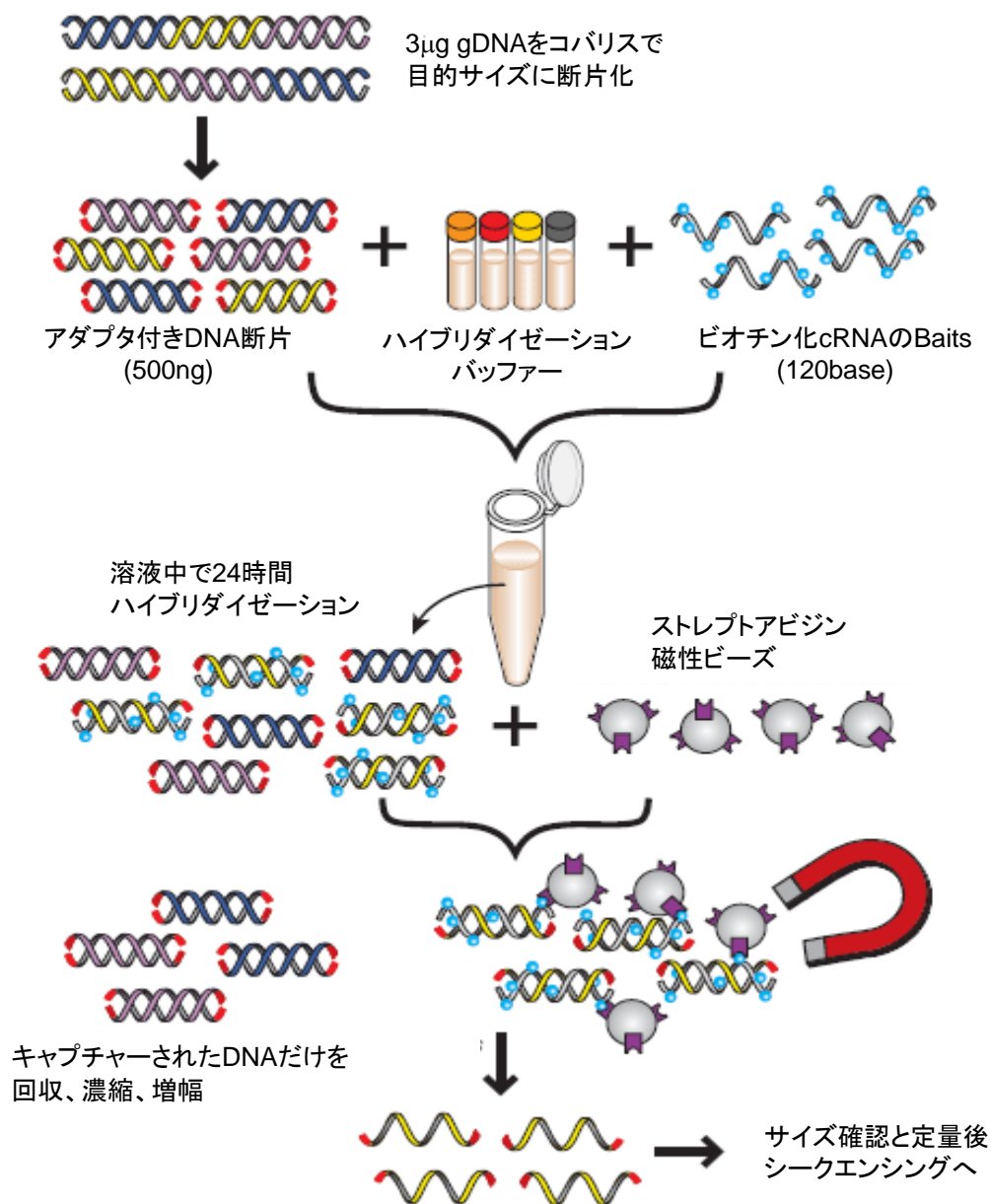


★ 5. リシーケンシング解析



Sure Select Human All Exon 50Mb GENCODE and Sanger (also includes CCDS and Broad defined v2 content)	
CCDS (Sept 2009)	99.86%
CNV (Mar 2010)	30.62%
Ensembl*	75.24%
miRNA (miRBase 14)	92.78%
GenBank*	90.74%
RefSeq Genes*	96.47%
RefSeq Transcripts*	97.50%
Total	50Mb
Developed with	Sanger
	*6/16/2010

