

Physician Attitudes Toward Newborn Screening for Muscular Dystrophy and Spinal Muscular Atrophy

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NEWBORN SCREENING PROTOCOL FOR DMD/BMD/LGMD

- 1st tier: CK test of blood spots
 - >500 IU/L = retest blood spot
 - Isozyme analysis (Rule Out CK-BB)
 - DNA for deletion analysis
- 2nd tier: Repeat whole blood for CK
 - Next Generation Sequencing (40+ genes)
 - Referral to physician/MDA clinic
 - Biopsy for dystrophin analysis

PENNSYLVANIA EXPERIENCE WITH NEWBORN DMD/BMD SCREENING (July 1986 – December 1995)

• West Penn Hospital	182,411	1
• Magee-Womens Hospital	221,165	16
• MDA Grant (PA, TX, NY, PR)	194,439	19
• Brazil	25,566	1
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Total	623,581	59

Frequency in males: 1/5,334

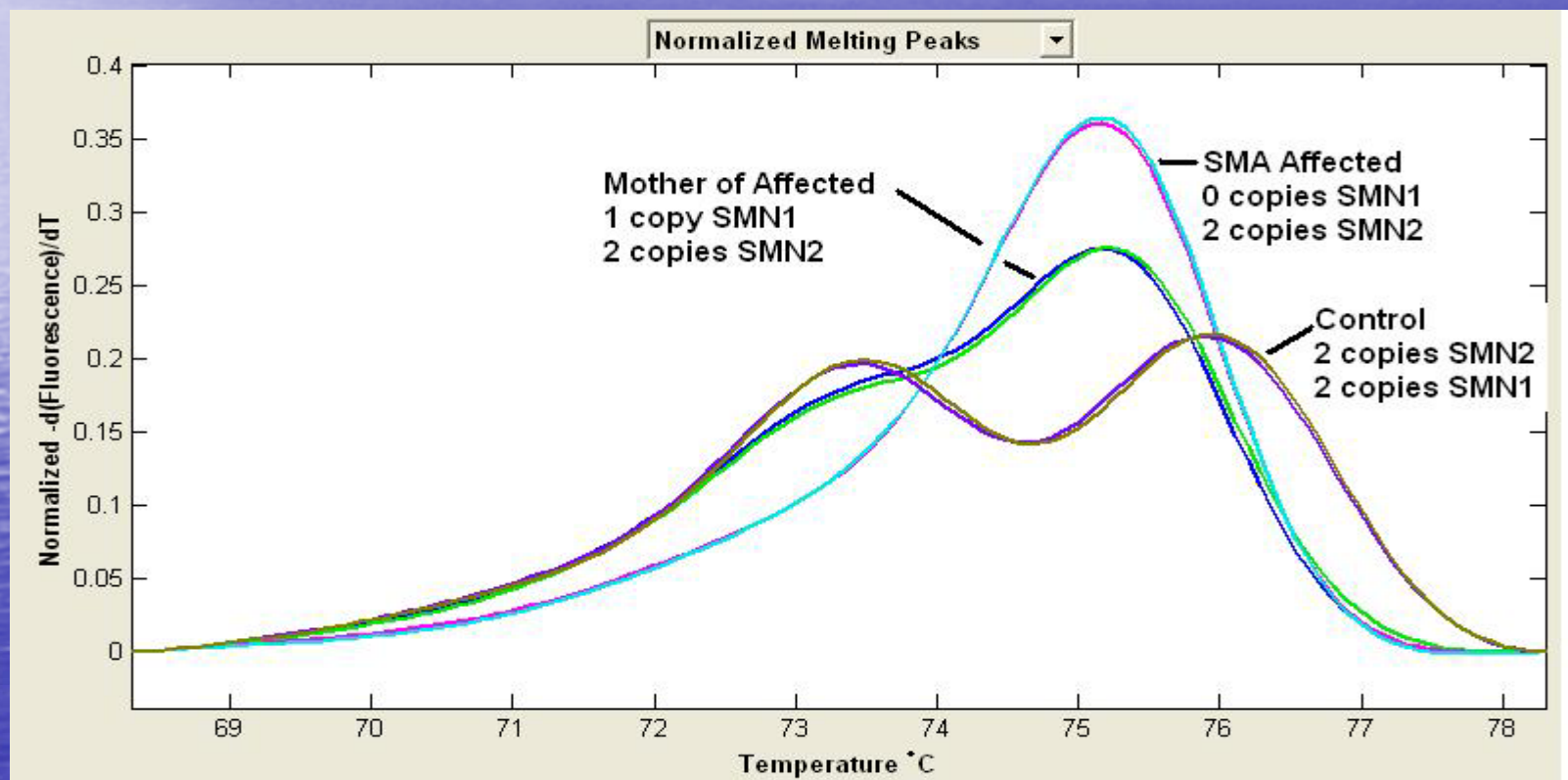
PENNSYLVANIA EXPERIENCE WITH NEWBORN DMD/BMD SCREENING (July 1986 – December 1995)

