

Sample Prioritization: Scoring system for genome selection

	Available samples	Pre-screening	Consanguinity	Age at onset	Reviewer's personal interpretation (depending on specific clinical presentation)
Best	6 points: > 2 affected	3 points: MLPA-neg, exome or panel-neg, VCF file available	3 points: yes	3 points: <20 years	3 points
Second best	5 points: 1 affected, both healthy parents	2 points: MLPA negative, exome or panel negative, no raw data available		2 points: 21-40 years	2 points
Medium	4 points: 2 affected	1 point: some pre-screening		1 point: 41-50 years	1 point
Low	1 point: 1 affected but strong positive family history	0 points: no pre-screening	0 points: no	0 points: >50 years	0 points

This scoring system will be used to prioritize families where a monogenic cause is strongly suspected, please note this is a guide only.