Analysis of Shipments from the Coriell Institute and Results from Surveys at Genetics Meetings

Jeanne C. Beck, Ph.D.

Coriell Institute for Medical Research

Outline of the Presentation

- Shipments of Samples as Positive Controls
- Results of Surveys at Meetings

DNA Shipments
January 1, 2002 - October 31, 2005

	Number Purchased	Number for Positive Controls	%
USA	67,978	12,775	18.8
Non US	10,556	2,745	26.0
%	13.4	17.7	

Non US DNA Shipments For Use As Positive Controls January 1, 2002-October 31, 2005

Total non US DNA shipments for use as positive controls = 1,037

Disease (Gene)	Number	% of Total
Cystic Fibrosis (*CFTR*)	339	32.69
Fragile X (*FMR1)*	119	11.48
Coagulation Defects (*FVL/MTHFR*)	73	7.04
Breast Cancer (*BRCA1*)	35	3.38
Aneuploid Chromosome Number	34	3.28
Prader-Willi Syndrome (*PWS)*	33	3.18
Coagulation Factor II (*F2)*	30	2.89
Tay-Sachs Disease (*HEXA*)	26	2.51
Angelman Syndrome (*AS*)	22	2.12

Non US DNA Shipments For Use As Positive Controls (continued) January 1, 2002-October 31, 2005

Total non US DNA shipments for use as positive controls = 1,037

Disease (Gene)	Number	% of Total
Friedreich Ataxia (*FRDA*)	18	1.74
Canavan Disease (ASPA)	18	1.74
Gaucher Disease I, II, III (*GBA*)	15	1.45
Bloom Syndrome (*RECQL3*)	14	1.35
Spinocerebellar Ataxia I (*SCA1*)	13	1.25
Glycogen Storage Disease (*G6PC*)	12	1.16
Hemochromatosis (*HFE)*	11	1.06
Beta-Thalassemia (*HBB*)	11	1.06
Huntington Disease (*HD*)	10	0.96

Shipments of Disease Specific Positive Control DNA January 1, 2002-October 31, 2005

	CF	Fragile X	FVL/ MTHFR	HFE	HD
Total DNA	3,292	822	1,742	232	59
Use as Positive Control	1,994	705	1,334	133	51
% for Positive Control	60.5	85.8	76.6	57.3	86.4
Non US DNA	387	136	82	14	15
Non US Positive Control	339	119	73	11	11
% for Positive Control	87.6	87.5	89.0	78.6	73.3

Shipments of Disease Specific Positive Control DNA to Europe January 1, 2002-October 31, 2005

	CF	Fragile X	FVL/ MTHFR	HFE	HD
Non US DNA	339	119	73	11	11
DNA to Europe	199	70	49	3	8
% To Europe	58.7	58.8	67.1	27.3	72.7

Shipments of Disease Specific Positive Control DNA to Europe January 1, 2002-October 31, 2005

	CF	Fragile X	FVL/ MTHFR	HFE	HD
Belgium	121	4	25	1	0
France	0	0	6	1	0
Germany	25	8	6	0	0
Greece	1	8	2	0	1
Ireland	9	42	0	0	0
Netherlands	4	0	0	0	0
Norway	0	5	4	1	1
Poland	0	0	0	0	0
Sweden	25	3	0	0	6
Switzerland	14	0	6	9	0

Shipments of Disease Specific Positive Control DNA January 1, 2002-October 31, 2005

	Tay-Sachs	Gaucher	Familial Dysauto	Bloom Syndome
Total DNA	118	103	83	59
Use as Positive Control	89	78	66	44
% Positive Control	75.4	75.7	79.5	74.6
Non US DNA	28	20	10	15
Non US Positive Control	26	15	8	14
% for Positive Control	92.9	75.0	80.0	28.6

Shipments of Disease Specific Positive Control DNA January 1, 2002-October 31, 2005

	Canavan	Niemann-Pick	Fanconia Anemia C	Mucolipid-osis IV
Total DNA	52	26	15	28
Use - Positive Cont	44	20	10	24
% for Positive Control	84.6	76.9	66.7	85.7
Non US DNA	19	2	0	8
Non US Positive Control	18	1	0	7
% for Positive Control	94.7	50.0	0	87.5

Summary

- Approximately 20% of all DNA samples purchased is for use as positive/negative controls (18.8% in the US; 26% outside the US).
- Approximately 38% (1,037/2,745) of the non US samples shipped as positive controls were for genetic diseases.
- Since January 1, 2002, we have shipped more than 550 DNA samples for use as positive controls for CF, Fragile X, FVL/MTHFR, HFE, and HD abroad. These diseases represent 20% of the samples purchased outside of the US for use as positive controls. Three hundred twenty-nine [329 (12%)] were shipped to Europe.

Summary (continued)

- Approximately 33% (339/1039) of DNA samples purchased by non US researchers as positive controls for genetic diseases were cystic fibrosis, 11.5% were Fragile X, 1% was Huntington disease, and 8.5% were diseases in the Ashkenazi Jewish panel.
- More than 85% of the cystic fibrosis, Fragile X, and FVL/MTHFR DNA samples sent abroad were for use as positive controls.

