

Samples	Target Region Average Coverage	Target Region Minimum Coverage	Target Region Maximum Coverage	Number of Poorly Covered Regions	Number of Poorly Covered Exons
All samples	70	18	4784	118858	27746
Patient 1	69	37	4604	120424	15806
Patient 2	76	34	5097	118485	48557
Patient 3	70	1	5196	119817	15677
Patient 4	79	32	5545	117523	48851
Patient 5	76	5	5754	118301	48068
Patient 6	70	7	5794	118664	49305
Patient 7	71	33	4529	120018	16990

Table S1. Coverage statistics. Average coverage statistics of all 342 sequence samples and detailed coverage statistics of Patients 1 to 7 are reported. Minimum coverage is reported considering bases with coverage depth >15. A coverage <15 is considered to define the number of poorly covered regions and poorly covered exons (target regions).