• Duchenne Muscular Dystrophy	32
• Becker Muscular Dystrophy	4
• DMD/BMD (No Deletion)	19
• Congenital/Limb Girdle MD	3
• Unknown	1
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Total	59

NEWBORN SCREENING PROTOCOL FOR SPINAL MUSCULAR ATROPHY

- 1^o Screen using Dried Blood Specimens (DBS)
 - SMN1 exon 7 homozygous deletion
 - 92-98% of cases
 - High Resolution Melting Assay
- 2^o Screen using same DBS
 - SMN2 assay on same DBS
 - Copy number indicates prognosis
- Pilot screening program underway
 - Utah and Colorado

HIGH RESOLUTION SMA ASSAY



DEVELOPING TREATMENTS

- Duchenne Muscular Dystrophy
 - Antisense Multiexon Skipping
 - Gene Therapy
- Spinal Muscular Atrophy
 - Enhance SMN2 Transcription
 - Stimulate Exon 7 Splicing
 - Protein Stabilization

PHYSICIAN SURVEYS

Specialty of Physicians (%)

<u>Specialty</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
Pediatrics	35.1	77.3	68.6
Neonatology	0.0	5.0	4.5
Genetics	0.5	0.0	1.7
Ped Neurology	2.3	0.6	1.0
Family Practice	34.7	1.1	1.0
Obstetrics	17.2	0.0	0.0
Other	10.4	0.0	3.8
No Response	0.0	16.0	19.4

PHYSICIAN SURVEY

Type of Practice of Physicians (%)

<u>Type</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
Private	63.3	65.2	57.9
Residency	19.8	0.6	5.9
Academic	16.6	13.8	20.0
Other	0.0	20.4	16.2

PHYSICIAN SURVEY

Age of Physicians (%)

<u>Age</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
21-31	18.6	2.2	5.8
31-40	43.9	17.1	27.6
41-50	13.1	25.4	26.9
51-60	14.0	34.3	21.7
61+	10.4	19.9	16.9
No Response	0.0	1.1	1.0

PHYSICIAN SURVEY

Gender of Physicians (%)

<u>Gender</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
Male	66.8	51.9	54.5
Female	33.2	47.0	44.8
No Response	0.0	1.1	0.0

PHYSICIAN SURVEY

Race/Ethnicity of Physicians (%)

<u>Race/Ethnicity</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
Caucasian	90.2	81.2	81.0
Asian	6.1	9.4	6.2
Afro-American	1.9	2.8	5.2
Hispanic	0.5	0.6	3.8
Other	0.0	0.6	0.0
No Response	1.4	5.5	3.8

Physician Attitudes Toward Newborn Screening For Muscular Dystrophy

<u>Attitude</u>	<u>PA 1990</u>	<u>PA 2012</u>	<u>SC 2012</u>
Strongly For	26.4	74.0	53.4
<u>Mildly For</u>	<u>33.8</u>	<u>18.8</u>	<u>36.6</u>
Total For	60.2	92.8	90.0
Strongly Against	4.6	1.1	1.4
<u>Mildly Against</u>	<u>10.2</u>	<u>6.1</u>	<u>7.9</u>
Total Against	14.8	7.2	9.3
No Opinion	25.0	0.0	0.7

Physician Attitudes Toward Newborn Screening For Spinal Muscular Atrophy

<u>Attitude</u>	<u>PA 1990</u> (Not Done)	<u>PA 2012</u>	<u>SC 2012</u>
Strongly For		74.0	51.4
<u>Mildly For</u>		<u>17.1</u>	<u>37.9</u>
Total For		91.1	89.3
Strongly Against		1.7	2.4
<u>Mildly Against</u>		<u>5.5</u>	<u>7.6</u>
Total Against		7.2	10.0
No Opinion		1.7	0.7

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