

Supplementary file 2:overview of genome positions that did not comply with quality requirements per method

tNGS

Chr	Start	End	Gene
1	24135767	24135960	HMGCL
1	24135964	24135964	HMGCL
1	41445871	41445871	CTPS1
1	41466475	41466494	CTPS1
1	41466497	41466497	CTPS1
1	45975488	45975488	MMACHC
1	45975490	45975631	MMACHC
1	45975740	45975741	MMACHC
1	45977851	45978113	MMACHC
1	53679365	53679529	CPT2
1	64120651	64120690	PGM1
1	64120693	64120695	PGM1
1	76190049	76190123	ACADM
1	76216469	76216470	ACADM
1	76216546	76216586	ACADM
1	76220608	76220634	ACADM
1	76220722	76220784	ACADM
1	76229263	76229374	ACADM
1	100654908	100654908	DBT
1	100657306	100657331	DBT
1	100659255	100659257	DBT
1	100659265	100659269	DBT
1	100659273	100659320	DBT
1	100659960	100660192	DBT
1	100662091	100662092	DBT
1	100665207	100665207	DBT
1	100665209	100665395	DBT
1	100665914	100666120	DBT
1	100684836	100684842	DBT
1	100684846	100684846	DBT
1	100684849	100685049	DBT
1	100685826	100685826	DBT
1	120296000	120296019	HMGCS2
2	26457949	26458109	HADHA
2	26458112	26458112	HADHA
2	27462862	27463038	CAD
2	27463040	27463041	CAD
2	71336804	71336837	MCEE
2	71343893	71343897	MCEE
2	211539006	211539136	CPS1
2	211539207	211539208	CPS1
2	228548610	228548612	SLC19A3
2	228548621	228548621	SLC19A3
2	228548624	228548714	SLC19A3
2	228548719	228548719	SLC19A3

2	228548723	228548723 SLC19A3
2	228549501	228549596 SLC19A3
2	228550644	228550828 SLC19A3
2	228550830	228550834 SLC19A3
2	228560801	228560801 SLC19A3
2	228566352	228566352 SLC19A3
2	228571305	228571317 SLC19A3
2	228571319	228571319 SLC19A3
2	228571331	228571370 SLC19A3
2	241819023	241819024 AGXT
2	241819030	241819143 AGXT
2	241819923	241819929 AGXT
3	15691945	15691946 BTD
3	15692380	15692492 BTD
3	15693693	15693693 BTD
3	15693696	15693707 BTD
3	15693709	15693709 BTD
3	15753330	15753385 BTD
3	15753388	15753392 BTD
3	15753396	15753397 BTD
3	135971230	135971241 PCCB
3	135978982	135979019 PCCB
3	135979022	135979023 PCCB
3	136053694	136053983 PCCB
3	136055803	136055806 PCCB
3	164696676	164696803 SI
3	164714561	164714570 SI
3	164751174	164751174 SI
3	170727737	170727755 SLC2A2
3	182833424	182833599 MCCC1
4	993598	993598 IDUA
4	993603	993624 IDUA
4	996955	997070 IDUA
4	17488508	17488508 QDPR
4	17513758	17513758 QDPR
4	159601032	159601263 ETFDH
4	159630172	159630193 ETFDH
4	159630196	159630196 ETFDH
4	159630198	159630198 ETFDH
4	159630201	159630202 ETFDH
4	159630206	159630206 ETFDH
4	159630425	159630428 ETFDH
4	159630432	159630630 ETFDH
5	41739828	41739833 OXCT1
5	70932287	70932548 MCCC2
5	70953392	70953392 MCCC2
5	70953456	70953505 MCCC2
5	125878955	125878955 ALDH7A1
5	125900040	125900056 ALDH7A1
5	125907052	125907054 ALDH7A1

5	125929033	125929041 ALDH7A1
5	131713420	131713421 SLC22A5
5	131715273	131715392 SLC22A5
5	131730053	131730053 SLC22A5
6	81053717	81053719 BCKDHB
6	81053784	81054305 BCKDHB
6	81055961	81055962 BCKDHB
6	81055966	81055972 BCKDHB
6	81055979	81055997 BCKDHB
7	44182802	44182820 GCK
7	44187412	44187442 GCK
7	137783050	137783066 AKR1D1
7	137802498	137802706 AKR1D1
7	144316264	144316275 TPK1
7	144316282	144316300 TPK1
8	63961102	63961159 TTPA
8	63972316	63972332 TTPA
8	63972336	63972344 TTPA
8	63972373	63972374 TTPA
8	63972377	63972384 TTPA
8	63972398	63972455 TTPA
8	63973233	63973235 TTPA
8	63978659	63978659 TTPA
8	144297961	144298018 GPIHBP1
9	35741307	35741502 GBA2
9	35749100	35749101 GBA2
9	35749132	35749146 GBA2
9	35749151	35749155 GBA2
9	35749202	35749235 GBA2
9	97365415	97365415 FBP1
9	97401801	97401810 FBP1
9	104147127	104147298 BAAT
9	133320084	133320189 ASS1
9	133320195	133320198 ASS1
9	133320205	133320207 ASS1
9	133320244	133320284 ASS1
9	133376649	133376671 ASS1
10	69587290	69587395 DNAJC12
10	69587397	69587397 DNAJC12
10	69587429	69587447 DNAJC12
10	88835782	88835790 GLUD1
10	126100238	126100240 OAT
10	126103628	126103651 OAT
11	2189922	2189966 TH
11	2191995	2192010 TH
11	68522950	68523502 CPT1A
11	68586145	68586170 CPT1A
11	68609233	68609298 CPT1A
11	68609306	68609309 CPT1A
11	68609368	68609409 CPT1A

