

S1\_Table. Summary statistics for Ion Torrent sequencing across the ePAR

individual			bases	>=Q20 bases	>=Q20 bases, %	reads	mapped reads	% of reads mapped
NA10847	female	da	43,979,848	39,307,954	89.38	193,337	179,808	93.00
NA12146	ePAR	fa	71,728,327	64,684,810	90.18	290,574	271,962	93.59
NA12239	female	mo	86,728,887	77,742,730	89.64	359,398	334,770	93.15
man 20	ePAR		78,320,031	70,679,322	90.24	316,557	306,178	96.72
man 53	ePAR		72,216,306	65,159,010	90.23	284,570	279,360	98.17
B1 *	ePAR	br	66,763,804	59,929,318	89.76	264,729	232,026	87.65
P1 *	ePAR	pa	66,218,114	59,721,814	90.19	262,450	235,900	89.88
F1 *	ePAR	fa	80,785,440	72,804,184	90.12	336,278	307,895	91.56
P2 *	ePAR	pa	78,977,048	70,732,103	89.56	325,234	310,205	95.38
F2 *	ePAR	fa	36,711,977	32,891,346	89.59	163,180	157,325	96.41
P3 *	ePAR	pa	149,637,283	134,452,533	89.85	810,574	753,664	92.98
F3 *	ePAR	fa	84,790,584	75,859,154	89.47	345,952	320,717	92.71
P4 *	ePAR	pa	85,253,719	76,863,868	90.16	355,542	347,721	97.80
F4 *	ePAR	fa	85,659,925	77,183,601	90.10	343,893	335,654	97.60
P5 *	ePAR	pa	26,747,753	24,051,513	89.92	118,408	117,148	98.94
F5 *	ePAR	fa	85,472,838	76,732,964	89.77	346,425	321,536	92.82
P6 *	ePAR	pa	113,795,421	101,829,085	89.48	476,539	457,852	96.08
F6 *	ePAR	fa	75,312,769	66,838,973	88.75	300,658	276,314	91.90
6689_01	ePAR		83,255,363	75,124,261	90.23	333,594	321,682	96.43
333	ePAR		72,691,290	64,267,808	88.41	320,268	309,115	96.52
		<i>min</i>	<b>26,747,753</b>	<b>24,051,513</b>	<b>88.41</b>	<b>118,408</b>	<b>117,148</b>	<b>87.65</b>
		<i>max</i>	<b>149,637,283</b>	<b>134,452,533</b>	<b>90.24</b>	<b>810,574</b>	<b>753,664</b>	<b>98.94</b>
		<i>mean</i>	<b>77,252,336</b>	<b>69,342,818</b>	<b>89.75</b>	<b>327,408</b>	<b>308,842</b>	<b>94.46</b>

\* from Mensah *et al.* 2014 PLoS genetics, 10: e1004578.  
da = daughter, fa=father, mo=mother, br=brother, pa=patient