10

NHP Southern Command
@NHPSouthernComm

Follow

If the person who drove their car onto a rock leaving #edc this morning wants their car they can come talk to NHP Troopers to get it back!!!

Nevada Highway Patrol

Retweets 20,606 Likes 50,665


BALLSTON SPA
CCP 7893
SCOTTIES

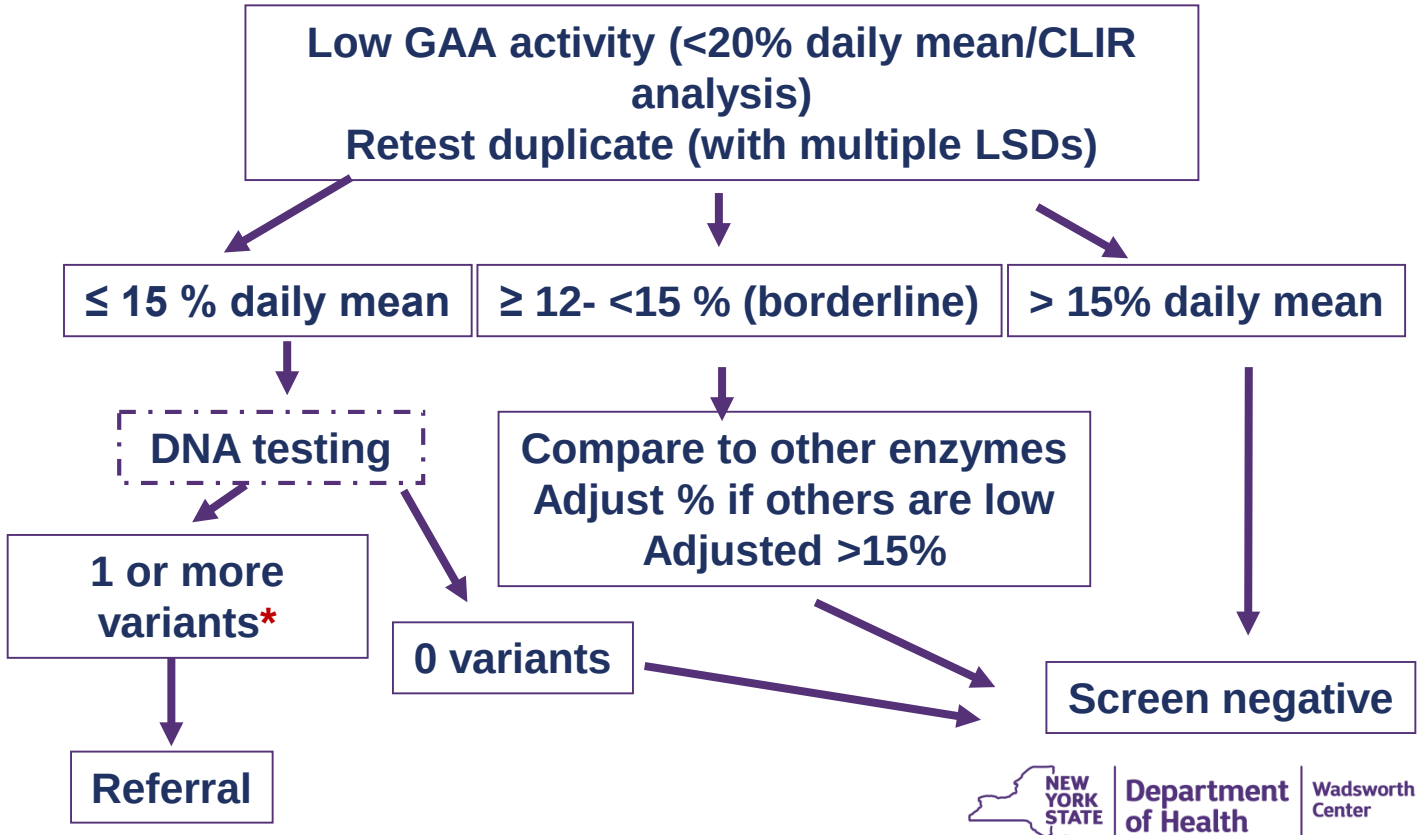
Why Pompe Disease?

