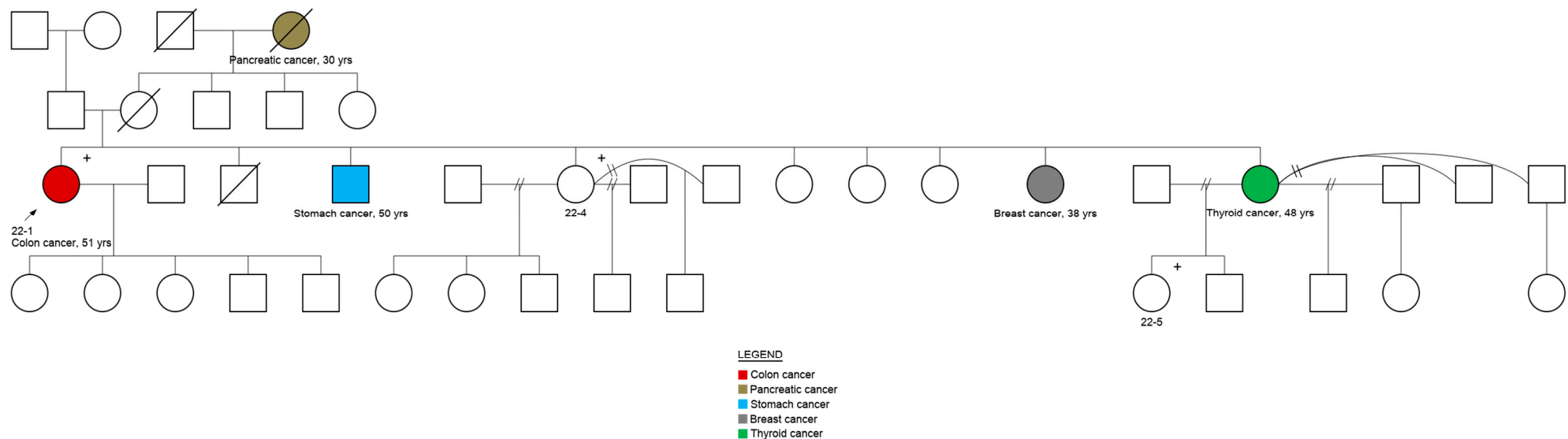


# Supplementary Material: Genetic Characterization in High-Risk Individuals from a Low-Resource City of Peru

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**Supplementary figure S1.** Family pedigree where the proband carried the variant *BRCA1* (ENST00000471181.2) c.4357+1G>A.



+: Carrier of *BRCA1* (ENST00000471181.2) c.4357+1G>A variant.

**Supplementary Table S1.** Summary of various quality metrics related to the primary analysis of the 84 analysed NGS samples.

