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ERRATUM

Cost-effectiveness of exome and genome sequencing for children with rare and undiagnosed conditions

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In the article "Cost-effectiveness of exome and genome sequencing for children with rare and undiagnosed conditions" by Lavelle et al (*Genet Med* 2022;24:1349-1361), there were errors in Table 1 and Table 3 where the table footnote citations were incorrect. Please see the revised Table 1 and Table 3 shown below. The article has been corrected online and can be accessed at https://www.sciencedirect.com/science/article/pii/S1098360022006827?via%3Dihub.

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Table 1 Select model parameters for base case and sensitivity analysis

Variable	Base Case	Range (min-max)[a]	Reference
Probability of diagnosis following testing strategy			
Infants			
First tier SOC	0.09	0.02-0.23	Farnaes et al[20], Willig et al[21]
First tier trio ES	0.37	0.28-0.43	Used estimates from Lionel et al[22] to adjust GS diagnostic rates to ES diagnostic rates
First tier trio GS	0.49	0.37-0.74	Farnaes et al[20], Willig et al[21]
All children			
First tier trio SOC	0.19	0.07-0.28	Lionel et al[22], Stavropoulos et al[23]
First tier trio ES	0.28	0.21-0.32	Used estimates from reference Lionel et al[22] to adjust GS diagnostic rates to ES diagnostic rates
First tier trio GS	0.37	0.28-0.51	Lionel et al[22], Stavropoulos et al[23]
Testing costs			
Infants			
SOC (strategy A) testing before Dx	$2154[b]	$1077-$6462	Shashi et al[24]
SOC testing before ES/GS	$2154[b]	$1077-$6462	Shashi et al[24]
Diagnostic odyssey (SOC testing w/no Dx)	$6566[c]	$3283-$19,698	Dragojlovic et al[25]
ES trio rapid test	$10,320	$6720-$13,920	Adjusted list prices[d]
GS trio rapid test	$12,000	$9000-$15,000	Adjusted list prices[d]
All children			
SOC (strategy A) testing before Dx	$2154[b]	$1077-$3231	Shashi et al[24]
SOC testing before ES/GS	$2154[b]	$1077-$3231	Shashi et al[24]
Diagnostic Odyssey (SOC testing w/no Dx)	$6566[c]	$3283-$19,698	Dragojlovic et al[25]
ES trio standard test	$8112	$6720-$10,560	Adjusted list prices[d]
GS trio standard test	$10,450	$7008-$14,304	CMS reimbursement rate 2019 Q4
After ES and GS testing costs, all ages			
With Dx	$135[e]	$135-$1350	Vrijenhoe et al[26]
Without Dx	$239[e]	$239-$2390	Vrijenhoe et al[26]

CAD, Canadian dollar; *CMS*, Centers for Medicare and Medicaid Services; *Dx*, diagnosis; *ES*, exome sequencing; *GS,* genome sequencing; *min*, minimum; *max*, maximum; *Q4*, fourth quarter; *USD*, US dollar; *w/no*, with no.

[a]Used in 1-way sensitivity analyses.

[b]This cost was derived from the study by Shashi et al,[24] which reported a charge of $3285 (2013 USD) for genetic testing performed on patients with suspected genetic conditions—who receive a diagnosis. This average charge included 41% of patients who incurred no charges ($0) for genetic testing because they were diagnosed by clinical criteria only, and 59% of the patients received testing at a charge of $5569 in 2013 USD. We converted this estimate of $5569 from charges to costs (using a cost-to-charge ratio of 0.33) and updated costs from 2013 to 2019 USD using the Consumer Price Index.

[c]This cost is derived from the study by Dragojlovic et al,[25] which estimated 10-year cost of the diagnostic odyssey for children with suspected genetic conditions. We included the cost estimates for diagnostic tests only. Costs were converted from 2016 CAD to 2019 USD. Future costs (years 2-10) were discounted to present value (year 1) costs using a 3% discount rate.

[d]All list prices were adjusted to expected CMS reimbursement rates using a ratio of 0.96. This ratio was derived by comparing mean list prices with 2019 Q4 CMS reimbursement rates for 3 tests for which we had both list prices and reimbursement rates: ES proband, GS proband, and GS trio.

[e]These estimates are derived using information from the study by Vrijenhoek et al[26] that reported that laboratory, diagnostic, and genetic testing costs are 70% to 90% lower after ES than before and vary on the basis of whether the child does or does not receive a diagnosis.

