

The Untold PKU Testing Story...and Why it Challenges Government-Mandated Newborn (Genetic) Screening

by Twila Brase*

Newborn screening advocates often refer to the newborn PKU (phenylketonuria) test as evidence of the benefit of screening—and as a rationale for compulsory testing of newborns nationwide.

However, a brief look into the history of PKU testing challenges these assertions. Inaccurate test results, harmed children, untested treatments, and an *increase* in mental retardation mark the untold PKU story.

Now, in the 21st century, an attempt to test all newborn babies for a broad range of genetic conditions is emerging—again without scientific evidence of benefit or proper assessment of risk.

Only two states require parent consent.

With children at risk for discrimination and concerns about eugenics arising—and because Congress enacted a 2008 law to nationalize screening—initiatives to protect children, including informed consent as recommended by the Institute of Medicine should be required for newborn (genetic) screening.

Overview

The nearly 50-year old PKU test, performed by taking blood from the baby's heel at birth, is often used as rational to push for compulsory genetic testing of newborn babies today. The test is hailed as having saved countless children from mental retardation.

While the test has been beneficial to many babies, it has also caused devastating harm to

other children, including nutritional deficiencies, death, and an *increase* in the number of children with mental retardation.

History often repeats itself. The failure to properly evaluate the PKU story has led to today's untested expansion of compulsory genetic testing programs for newborns, say experts in research and ethics.

PKU is a rare condition. Approximately, one child out of every 15,000 infants born in the

United States is diagnosed with phenylketonuria (PKU),¹ an enzyme deficiency causing high phenylalanine blood levels that can lead to mental retardation.²

Despite protests from physicians and concerned researchers, the PKU test was mandated nationwide in the mid-1960s. After few infants were discovered to have PKU, some state legislators threatened to discontinue the testing program. In response, advocates of screening children at birth fought to expand the testing program beyond PKU—without proper study of the benefits or risks.

In recent years, the testing program for a broader list of genetic conditions became known as “newborn screening.”³ Attempts to expand the number of conditions for which all infants are tested continue today with little scientific evidence, or legislative oversight.

Like the PKU story, the benefits of genetic testing may be exaggerated and the pitfalls minimized.

Experts in the fields of genetics and bioethics have already expressed concerns about the impact of emerging DNA chip-enhanced testing of infants, including violation of the child’s right “not to know,” potential for future discrimination, use of newborn screening for research purposes, and the possibility of eugenics.

It is time for informed parent consent to be required for all newborn genetic testing. As Dr. Diane Paul, author of “The History of Newborn Phenylketonuria Screening in the U.S.,” a U.S. Task Force on Genetic Testing report, says,

“[A] ‘technological imperative’ has combined with unrealistic assumptions about benefits [of newborn screening], and that drives the expansion of screening programs. The lesson that such wholesale expansion is unwarranted has been repeatedly

drawn since the early 1960s. Surely it is time to heed it.”⁴

It’s Time to Act

Medical practice standards have long upheld informed consent, and nearly 15 years ago, the Institute of Medicine recommended explicit informed parent consent for newborn genetic testing, including the controversial PKU test. For the protection of all children and families, it is time to:

- Require explicit informed parent consent for newborn genetic testing
- Require that parents be fully informed of the many potential risks associated with genetic testing of children.
- Allow parents to choose and limit what conditions their child is tested for at birth.
- Limit testing to newborn conditions, not adult-onset diseases.
- Require informed parent consent for government storage, use, and dissemination of newborn blood and DNA.
- Provide individual property rights to the newborn blood and DNA stored in state health departments across the United States.
- Consider making newborn genetic testing a function of the private sector again, rather than a function of government.

Full Report Available Online:

www.cchconline.org/CCHCpkunbsReport092408.pdf

¹ “Minnesota Newborn Screening Program Provider Fact Sheet: Phenylketonuria (PKU).” Minnesota Department of Health. May 2005.

² <http://www.medhelp.org/lib/pku.htm>

³ “What’s in a name? Newborn Screening—It’s not just PKU.” Minnesota Department of Health Newborn Screening Bulletin – July 2005: Vol 3, Issue 2.

⁴ “Appendix 5. The History of Newborn Phenylketonuria Screening in the U.S.” Diane B. Paul. Final Report of the Task Force on Genetic Testing. National Human Genome Research Institute. Sept. 1997.

