

Table S1: Model parameter and expected values with ranges for Markov model

Parameters	Expected values	Distribution	Low	High	Source
Cost parameters	US\$ (A\$)				
Screening test cost	\$4.82 (\$7)	Gamma	-10%	+10%	NBS Pilot
Repeat screening test cost	\$6.89 (\$10)	Gamma	7	25	NBS Pilot
SMA1 annual cost	\$115,858 (\$231,717)	Gamma	\$50,000	\$490,000	[34]
SMA2 annual cost	\$76,234 (\$152,469)	Gamma	\$40,000	\$360,000	[34]
SMA3 annual cost	\$47,983 (\$95,966)	Gamma	\$20,000	\$180,000	[34]
Gene therapeutic cost	\$1,540,000 (\$2,234,540)		\$1,270,000	\$2,100,000	[36]
Gene Tx follow-up cost	\$158 (\$230)				[37]
Gene Tx initial year cost	\$4,312 (\$4574)				[37,38]
Nusinersen Injection episode cost	\$3,731 (\$5,414)				[37,38]
Nusinersen loading cost	\$318,164 (\$461,656)				[2,39]
Nusinersen maintenance cost	\$119,311 (\$173,123)		\$79,939	\$131,242	[2,39]
Nusinersen one dose cost	\$75,810 (110,000)		\$68,228	\$83,390	[39]
Confirmatory diagnostic SCID testing cost (after positive screening test)	\$2119 (\$3,074)				[13]
Diagnostic SCID testing cost (without NBS)	\$3446 (\$5,000)				[13]
Pre-symptomatic SCID cost	\$0 (\$0)				Assumption
HSCT cost (early diagnosis)	\$119,282 (\$173,078)	Gamma	-10%	+10%	[13]
HSCT cost (late diagnosis)	\$271,697 (\$394,233)	Gamma	-10%	+10%	[13]
Treatment cost for SCID patient dying prior to HSCT	\$178,923 (\$259,617)				[13]
SCID Well treatment cost (per year)	\$34 (\$50)				[13]
SCID Moderate treatment cost (per year)	\$24,052 (\$34,900)				[13]
SCID Poor treatment cost (per year)	\$12,873 (\$18,679)				[13]
End-of-life costs (SCID Moderate & SCID Poor)	\$41,841 (\$60,712)				[13]

SCID Well productivity cost	\$0 (\$0)			Assumption	
SCID Moderate productivity cost	\$1,394 (\$2,023)			[13]	
SCID Poor productivity cost	\$0 (\$0)			Assumption	
Netherlands, 2016 PPP Euros/US\$	0.796			[24]	
Australia, 2016 PPP AU\$/US\$	1.45			[24]	
CPI inflation rate 2016 to 2019 AU\$	0.0557			[40]	
Discount rate	0.03	-	0.05		
Outcome parameters					
SMA INCIDENCE	0.000091	Beta	0.000065	0.000119	[41]
SMA 1 mortality	0.29	Beta	0.227	0.353	[42]
SMA 2 mortality	0.004	Beta	0.0025	0.0059	[43]
SMA 3 mortality	Population background mortality				
SMA phenotype (SMA1, SMA2, SMA3)	0.58, 0.29, 0.13			[43]	
False negative % in screen	0.00000576			[25]	
Screen retest due to non-amplification	0.012			[25]	
Further test % in screen	0.0000854	Beta	0.000038	0.000134	[25]
SMA confirmation in re-test	1			[25]	
Motor milestone half-yearly transition					
Non-sitter to sit in early gene therapy	0.43	Beta	0.35	0.50	[2,44]
Non-sitter to sit in late nusinersen Tx	0.45	Beta	0.40	0.50	[45,46]
Sit to stand in early gene therapy	0.84	Beta	0.80	0.87	[2,44]
Sit to stand in late nusinersen Tx	0.55			[45,46]	
Stand to assisted walk in early gene therapy	0.74	Beta	0.70	0.78	[2,44]
Stand to assisted walk in late nusinersen Tx	0.21			[45,46]	
Walk assisted to independent walk in early gene therapy	0.81	Beta	0.73	0.88	[2,44]
Walk assisted to independent walk in late nusinersen Tx	0.58			[45,46]	
SCID incidence	0.00002	Beta	0.000012	0.000025	[3,47]
False Negative % in Screen (1-sensitivity)	0.005	Beta	0	0.01	[10]
False Positive % in Screen (1-specificity)	0.0003	Beta	0.0002	0.0008	[10]
% patients early diagnosed without NBS	0.2	Beta	0.1	0.3	[9]

Probability to survive until treatment (early diagnosis)	0.9423				[3]
Probability to survive until treatment (late diagnosis)	0.78				[10]
5-year survival (early diagnosis)	0.94	Beta	0.91	0.98	[48]
5-year survival (late diagnosis)	0.82	Beta	0.7	0.9	[48]
SCID Well after HSCT (early diagnosis; surviving subjects)	0.8				[13]
SCID Moderate after HSCT (early diagnosis; surviving subjects)	0.15				[13]
SCID Poor after HSCT (early diagnosis; surviving subjects)	0.05				[13]
SCID Well after HSCT (late diagnosis; surviving subjects)	0.5				[13]
SCID Moderate after HSCT (late diagnosis; surviving subjects)	0.3				[13]
SCID Poor after HSCT (late diagnosis; surviving subjects)	0.2				[13]
Quality of life utility value					
Non-sitter	0.02				[15,34,49]
Sit without support	0.11				[19,34,49]
Stand with assistance	0.25				[34]
Walk with assistance	0.38				[49]
Stand walk unaided	0.64				[49]
Utility value for presymptomatic SCID	0.95				Assumption
Utility value for SCID Well	0.95				[13]
Utility value for SCID Moderate	0.75				[13]
Utility value for SCID Poor	0.5				[13]
Utility value for HSCT	0.5				Assumption
Utility value for Deceased	0				Assumption