SampleID	TotalReadCount	MarkedDuplicatesCount	MarkedDuplicates (%)	UniqueMappedCount	UniqueMapped (%)	MAPQ>39Count	MAPQ>39%	TargetMeanCoverage	SmallVariantsCount
SAMPLE_01	4306702	808172	18.77	3494143	81.13	4072617	94.56	329.26	1352
SAMPLE_02	5422346	1218150	22.47	4198994	77.44	5121517	94.45	407.48	1435
SAMPLE_03	4136992	780680	18.87	3351539	81.01	3888460	93.99	324.00	1287
SAMPLE_04	4217184	819023	19.42	3395240	80.51	3988817	94.58	330.65	1434
SAMPLE_05	3154060	510588	16.19	2641272	83.74	2974170	94.30	256.87	1426
SAMPLE_06	3667028	666226	18.17	2996904	81.73	3484546	95.02	296.26	1425
SAMPLE_07	4286010	832490	19.42	3449507	80.48	4041816	94.30	335.17	1338
SAMPLE_08	4400140	850979	19.34	3544581	80.56	4172516	94.83	344.74	1419
SAMPLE_09	3858364	707594	18.34	3146318	81.55	3638934	94.31	306.46	1356
SAMPLE_10	3988152	761105	19.08	3223256	80.82	3762974	94.35	310.24	1339
SAMPLE_11	4537970	946067	20.85	3587637	79.06	4288345	94.50	347.24	1294
SAMPLE_12	5362140	1244115	23.20	4112643	76.70	5084934	94.83	398.73	1412
SAMPLE_13	5410000	1355376	25.05	4048529	74.83	5228801	96.65	406.85	1369
SAMPLE_14	4472046	946908	21.17	3520519	78.72	4253573	95.11	345.48	1449
SAMPLE_15	4752614	1035745	21.79	3712154	78.11	4502667	94.74	362.38	1407
SAMPLE_16	3537878	629622	17.80	2904750	82.10	3321152	93.87	278.68	1295
SAMPLE_17	4839334	1073494	22.18	3760264	77.70	4572859	94.49	364.52	1405
SAMPLE_18	4707644	1166239	24.77	3536926	75.13	4452290	94.58	335.19	1212
SAMPLE_19	4363940	991754	22.73	3366396	77.14	4210221	96.48	343.78	1410
SAMPLE_20	4392822	885992	20.17	3502380	79.73	4155814	94.60	339.47	1358
SAMPLE_21	4147762	900059	21.70	3241837	78.16	4015566	96.81	338.09	1267
SAMPLE_22	4825444	1060434	21.98	3760443	77.93	4585233	95.02	366.01	1282
SAMPLE_23	5860556	1671934	28.53	4183130	71.38	5494262	93.75	395.39	1197
SAMPLE_24	5067370	1255720	24.78	3806225	75.11	4901888	96.73	393.60	1388
SAMPLE_25	3363226	670952	19.95	2688266	79.93	3254142	96.76	290.19	1396
SAMPLE_26	3458826	613676	17.74	2841369	82.15	3335661	96.44	271.70	1338
SAMPLE_27	3695812	674941	18.26	3018316	81.67	3482143	94.22	291.02	1344
SAMPLE_28	4989922	1280004	25.65	3704665	74.24	4817971	96.55	384.11	1368
SAMPLE_29	4485526	947248	21.12	3534256	78.79	4264763	95.08	344.69	1424
SAMPLE_30	5039630	1152117	22.86	3882771	77.04	4785938	94.97	377.40	1518
SAMPLE_31	4235578	821213	19.39	3410728	80.53	3987354	94.14	323.62	1283
SAMPLE_32	4177210	845637	20.24	3326007	79.62	3951943	94.61	319.53	1402
SAMPLE_33	5701898	1583567	27.77	4112106	72.12	5372858	94.23	390.83	1260
SAMPLE_34	4870290	1042338	21.40	3823458	78.51	4612145	94.70	370.58	1368
SAMPLE_35	4002138	751960	18.79	3245440	81.09	3770873	94.22	313.51	1296
SAMPLE_36	5393030	1416734	26.27	3966695	73.55	5096465	94.50	377.37	1240
SAMPLE_37	4041608	762968	18.88	3274249	81.01	3823807	94.61	312.94	1360
SAMPLE_38	5399666	1477613	27.36	3916460	72.53	5095500	94.37	367.92	1235