PKU Prevalence

State	Total 2006 confirmed PKU cases ⁵	Total 2007 confirmed PKU cases ⁶	Approx. Annual # of Births ⁷
Alabama	4	6	58,900
Alaska	0	2	9,800
Arizona	4	3	87,400
Arkansas	3	1	36,800
California	18	12	529,500
Colorado	0		68,500
Connecticut	1	0	42,600
Delaware	0	1	11,300
District of Columbia	0		15,000
Florida	12	5	205,500
Georgia	1		134,600
Hawaii		0	17,500
Idaho	6	2	20,400
Illinois	7	8	177,600
Indiana	11	8	85,500
Iowa	2	2	37,800
Kansas	2	1	39,700
Kentucky	1		52,700
Louisiana	1	4	65,100
Maine	0	0	13,400
Maryland	0	5	68,800
Massachusetts	8	2	81,700
Michigan	3	2	126,000
Minnesota	7	7	73,000
Mississippi	1	2	40,500
Missouri	5	5	76,400
Montana	0		11,000
Nebraska	0		27,000+
Nevada	2	1	32,200
New Hampshire	2		13,900
New Jersey	6	2	111,800
New Mexico	0	1	27,300
New York	3		252,300
North Carolina	5	3	118,200
North Dakota	0	2	8,900
Ohio	10		149,000
Oklahoma	2	2	51,000
Oregon	2	4	46,100
Pennsylvania	8	13	142,950
Rhode Island			13,550
South Carolina	1	2	52,200
South Dakota	1	2	11,000
Tennessee	0	0	82,600
Texas	10	13	374,100
Utah	8	4	50,300
Vermont	0	0	6,100
Virginia	7	2	97,400
Washington	3	5	78,600
West Virginia	0	1	21,100
Wisconsin	4	5	67,400
Wyoming	1	0	5,800
TOTAL CASES	172	141	
2006 Birth Rate	4,224,267		
2007 Birth Rate		4,253,538	

⁵ Cases of Classical PKU, National Newborn Screening and Genetics Resource Center. Accessed February 21, 2008.

⁶ Cases of Classical PKU, National Newborn Screening and Genetics Resource Center. Accessed February 21, 2008.

⁷ "State Map Page," National Newborn Screening & Genetics Resource Center, as updated August 2007.

No Consent⁸

State	Consent for Newborn Testing Required ^{9 10}	Number of Mandated Conditions for Testing ¹¹	Newborn Testing for Cystic Fibrosis ¹²
Alabama	N	39	Y
Alaska	N	45	Y
Arizona	N	28	Y
Arkansas	N	30	Y
California	N	50	Y
Colorado	N	49	Y
Connecticut	N	46	Offered/By Request
Delaware	N	44	Y
District of Columbia	N	52	Y
Florida	N	35	Y
Georgia	N	44	Y
Hawaii	N	45	Y
Idaho	N	45	Y
Illinois	N	48	Y
Indiana	N	50	Y
Iowa	N	50	Y
Kansas	N	30	Y
Kentucky	N	31	Y
Louisiana	N	30	Y
Maine	N	40	Y
Maryland	Y	50	Y
Massachusetts	N	37	Y
Michigan	N	49	Y
Minnesota	N	53	Y
Mississippi	N	50	Y
Missouri	N	36	Y
Montana	N	43	Y
Nebraska	N	34	Y
Nevada	N	44	Y
New Hampshire	N	35	Y
New Jersey	N	24	Y
New Mexico	N	29	Y
New York	N	51	Y
North Carolina	N	40	N
North Dakota	N	51	Y
Ohio	N	40	Y
Oklahoma	N	53	Y
Oregon	N	34	Y
Pennsylvania	N	16	Offered/By Request
Rhode Island	N	32	Y
South Carolina	N	52	Y
South Dakota	N	49	Y
Tennessee	N	52	Y
Texas	N	30	Not yet implemented
Utah	N	45	N
Vermont	N	34	Y
Virginia	N	28	Y
Washington	N	28	Y
West Virginia	N	33	Y
Wisconsin	N	49	Y
Wyoming	Y	30	Y

⁸ This table reflects all conditions mandated by law or rule, not: 1) testing states may offer for conditions not mandated, 2) other conditions revealed as byproducts of testing, or 3) required HIV testing (CT, IL, NY).

⁹ According to the March 2003 GAO report, "Newborn Screening," 33 states allow religious objection; 13 states allow objection for any reason; 5 states allow no exemptions. Allowing for objections does not require parents to be informed of the right to object. Only 11 states require parents be informed of the program at the time of screening.

¹⁰ "Newborn Screening," Government Accountability Office, 3/2003.

¹¹ "National Newborn Screening Status Report [Updated 09/16/08]" National Newborn Screening and Genetics Resource Center.

¹² "National Newborn Screening Status Report [Updated 09/16/08]" National Newborn Screening and Genetics Resource Center