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Virtual Roundtable on States' Experiences Screening for Spinal Muscular Atrophy (SMA)

Maryland SCID/SMA Multiplex Assay

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Maryland Department of Health Newborn Screening Division

10/24/19

Screening Method

- Method developed at CDC
  - PCR performed with 2-mm DBS punch in PCR plate
  - TREC and SMN1 (exon 7) multiple assay
  - RNaseP (RPP30) is used as a reference gene
  - AriaMX real-time PCR instrument
  - Biomek NXp is used to process the plates
- Analytical Considerations
  - No increase in retest rate with the addition of SMA
  - Most specimens that are inconclusive for SMA are inconclusive for SCID

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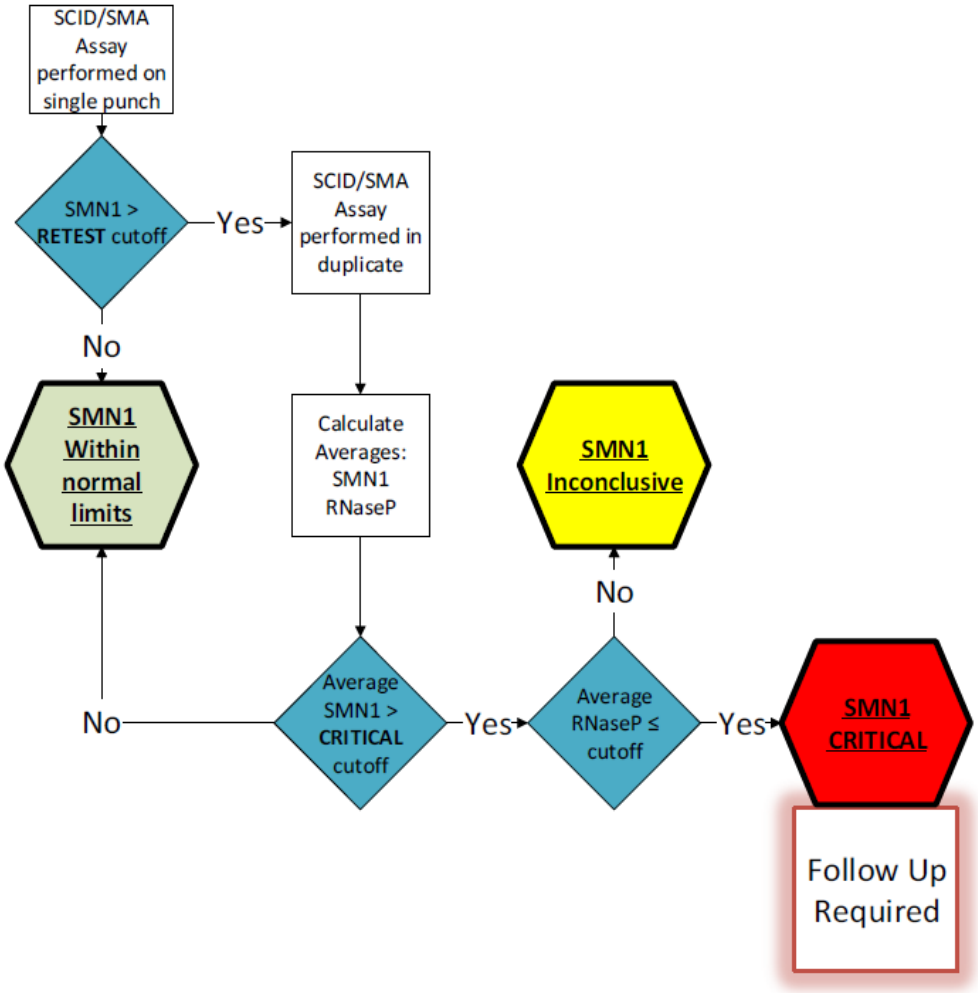
Screening Algorithm