- Pilot study – consented 3-4 NYC hospitals
- Dr. Melissa Wasserstein, PI – NICHD
- MC recipient of contract from NICHD
- ACHDNC
- Universal screening in NY
- Substituted MPS1
- Added to regulation (when validated) along with ALD

Pompe Disease

- Conservatively set cut-off at 15.0% for NIH pilot study after >5,000 tests
- MO provided screen positive specimens for interlaboratory comparison
- 12.0% was highest positive result on panel
- Much discussion on case definition for Pompe disease
- Issue with many LSDs having later onset
- Newborn screening???

Pompe Disease Screening Algorithm



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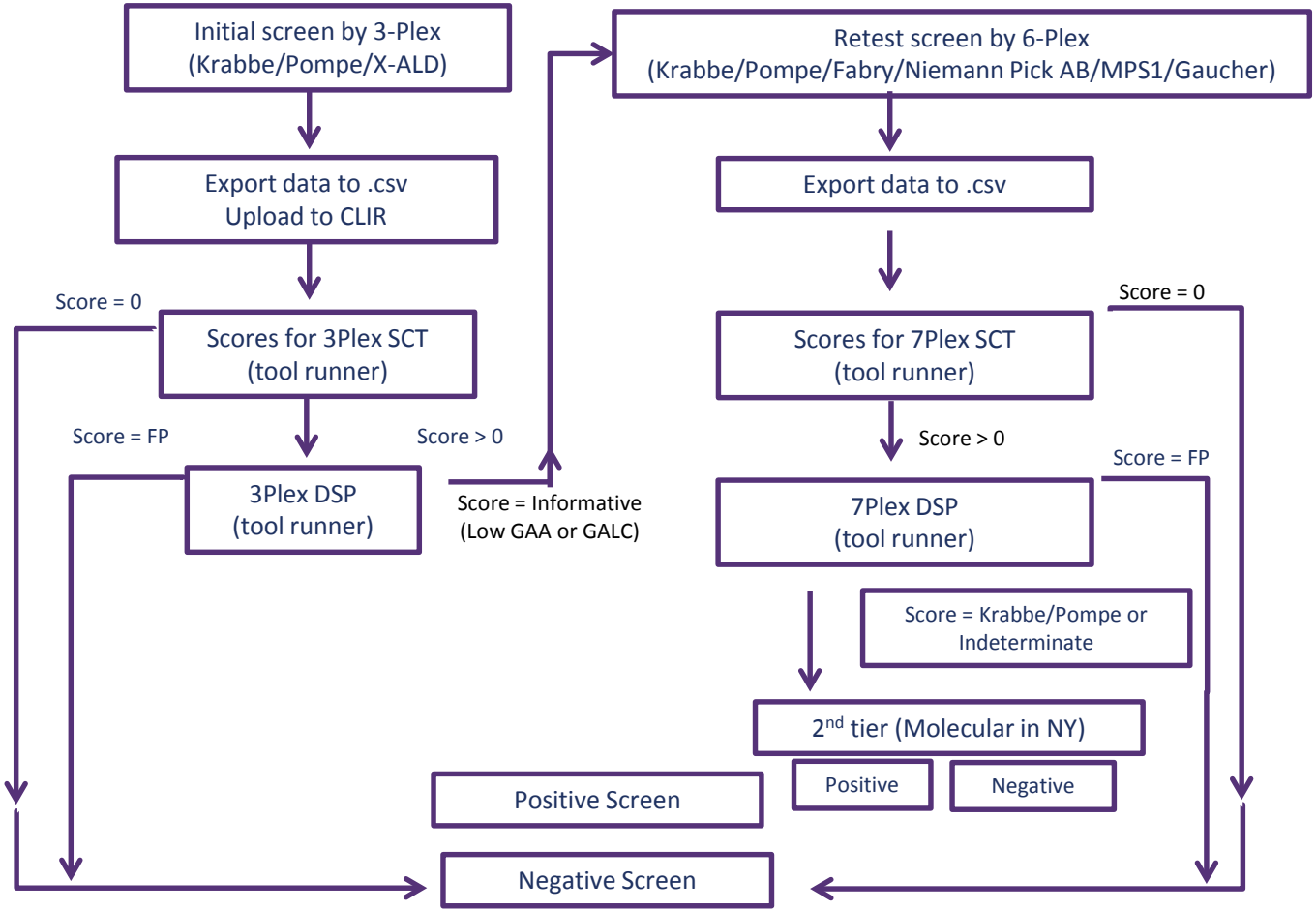
Initial screen by 3-Plex
(Krabbe/Pompe/X-ALD)
Export data to .csv
Upload to CLIR
Score = 0
Scores for 3Plex SCT
(tool runner)
Score = FP
Score > 0
3Plex DSP
(tool runner)
Score = Informative
(Low GAA or GALC)
Retest screen by 6-Plex
(Krabbe/Pompe/Fabry/Niemann Pick AB/MPS1/Gaucher)
Export data to .csv
Score = 0
Scores for 7Plex SCT
(tool runner)
Score > 0
Score = FP
7Plex DSP
(tool runner)
Score = Krabbe/Pompe or
Indeterminate
2nd tier (Molecular in NY)
Positive
Negative
Positive Screen
Negative Screen