SAMPLE_39	3919572	875210	22.33	3042270	77.62	3691856	94.19	291.95	1293
SAMPLE_40	4332984	892022	20.59	3436977	79.32	4104886	94.74	334.20	1393
SAMPLE_41	3971380	781224	19.67	3186209	80.23	3775194	95.06	313.85	1380
SAMPLE_42	3732816	681963	18.27	3047544	81.64	3543952	94.94	295.02	1336
SAMPLE_43	3729244	686417	18.41	3040168	81.52	3516194	94.29	294.79	1391
SAMPLE_44	4124460	859458	20.84	3259705	79.03	3980052	96.50	331.44	1372
SAMPLE_45	5512724	1493879	27.10	4012552	72.79	5193536	94.21	375.35	1244
SAMPLE_46	4138582	966148	23.34	3165224	76.48	4009129	96.87	333.86	1405
SAMPLE_47	3538128	620712	17.54	2913541	82.35	3344114	94.52	281.04	1352
SAMPLE_48	4655008	1119190	24.04	3530653	75.85	4506732	96.81	370.56	1398
SAMPLE_49	4057396	851084	20.98	3202842	78.94	3912648	96.43	301.14	1317
SAMPLE_50	5203538	1297771	24.94	3899184	74.93	5037737	96.81	406.83	1322
SAMPLE_51	4869284	1268279	26.05	3595657	73.84	4708973	96.71	369.68	1425
SAMPLE_52	3748462	796364	21.25	2948236	78.65	3630872	96.86	315.76	1403
SAMPLE_53	4503968	1057702	23.48	3440000	76.38	4352802	96.64	355.35	1367
SAMPLE_54	1847982	294654	15.94	1550856	83.92	1786735	96.69	168.79	1343
SAMPLE_55	4747334	1181833	24.89	3558987	74.97	4594479	96.78	358.84	1285
SAMPLE_56	5811628	1595324	27.45	4208612	72.42	5626443	96.81	433.62	1419
SAMPLE_57	2693326	458770	17.03	2231811	82.86	2602974	96.65	237.62	1351
SAMPLE_58	3412974	692539	20.29	2715589	79.57	3293492	96.50	285.83	1283
SAMPLE_59	3538404	669174	18.91	2865978	81.00	3416385	96.55	279.16	1230
SAMPLE_60	5020500	1332814	26.55	3682305	73.35	4867239	96.95	380.88	1303
SAMPLE_61	4798092	1180693	24.61	3608026	75.20	4628447	96.46	359.22	1386
SAMPLE_62	4089686	880592	21.53	3204073	78.35	3945953	96.49	318.64	1340
SAMPLE_63	4095102	935844	22.85	3153206	77.00	3964662	96.81	334.22	1291
SAMPLE_64	5846254	1461675	25.00	4376723	74.86	5652672	96.69	428.66	1288
SAMPLE_65	5366774	1425054	26.55	3933705	73.30	5195364	96.81	409.48	1362
SAMPLE_66	2328898	341598	14.67	1984426	85.21	2251353	96.67	208.85	1296
SAMPLE_67	6109638	1701443	27.85	4401148	72.04	5903595	96.63	455.12	1316
SAMPLE_68	4722916	1144064	24.22	3573003	75.65	4558605	96.52	364.47	1358
SAMPLE_69	6218248	1779750	28.62	4432586	71.28	5866692	94.35	418.83	1207
SAMPLE_70	4310554	882439	20.47	3424267	79.44	4089256	94.87	335.73	1347
SAMPLE_71	3937946	753770	19.14	3180169	80.76	3730536	94.73	312.18	1346
SAMPLE_72	4986154	1284257	25.76	3698744	74.18	4707392	94.41	352.87	1314
SAMPLE_73	4061440	777417	19.14	3279642	80.75	3841684	94.59	320.33	1419
SAMPLE_74	4405010	1063782	24.15	3337005	75.75	4162780	94.50	319.51	1263
SAMPLE_75	5071674	1341029	26.44	3725592	73.46	4766355	93.98	352.68	1234
SAMPLE_76	4143456	753477	18.18	3384536	81.68	3976468	95.97	309.01	1346
SAMPLE_77	5138396	1201933	23.39	3930057	76.48	4953834	96.41	377.95	1279
SAMPLE_78	4782692	1027324	21.48	3749082	78.39	4611111	96.41	360.89	1331
SAMPLE_79	4838792	946789	19.57	3886423	80.32	4631115	95.71	343.38	1354

<b>SAMPLE_80</b>	4014524	805358	20.06	3204160	79.81	3868949	96.37	317.23	1354
<b>SAMPLE_81</b>	5525118	1478170	26.75	4040892	73.14	5349764	96.83	417.73	1320
<b>SAMPLE_82</b>	3579440	644938	18.02	2930753	81.88	3376713	94.34	288.06	1412
<b>SAMPLE_83</b>	3252646	533731	16.41	2716705	83.52	3070473	94.40	258.97	1387
<b>SAMPLE_84</b>	5332234	1431476	26.85	3895773	73.06	5043888	94.59	371.97	1310

The columns contain the following information: (1) SampleID, (2) total count of the sequenced reads, (3) number of reads marked as duplicates, (4) duplicates as a proportion of the total number of sequenced reads, (5) number of mapped reads without duplicates, (6) proportion of non-duplicated reads in the total number of sequenced reads, (7) number of reads mapping to the reference genome with MAPQ at least 40, (8) proportion of the MAPQ40 and above reads in the total number of sequenced reads, (9) mean depth of coverage in the targeted regions, (10) number of called small variants.