Human All Exon V4 (51Mb) + UTRs (20Mb)

8,263 lincRNA Exon
HGNC 979 non-codingRNA Exon
(The HUGO Gene Nomenclature Committee)
(9.3M)

Illumina HiSeq 2000
75base, paired-end sequencing

★5. リシークエンシング解析

Whole Exome Sequencing

シーケンス検体数: 97

Baits: Exon 50M + UTR + LincRNA, 約80Mb

<参考: 平均>

#Cluster: 87M, PF%: 91.1, %ident: 16.2, Residue: 60M

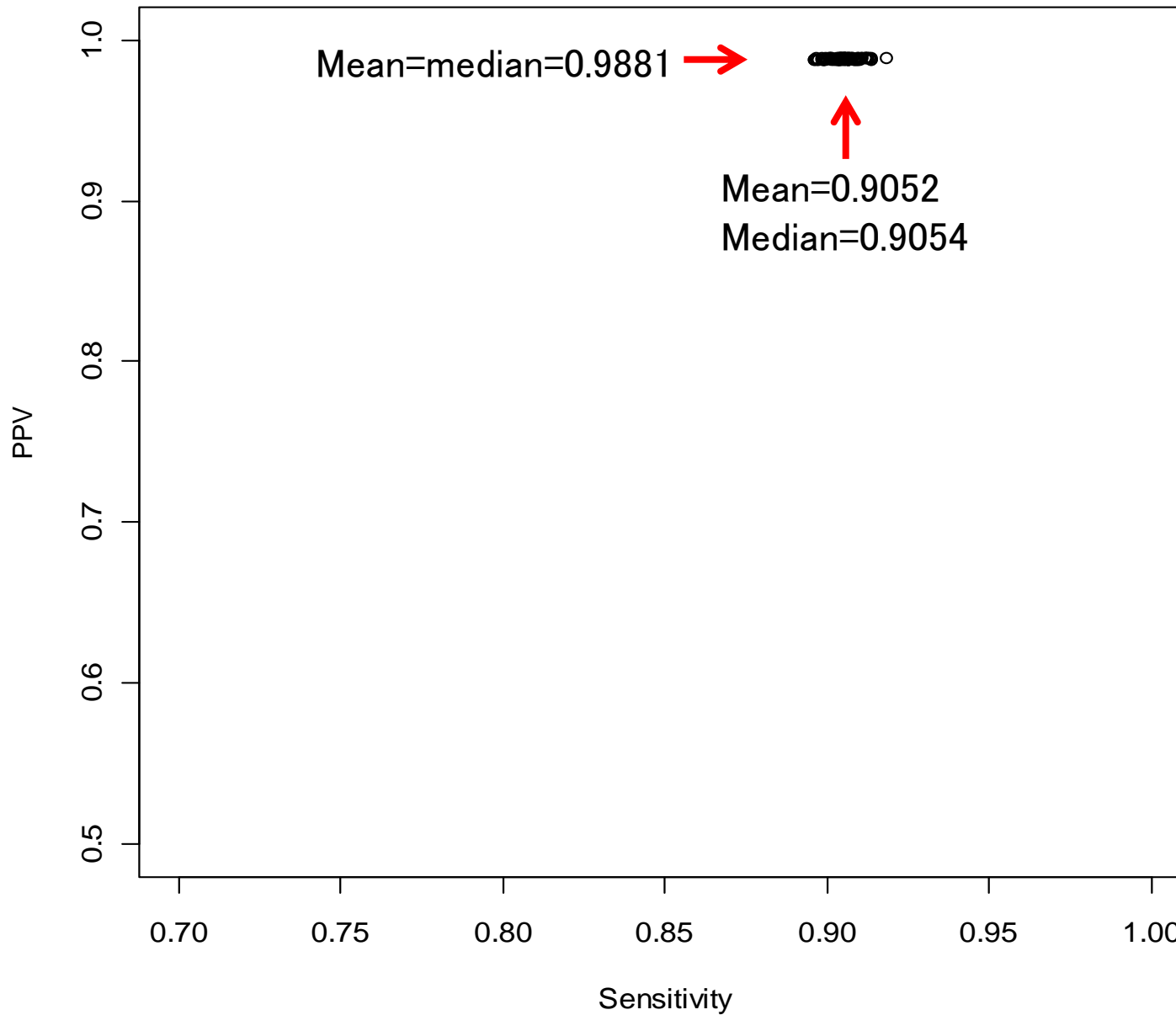
Mean depth on baits: 80.7

(on 50M+UTR: 76.7, on Linc: 114.1)