Results from Surveys

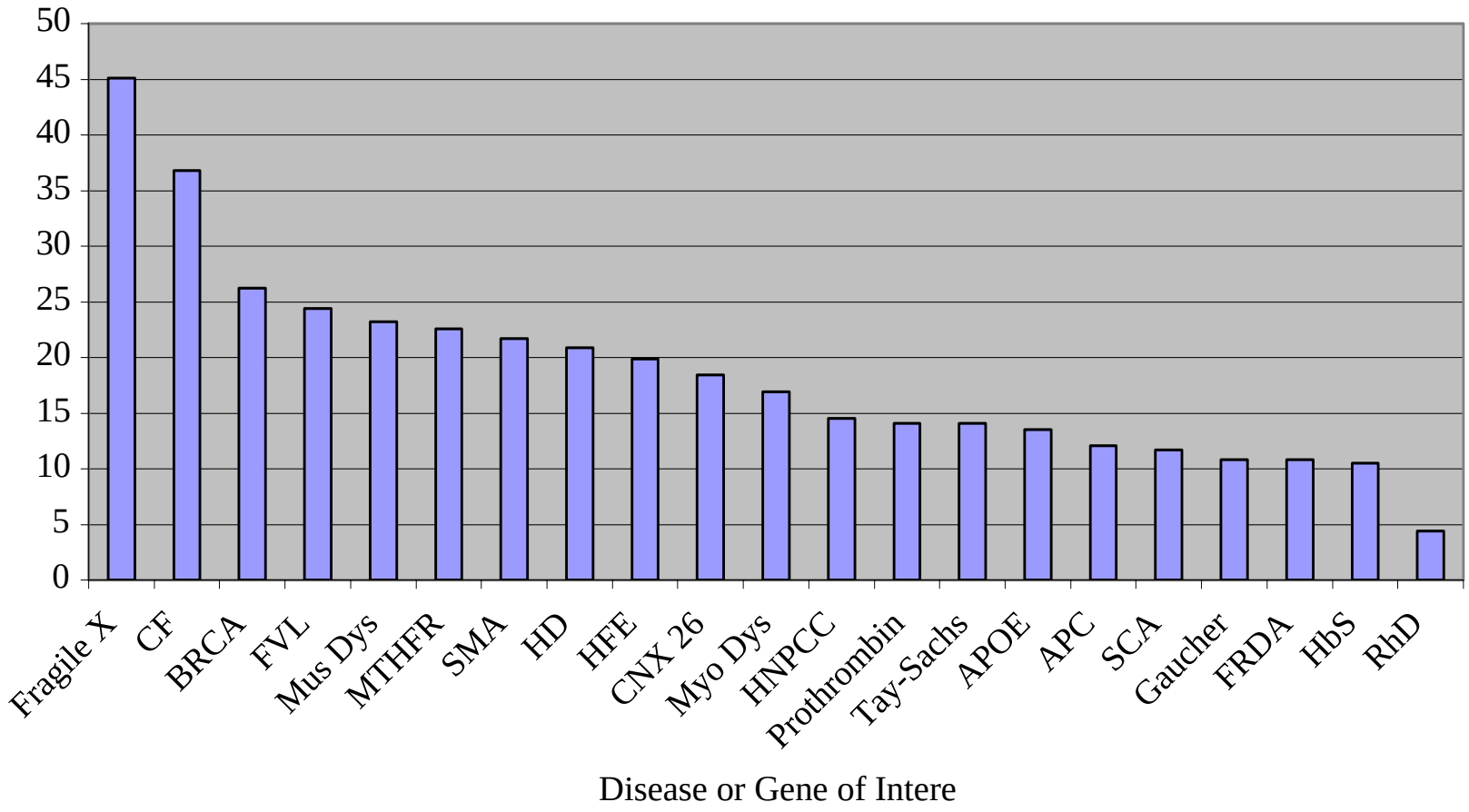
Data Acquisition

- Surveys were conducted at seven meetings (ESHG, ASHG) from 2001 to 2005 to gauge interest in samples with characterized mutations.
- The survey was administered to 633 individuals and 378 (59.7%) indicated that they would use samples with characterized mutations.
- Those who answered in the affirmative were asked to indicate the diseases of interest to them.

Cumulative Results

Disease/Gene	%*		Disease/Gene	%
Fragile X	45.1		CNX 26	18.4
Cystic Fibrosis	36.8		Myo Dys	16.9
BRCA1/2	26.2		*HNPCC*	14.5
FVL	24.4		Prothrombin	14.1
Mus Dys	23.2		Tay-Sachs	14.1
MTHFR	22.6		APOE	13.5
SMA	21.7		APC	12.1
HD	20.9		SCA	11.7
HFE	19.9		Gaucher	10.8

*Percentage of Those Responding


Requests for Characterized Mutations from Surv
50
45
40
35
30
25
20
15
10
5
0
Fragile X
CF
BRCA
FVL
Mus Dys
MTHFR
SMA
HD
HFE
CNX 26
Myo Dys
HNPCC
Prothrombin
Tay-Sachs
APOE
APC
SCA
Gaucher
FRDA
HbS
RhD
Disease or Gene of Intere

Summary

- Samples requested most frequently include those for Fragile X, cystic fibrosis, *BRCA1/2*, and FVL/MTHFR.
- This correlates well with the data from shipments.