

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

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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 (<i>CFTR</i>)	339	32.69
Fragile X (<i>FMR1</i>)	119	11.48
Coagulation Defects (<i>FVL/MTHFR</i>)	73	7.04
Breast Cancer (<i>BRCA1</i>)	35	3.38
Aneuploid Chromosome Number	34	3.28
Prader-Willi Syndrome (<i>PWS</i>)	33	3.18
Coagulation Factor II (<i>F2</i>)	30	2.89
Tay-Sachs Disease (<i>HEXA</i>)	26	2.51
Angelman Syndrome (<i>AS</i>)	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 (<i>FRDA</i>)	18	1.74
Canavan Disease (<i>ASPA</i>)	18	1.74
Gaucher Disease I, II, III (<i>GBA</i>)	15	1.45
Bloom Syndrome (<i>RECQL3</i>)	14	1.35
Spinocerebellar Ataxia I (<i>SCA1</i>)	13	1.25
Glycogen Storage Disease (<i>G6PC</i>)	12	1.16
Hemochromatosis (<i>HFE</i>)	11	1.06
Beta-Thalassemia (<i>HBB</i>)	11	1.06
Huntington Disease (<i>HD</i>)	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 Syndrome
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	Mucopolid- 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

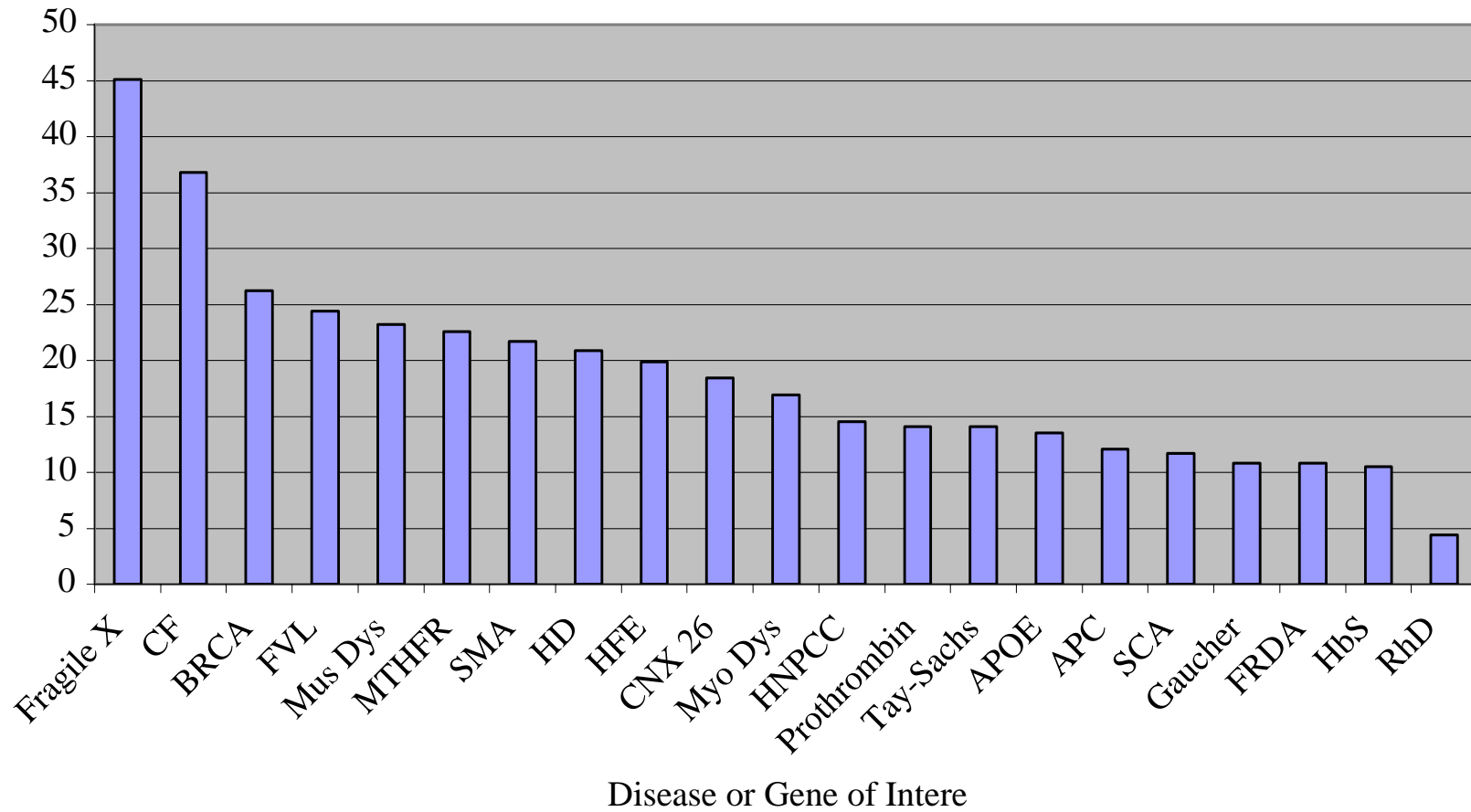
- Surveys were conducted at seven meetings (ESHG, ASHG) from 2001 to 2005 to gauge interest in samples with characterized mutations.
- The survey was administered to 633 individuals and 378 (59.7%) indicated that they would use samples with characterized mutations.
- Those who answered in the affirmative 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
<i>BRCA1/2</i>	26.2		<i>HNPCC</i>	14.5
<i>FVL</i>	24.4		Prothrombin	14.1
Mus Dys	23.2		Tay-Sachs	14.1
<i>MTHFR</i>	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



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.