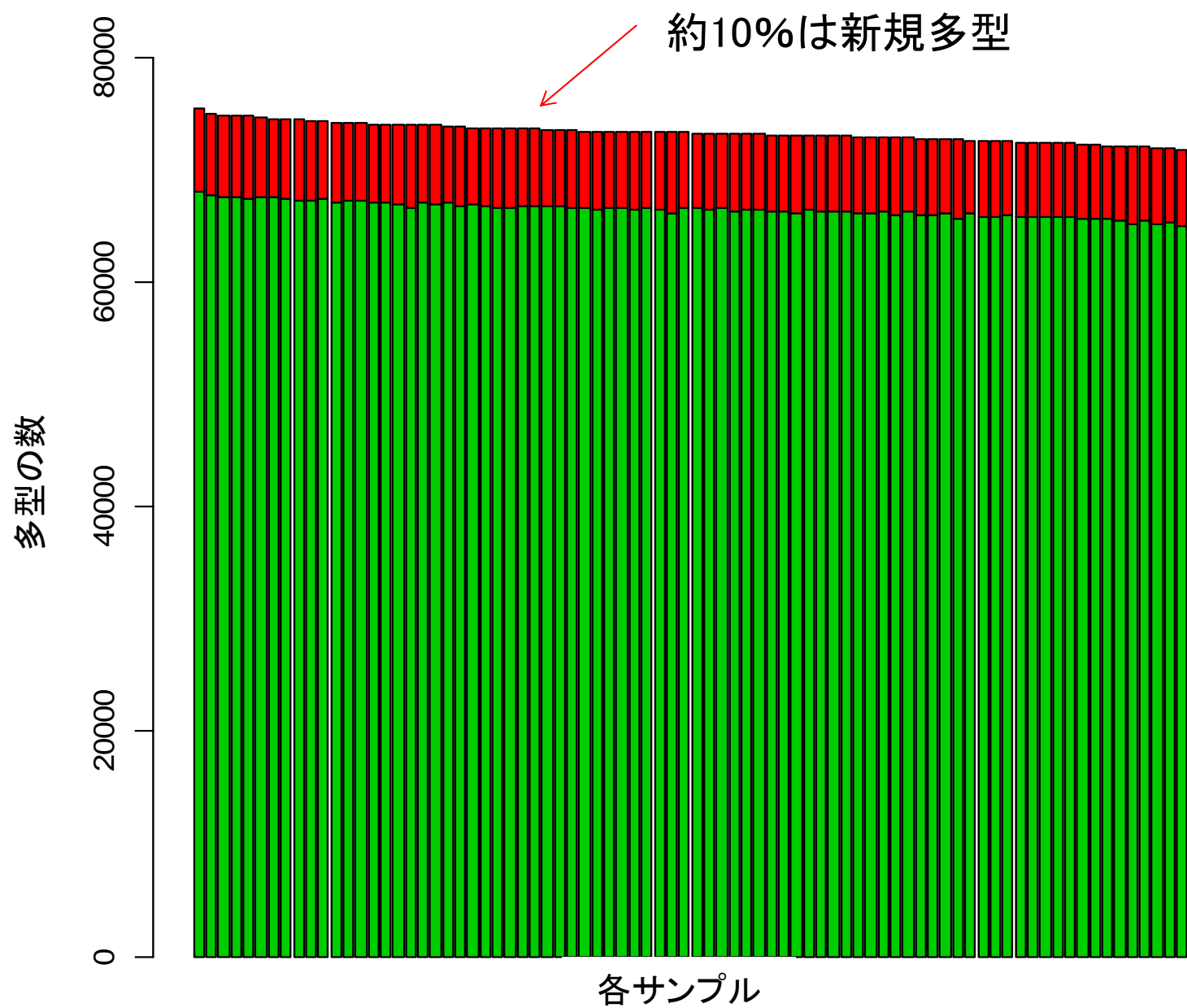
Sample	Reads	PF	PF%	#identical	%ident	#paired-read	residue	Mean depth
022481-1A1	65,857,029	59,874,108	89.56	10,828,267	18.1	49,045,841	42,784,295	57.9
022481-1A2	72,633,165	65,457,525	90.12	13,789,323	21.1	51,668,202	44,081,304	59.8
022481-1A3	71,310,637	63,763,158	89.44	11,262,822	17.7	52,520,336	45,899,172	61.9
022481-1A4	72,295,294	65,061,635	90.02	9,155,219	14.1	55,926,416	50,439,566	68.2
022481-1A6	67,346,307	60,916,139	90.30	9,051,956	14.8	51,784,180	46,206,993	62.7
022481-1A7	69,674,676	62,784,345	90.11	9,226,025	14.7	53,558,320	48,037,228	64.8
022481-1A10	70,709,588	63,788,286	90.23	7,710,221	12.1	56,088,065	51,444,427	69.6
022481-1A8	62,547,653	58,713,187	90.67	10,462,818	18.4	46,250,349	40,592,277	54.6
022481-1A9	68,272,332	62,865,223	90.58	10,795,447	17.2	51,295,876	46,006,162	62.2
022481-1A11	69,666,206	63,017,359	90.46	11,133,509	17.7	51,833,850	46,045,064	62.1
022481-1A12	65,526,445	59,363,781	90.64	12,789,998	21.5	46,603,783	40,205,121	54.4
022481-1B1	71,287,499	64,320,307	90.23	10,765,638	18.7	53,554,669	47,644,658	63.8
022481-1B2	72,892,156	65,361,758	89.70	7,989,811	12.2	57,401,147	51,696,181	69.8
022481-1B3	69,985,425	63,216,663	90.33	8,523,804	12.5	54,892,858	48,809,887	65.8
022481-1B4	70,346,638	62,895,228	89.41	7,344,999	11.7	55,550,529	49,974,311	68.3
022481-1B5	70,434,958	62,862,803	89.42	7,615,733	12.1	55,367,070	50,202,281	67.6
022481-1B6	71,099,586	63,732,799	89.64	6,837,236	10.7	56,895,599	52,156,518	70.0
022481-1B7	67,746,893	60,923,893	89.96	5,791,004	9.5	54,901,798	50,794,398	68.1
022481-1B10	71,460,756	63,328,897	88.62	5,573,908	8.8	57,754,989	53,659,579	71.4
022481-1B9	74,732,113	66,515,379	89.01	6,303,783	9.5	60,211,598	55,746,674	74.8
022481-1B8	67,102,546	59,968,723	89.13	6,451,587	10.8	53,357,136	48,888,923	65.6
022481-1B11	72,211,412	64,920,041	89.89	7,266,028	11.2	56,999,413	51,753,842	69.1
022481-1B12	67,869,291	60,545,661	89.21	6,880,377	14.3	51,965,284	45,925,768	61.2
022481-1C1	67,059,728	59,680,165	89.00	6,472,423	10.8	53,207,742	48,495,943	64.7
0307A1-1C2	71,941,423	68,009,966	94.54	11,212,206	18.8	56,797,760	53,084,931	71.7
0307A1-1C3	71,472,886	67,847,207	94.83	7,672,422	12.2	60,174,785	56,502,363	78.2
0307A1-1C4	70,742,825	67,212,423	94.88	7,208,450	14.5	60,534,375	56,937,492	79.1
0307A1-1C6	79,560,633	74,726,187	93.92	8,379,049	11.7	66,347,138	62,964,674	81.8
0307A1-1C7	83,739,049	78,481,109	93.72	7,226,207	10.7	71,262,842	67,036,635	82.5
