The American Journal of Human Genetics, Volume 98

Supplemental Information

A Clinical Service to Support the Return

of Secondary Genomic Findings in Human Research

Andrew J. Darnell, Howard Austin, David A. Bluemke, Richard O. Cannon III, Kenneth Fischbeck, William Gahl, David Goldman, Christine Grady, Mark H. Greene, Steven M. Holland, Sara Chandros Hull, Forbes D. Porter, David Resnik, Wendy S. Rubinstein, and Leslie G. Biesecker

Cost estimates for a Secondary Genomics Findings Consultation service in the Intramural Research Program of the National Institutes of Health.

Table S1: Overall Costs

Number of									
Analyzed	Number of	Salaries							
Exomes or	secondary	&	Sample			Office &		Total	Cost
genomes	findings per	Benefits	intake	ABI	PCR	Computer	Fixed	Projected	per
per year	year	(Table 2)	costs	Arrays	Validations	Expenses	Costs*	Cost	Exome
1,000	50	\$82,600	\$500	\$1,000	\$3,000	\$2,500	\$50,000	\$139,600	\$140
5,000	250	\$188,000	\$2,500	\$1,000	\$15,000	\$5,000	\$50,000	\$261,500	\$52
10,000	500	\$327,600	\$5,000	\$1,000	\$30,000	\$10,000	\$50,000	\$423,600	\$42
20,000	1,000	\$516,000	\$10,000	\$2,000	\$60,000	\$15,000	\$50,000	\$653,000	\$33

Fixed costs include software licensing, sequencer service contract and amortization, etc.

Table S2: Staffing Costs

		Number of Exomes/Genomes Sequenced					
	Annual						
	Personnel						
	Cost	1,000	5,000	10,000	20,000		
Clinical Support	\$40,000	\$4,000	\$20,000	\$40,000	\$80,000		
Bioinformatics	\$200,000	\$20,000	\$40,000	\$80,000	\$80,000		
Lab Technician	\$60,000	\$9,000	\$12,000	\$30,000	\$60,000		
Lab Director	\$200,000	\$40,000	\$80,000	\$120,000	\$200,000		
Genetic Counselor	\$96,000	\$9,600	\$36,000	\$57,600	\$96,000		
Lab costs							
Total	\$82,600	\$188,000	\$327,600	\$516,000			
Per exome/genome	\$82.60	\$37.60	\$32.76	\$25.80			

We have specified personnel costs using an estimated fraction of effort (e.g., for 1,000 exomes per year, we estimate it would require about 10% effort or 4 hours per week of a support person whose salary and benefits would be \$40,000 per year). Laboratory costs were estimated to be

Scope of Staff Work:

Clinical Support: Arranging for sample intake/shipping, ordering, & patient scheduling.

Bioinformatician: Running filters to screen submitted variants for basic pathogenicity criteria, identifying relevant publications & database entries for variants that pass filters, programming duties, & updating filters.

Lab technician: Sample accessioning, DNA isolation, database entry, PCR validations, organizing and preparing data for Lab Dir. Lab Director: CLIA licensing, training staff, monitoring literature, performing final pathogenicity determinations, reviewing and signing final reports, proposing gene list changes.

Genetic Counselor: Returning findings to research participants (obtain pedigree, provide genetic counseling, and communicating result to identified local provider).