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SMA Screening Results

- Started Screening May 31st, 2019
- Screened over 57,894 specimens
  - Screened over 31,914 infants
- 3 cases sent to follow-up
  - All 3 cases had no amplification of SMN1 exon 7 in the initial and repeat specimen
  - Case 1: SMA type 1 and was treated with Zolgensma
  - Case 2: Diagnosed with SMA, but currently presymptomic
  - Case 3 is pending
- Screen-positive infants are seen at John Hopkins Hospital, Pediatric Neurology
- No known false negatives

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SMA Screening in Minnesota

Carrie Wolf

October 24, 2019

SMA in Minnesota

Year (Screening began 3/1/18)	Newborns Screened	SMA Positive Screen	SMA Confirmed
2018 (3/1/18 – 12/31/18)	55,871	8	8
2019 (through 10/1/19)	49,597	2	2
Total	105,468	10	10

SMA birth prevalence in Minnesota = 1:10,547

SMA Case Summary:

-SMN1: 0 copies (n=10)

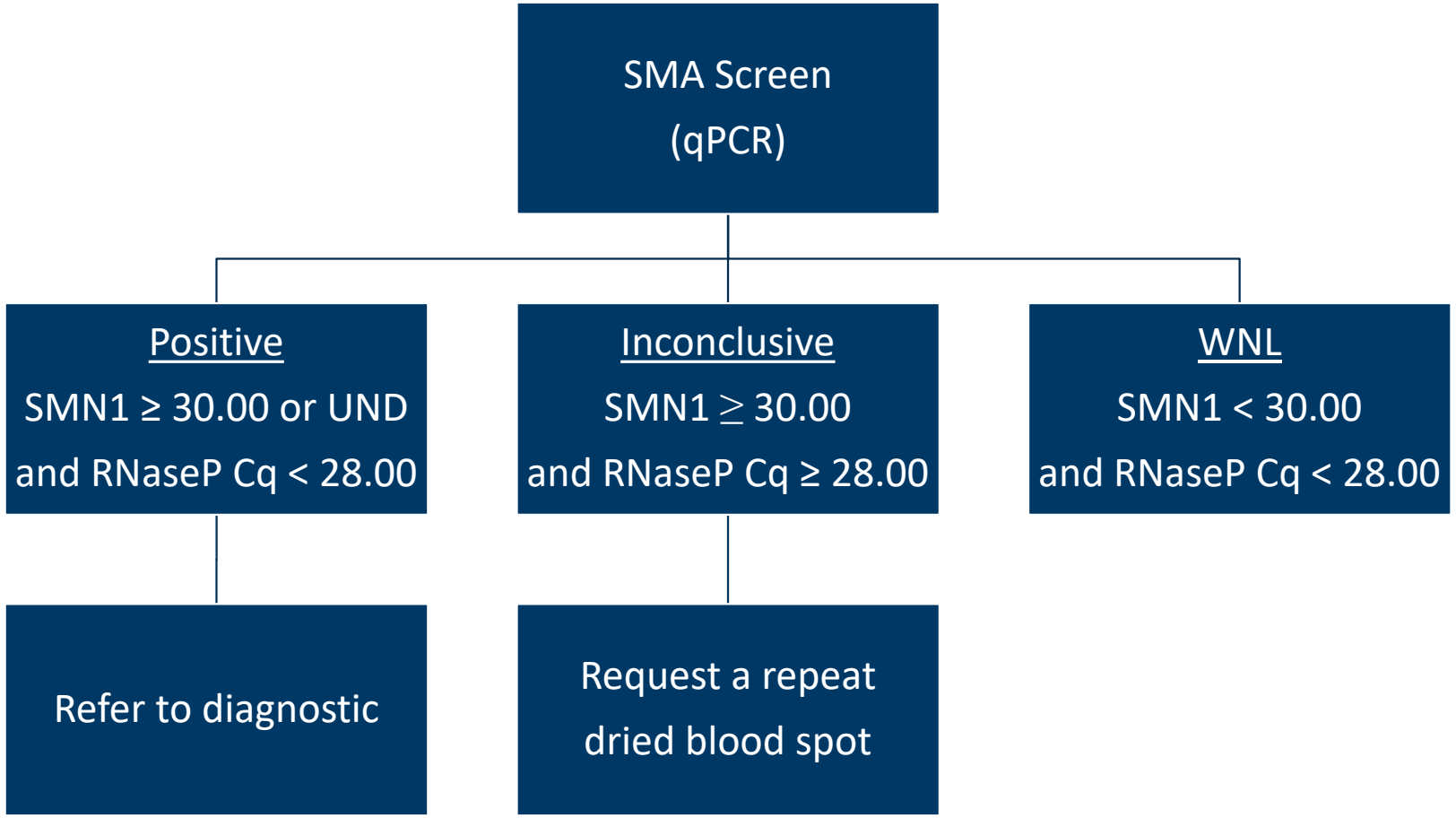
-SMN2:

- 2 copies (n=5)
- 3 copies (n=3)
- 4 copies (n=2)

All 10 have started treatment with Spinraza with a couple also receiving Zolgensma.

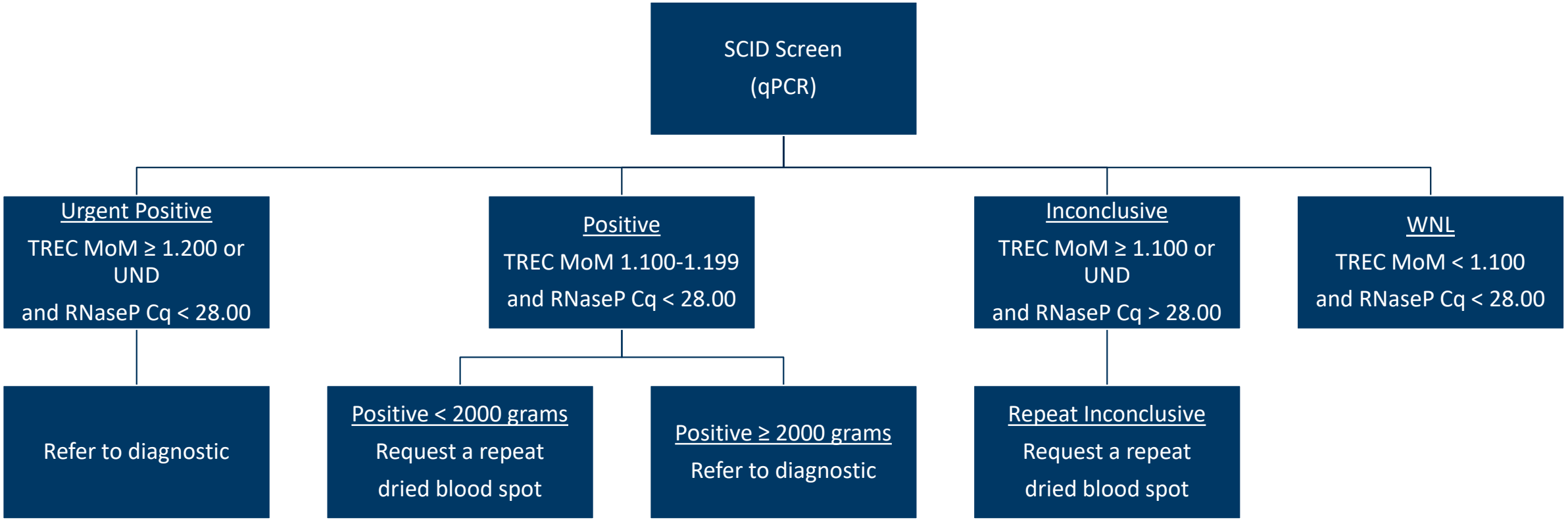
- Median time from birth to first injection = 19 days (range, 11 – 64 days)

SMA Algorithm



https://www.health.state.mn.us/people/newbornscreening/materials/factsheets/bloodspotdisorders.html

SCID Algorithm



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