0307A1-1C5	72,226,207	67,823,880	93.91	67,143,113	94.88	60,450,484	56,923,665	78.2
0307A1-1C10	67,143,113	62,843,696	93.75	76,204,653	112.3	72,379,484	68,949,948	94.9
0307A1-1C8	67,284,696	63,923,665	95.02	74,223,246	110.1	70,118,181	66,473,947	90.7
0307A1-1C9	76,204,653	72,379,484	94.98	68,063,993	94.56	64,309,819	60,450,484	78.2
0307A1-1C11	74,223,246	70,118,181	94.47	72,129,525	94.46	73,887,631	69,354,439	93.86
0307A1-1C12	68,063,993	64,309,819	94.56	76,387,238	112.3	71,960,572	68,146,916	94.1
0307A1-1D1	76,357,442	72,129,525	94.46	74,836,255	98.34	75,974,614	71,228,558	93.75
0307A1-1D2	73,887,631	69,354,439	93.86	76,815,895	102.3	72,077,330	68,888,900	91.8
0307A1-1D3	76,387,238	71,960,572	94.16	81,217,741	115.2	76,158,227	70,501,688	92.27
0307A1-1D4	74,836,255	70,501,688	93.27	79,036,523	104.7	73,810,047	69,340,820	93.39
0307A1-1D5	75,974,614	71,228,558	93.75	81,997,673	106.2	76,630,558	72,379,484	93.45
0307A1-1D6	76,815,895	72,077,330	93.88	79,449,926	106.2	74,350,667	70,501,688	93.58
0307A1-1D7	81,217,741	76,158,227	93.77	79,449,926	106.2	74,350,667	70,501,688	93.58
0307A1-1D8	79,036,523	73,810,047	93.39	79,449,926	106.2	74,350,667	70,501,688	93.58
0307A1-1D9	81,997,673	76,630,558	93.45	81,997,673	106.2	76,630,558	72,379,484	93.45
0307A1-1D10	79,449,926	74,350,667	93.58	81,997,673	106.2	76,630,558	72,379,484	93.45
0307A1-1D11	79,449,926	74,350,667	93.58	81,997,673	106.2	76,630,558	72,379,484	93.45
0307A1-1D12	73,076,071	68,445,239	93.66	81,997,673	106.2	76,630,558	72,379,484	93.45
0307A1-1E1	86,784,371	81,133,313	93.49	86,784,371	106.2	81,133,313	76,630,558	93.45
0427B1-1H8	102,562,335	93,132,458	90.81	102,562,335	106.2	93,132,458	88,938,061	92.04
0427B1-1H9	87,813,545	79,709,661	90.77	87,813,545	106.2	79,709,661	74,350,667	93.58
0427B2-1E3	88,938,061	81,860,876	92.04	88,938,061	106.2	81,860,876	76,630,558	93.45
0427B2-1E4	89,079,443	82,711,249	92.85	89,079,443	106.2	82,711,249	76,630,558	93.45
0427B3-1E12	89,241,712	81,719,929	91.57	89,241,712	106.2	81,719,929	76,630,558	93.45
0427B3-1E6	90,240,560	82,820,707	91.78	90,240,560	106.2	82,820,707	76,630,558	93.45
0427B4-1F1	83,772,140	76,843,368	91.73	83,772,140	106.2	76,843,368	71,228,558	93.75
0427B4-1F2	93,893,035	85,910,253	91.50	93,893,035	106.2	85,910,253	81,133,313	93.49
0427B5-1F3	89,214,640	81,555,629	91.42	89,214,640	106.2	81,555,629	76,630,558	93.45
0427B5-1F5	91,368,275	83,205,292	91.07	91,368,275	106.2	83,205,292	76,630,558	93.45