MAYO CLINIC

CLIR - Collaborative Laboratory Integrated Reports

Application: KMPX

monica.martin@health.ny.gov

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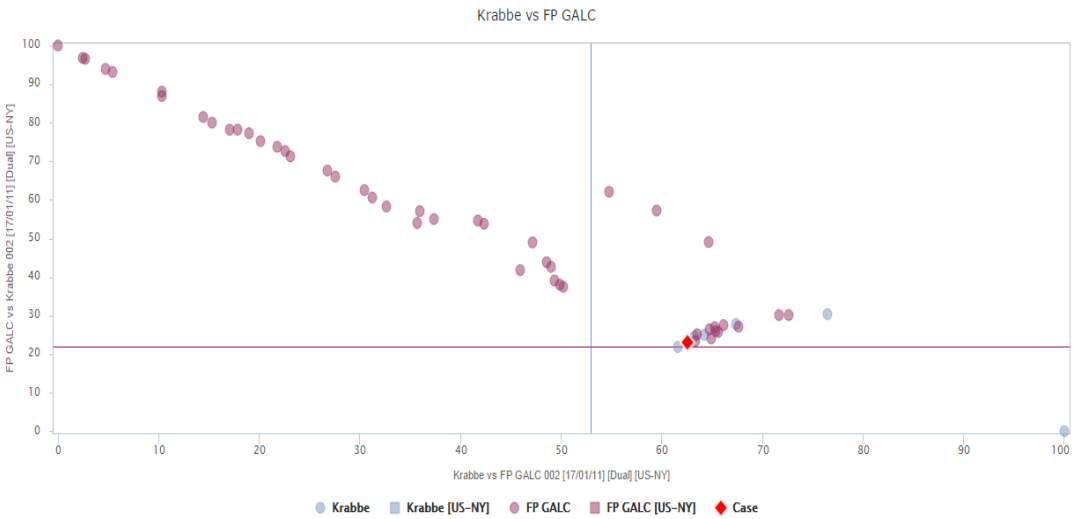
Dual Scatter Plot Runner: Krabbe vs FP GALC 002 [17/01/11] [Scatter] [US-NY]

Dual Scatter Runner | Results | Marker/Covariate Values | Score Calculation | View PDF...