Table 3 Sensitivity analysis results for primary incremental cost per diagnosis analyses

	Infants (Base Case: GS $15,048/Diagnosis vs SOC)				All children (Base Case: GS $27,349/Diagnosis vs SOC)			
	First Alternative Assumption		Second Alternative Assumption		First Alternative Assumption		Second Alternative Assumption	
Sensitivity Analysis[a] (Base Case Assumption)	Value #1	Incremental Cost Per Diagnosis	Value #2	Incremental Cost Per Diagnosis	Value #1	Incremental Cost Per Diagnosis	Value #2	Incremental Cost Per Diagnosis
Cost of diagnostic odyssey ($6566)	$3283	GS: $22,157/Dx vs SOC	$19,698	ES: cost-saving vs SOC GS: $13,896/Dx vs ES	$3283	GS: $42,122/Dx vs SOC	$19,698	ES: cost-saving vs SOC GS: $25,874/Dx vs ES
Cost of GS (infants: $12,000 children: $10,450)	$9000	GS: $7548/Dx vs SOC	$15,000	ES: $15,541/Dx vs SOC GS: $38,896/Dx vs ES	$7008	GS: $8227/Dx vs SOC	$14,304	ES: $28,824/Dx vs SOC GS: $62,147/Dx vs ES
GS diagnostic rate (infants: 0.49 children: 0.37)	0.37	ES[b]: $15,541/Dx vs SOC	0.74	GS: $9220/Dx vs SOC	0.28	ES[b]: $28,824/Dx vs SOC	0.51	GS: $15,338 /Dx vs SOC
SOC diagnostic rate (infants: 0.09 children: 0.19)	0.02	ES: $11,551/Dx vs SOC GS: $13,896/Dx vs ES	0.23	SOC/GS[b]: $23,076/Dx vs SOC	0.07	ES: $9832/Dx vs SOC GS: $25,874/Dx vs ES	0.28	GS: $50,112/Dx vs SOC
Cost of SOC testing ($2154)	$1077	SOC/GS[b]: $15,252/Dx vs SOC	$6462	GS: $14,079/Dx vs SOC	$1077	SOC/GS[b]: $23,296/Dx vs SOC	$6462	ES: $19,729/Dx vs SOC GS: $25,874/Dx vs ES
ES diagnostic rate (infants: 0.37 children: 0.28)	0.21	GS: $15,048/Dx vs SOC	0.43	ES: $12,780/Dx vs SOC GS: $27,896/Dx vs ES	0.21	GS: $27,349/Dx vs SOC	0.43	ES: $19,923/Dx vs SOC GS: $46,656/Dx vs ES
ES cost (infants: $10,320 children: $8112)	$6720	GS: $15,048/Dx vs SOC	$13,920	GS: $15,048/Dx vs SOC	$6720	ES: $13,357/Dx vs SOC GS: $41,340/Dx vs ES	$10,560	GS: $27,349/Dx vs SOC
Cost after GS or ES with Dx ($135)	$1350	GS: $16,536/Dx vs SOC	[c]	[c]	$1350	GS: $29,358/Dx vs SOC	[c]	[c]
Cost after GS or ES with no Dx ($239)	$2390	GS: $17,790/Dx vs SOC	[c]	[c]	$2390	GS: $34,877/Dx vs SOC	[c]	[c]
Family members tested (trio)	Proband	GS: $7022/Dx vs SOC[d]	[c]	[c]	Proband	ES: cost-saving vs SOC GS: $3076/Dx vs ES[e]	[c]	[c]
Reanalysis (no reanalysis)	Yes	ES: $12,142/Dx vs SOC GS: $15,980/Dx vs ES[f]	[c]	[c]	Yes	ES: $14,227/Dx vs SOC GS: $30,078/Dx vs ES[g]	[c]	[c]
Strategies included (SOC, ES, GS)	SOC, ES	ES: $15,543/Dx vs SOC[h]	[c]	[c]	SOC, ES	ES: $28,822/Dx vs SOC[i]	[c]	[c]
Cost of SOC testing ($2154) and cost of diagnostic odyssey ($6566)[j]	$1077, $3283	SOC/GS: $22,721/Dx vs SOC	$6462, $19,698	ES: cost-saving vs SOC GS: $13,896 vs ES	$1077, $3283	SOC/GS: $38,070/Dx vs SOC	$6462, $19,698	ES: cost-saving vs SOC GS: $25,874/Dx vs ES

Dx, diagnosis; *ES*, exome sequencing; *GS*, genome sequencing; *SOC*, standard of care.

[a]This table describes how changing base case assumptions in our model influences our model's projection for our primary analysis (10-year cost per infant or child diagnosed). In the first row, the far left column identifies the assumption to be altered—in this case, the assumed cost of the diagnostic odyssey and the value of this assumption in the base case analysis. The left portion of the table describes the effect of alternative values for this assumption on the incremental cost of diagnoses in infants. Adopting the first alternative assumption (diagnostic odyssey cost of $3282) means that the estimated incremental cost per diagnosis for GS is $22,157 compared with SOC. Adopting the second alternative assumption (diagnostic odyssey cost of $19,698) means that ES is cost-saving compared with SOC, and the incremental cost per diagnosis for GS is $13,896 compared with ES. The right portion of the table reports corresponding results for our projections for all children.

[b]GS has same diagnostic rate and higher cost.

[c]Only 1 alterative assumption was tested.

[d]See also Supplemental Table 10.

[e]See Supplemental Table 11.

[f]See Supplemental Table 12.

[g]See Supplemental Table 13.

[h]See Supplemental Table 14.

[i]See Supplemental Table 15.

[j]Values of these 2 parameters were varied simultaneously between low and high bounds.