Sample	#cluster	#PF	PF%	#identical	%ident	#paired-read	residue	Mean depth
0307A6-1D6	76,915,895	72,207,730	93.88	8,914,100	12.3	63,293,630	59,701,213	80.9
0307A6-1D7	81,217,741	76,158,227	93.77	7,817,531	10.3	68,340,696	65,001,667	87.5
0307A7-1D10	80,430,449	75,015,858	93.27	7,700,766	10.3	67,315,092	63,810,049	86.4
0307A7-1D8	79,036,523	73,810,047	93.39	9,340,820	12.7	64,469,227	60,334,505	79.8
0307A7-1D9	81,997,673	76,630,558	93.45	8,085,656	10.6	68,544,902	64,901,570	87.5
0307A8-1D11	79,449,926	74,350,667	93.58	10,855,408	14.6	63,495,259	59,438,612	80.2
0307A8-1D12	73,076,071	68,445,239	93.66	9,507,588	13.9	58,937,651	55,280,347	75.0
0307A8-1E1	86,784,371	81,133,313	93.49	13,048,111	16.1	68,085,202	63,300,744	85.8
0427B1-1H8	102,562,335	93,132,458	90.81	14,863,423	16.0	78,269,035	73,625,723	98.7
0427B1-1H9	87,813,545	79,709,661	90.77	7,654,800	9.6	72,054,861	69,403,885	93.5
0427B2-1E3	88,938,061	81,860,876	92.04	16,791,362	20.5	65,069,514	60,724,162	81.0
0427B2-1E4	89,079,443	82,711,249	92.85	18,249,797	22.1	64,461,452	59,985,158	81.2
0427B3-1E12	89,241,712	81,719,929	91.57	13,865,451	17.0	67,854,478	63,881,815	86.7
0427B3-1E6	90,240,560	82,820,707	91.78	18,775,501	22.7	64,045,206	58,937,192	80.1
0427B4-1F1	83,772,140	76,843,368	91.73	13,602,077	17.7	63,241,291	59,348,860	80.7
0427B4-1F2	93,893,035	85,910,253	91.50	15,137,139	17.6	70,773,114	66,396,875	90.0
0427B5-1F3	89,214,640	81,555,629	91.42	18,534,748	22.7	63,020,881	58,136,483	79.0
0427B5-1F5	91,368,275	83,205,292	91.07	16,557,094	19.9	66,648,198	62,048,305	84.4