- Krabbe [0]
- FP GALC [5]
  - 20171141147
  - 20171141213
  - 20171141334
  - 20171141456
  - 20171142487
- Indeterminate [1]
  - 20171141929
- Neither [0]

FP GALC	Krabbe
Score: 23 %ile Rank: 0.00 % Count: 47	Score: 63 %ile Rank: 11.36 % Count: 6



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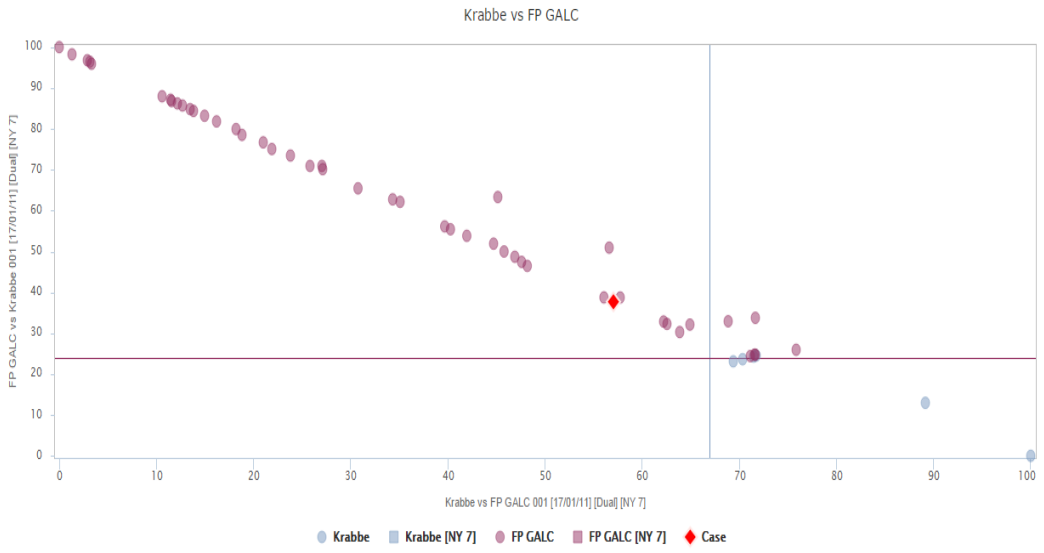
Dual Scatter Plot Runner: Krabbe vs FP GALC 001 [17/01/11] [Scatter] [NY 7]

Dual Scatter Runner | Results | Marker/Covariate Values | Score Calculation | View PDF...

- Krabbe [0]
- FP GALC [1]
  - 20171141929
- Indeterminate [0]
- Neither [0]

FP GALC	Krabbe
Score: 38 %ile Rank: 21.28 % Count: 47	Score: 57 %ile Rank: 0.00 % Count: 6



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Pompe Disease by the Numbers

Validation study 5,000 spring 2013
Pilot 30,358 May 2013 – Sept 2014
Live 701,332 since October 1, 2014
815 <20%

Samples from Missouri – all infantile <8%
Of all affected specimens confirmed 12-13%
Set cutoff at ≤ 15% of mean

Pompe Disease by the Numbers

Live 701,332 October 1, 2014
131 specimens sequenced
10 common benign variants
19 pseudodeficiency / common benign

102	infants referred
52	carriers
4	infantile onset – 3 classical / 1 non-classical
17	2 pathogenic variants
20	1 pathogenic / 1 VOUS
9	2 VOUS

MPS1 Disease by the Numbers

Added to pilot May 20, 2015
Total tested: 31,235
43 <20%; 31 <16%
10 positive infants referred for DNA
5 were pseudodeficiency
1 carrier
1 pseudodeficiency allele / VOUS
3 are being followed clinically/pending

Thank You !!

Joe Orsini, PhD
Erin Hughes, MLS
Patrick Hopkins
Colleen Stevens, PhD

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