★ HumanOmni2.5-8 BeadChipのデータとの一致率（感度/PPV：サンプルごと）



★検出された多型 (サンプルごと)



★Feasibility Study開始8ヶ月時点での主な進捗状況（B. ゲノム解析）

1. ゲノムコホート研究の枠組みの中で行う多因子疾患の罹患リスク要因分析の方法論について実証的に検討するためのデータセットサンプルの確保が進んだ。
 - ✓ 疾患の例として大腸がんを取り上げ、生活習慣・バイオマーカー・複数の候補遺伝素因のデータセットの収集を進めた。
2. 1990-1994年に採血され、2006-2007年にDNA抽出され、現実的な方法（普及型-80°C冷凍庫）で長期保管されていた末梢血ゲノム試料に対して、最新のSNP遺伝子型判別アレイ技術により、新鮮に調製したDNAと同等の品質のゲノム網羅的多型情報が得られることを確認。
3. 同様の長期保管ゲノム試料に対して、最新のbisulfite変換遺伝子型判別アレイ技術を用いれば、新鮮に調整したDNAと同等の品質のゲノム網羅的DNAメチル化情報が得られることを確認し、制御すべき系統誤差の特徴を把握、エピゲノム修飾の個人差を捕捉できる可能性を示唆した。
4. 同様の長期保管ゲノム試料に対して、全ゲノムシーケンスよりもwetの実験操作としては工程が多く煩雑な全エクソームシーケンス情報も得られることを確認。
5. 「ヒトゲノム・遺伝子解析研究に関する倫理指針」や米国のCommon Rule（連邦法45CFR46）の見直しの動きについて、特に包括的同意（General Consent）許容に向けた動き（批判・反対を含む）に関して、様々な確度から検討を進め、FS終了時の「大規模ゲノムコホート推進体制」への提言とりまとめについての構想を開始。