11	107999758	107999938 ACAT1
11	108005143	108005185 ACAT1
11	112140594	112140594 PTS
11	118898436	118898436 SLC37A4
13	41363538	41363637 SLC25A15
13	41363731	41363734 SLC25A15
13	41363736	41363809 SLC25A15
13	41384345	41384504 SLC25A15
13	41385306	41385537 SLC25A15
13	100813228	100813229 PCCA
15	40711511	40712283 IVD
15	40712349	40712349 IVD
15	40713491	40713491 IVD
15	45665424	45665539 GATM
15	45670725	45670737 GATM
15	45670742	45670742 GATM
15	45670746	45670746 GATM
15	45670748	45670753 GATM
15	76518681	76518686 ETFA
15	76522835	76523000 ETFA
15	76576696	76576811 ETFA
15	76577136	76577278 ETFA
15	76602358	76602358 ETFA
15	76602360	76602415 ETFA
15	80444822	80444897 FAH
15	80445112	80445291 FAH
15	80445294	80445297 FAH
15	80448283	80448425 FAH
15	80448429	80448432 FAH
16	30999652	30999653 HSD3B7
16	30999660	30999939 HSD3B7
16	71601098	71601257 TAT
16	71611002	71611003 TAT
17	3564838	3564838 CTNS
17	3565388	3565535 CTNS
17	3565606	3565606 CTNS
17	7121025	7121027 ACADVL
17	7127900	7127900 ACADVL
17	7127905	7127966 ACADVL
17	26727722	26727722 SLC46A1
17	41064988	41064995 G6PC
17	73747665	73747748 GALK1
17	73759039	73759039 GALK1
19	1398155	1398594 GAMT
19	1401564	1401579 GAMT
19	13003070	13003107 GCDH
19	13003121	13003258 GCDH
19	40736733	40736906 AKT2
19	40743339	40743473 AKT2
19	40748831	40749009 AKT2

19	40749692	40749933 AKT2
19	40749964	40749966 AKT2
19	40774718	40775002 AKT2
19	40790946	40791009 AKT2
19	40791078	40791312 AKT2
19	41903574	41903574 BCKDHA
19	41924620	41924669 BCKDHA
19	45409607	45409614 APOE
19	45410124	45410360 APOE
19	45411199	45411219 APOE
19	45411780	45411783 APOE
19	45452269	45452269 APOC2
19	45452271	45452320 APOC2
19	45452327	45452329 APOC2
19	45452334	45452336 APOC2
19	51849086	51849189 ETFB
19	51851481	51851482 ETFB
19	51851489	51851505 ETFB
19	51851550	51851558 ETFB
19	51851563	51851642 ETFB
19	51858400	51859327 ETFB
19	51869595	51869682 ETFB
20	746424	746432 SLC52A3
20	746741	746742 SLC52A3
20	746764	746768 SLC52A3
20	746772	746798 SLC52A3
20	32889903	32890008 AHCY
20	32890011	32890013 AHCY
20	32890015	32890015 AHCY
20	32890017	32890018 AHCY
20	32890020	32890023 AHCY
20	43280384	43280409 ADA
21	38121747	38121747 HLCS
21	38302544	38302544 HLCS
21	38320323	38320597 HLCS
21	38338734	38338979 HLCS
21	44495977	44496065 CBS
21	44496475	44496488 CBS
21	44496491	44496512 CBS
22	51062090	51062394 ARSA
22	51062562	51062562 ARSA
22	51062565	51062753 ARSA
22	51063152	51063170 ARSA
22	51066570	51066617 ARSA
X	152991611	152991631 ABCD1

WES

Gene	Genomic location (GRCh37)	GENCODE transcript	Exon location in transcript
ARG1	Chr6:131,868,263-131,868,338	ENST00000672233.1_1	Exon 2 (from start) out of 8
BTB	Chr3:15,709,536-15,709,748	ENST00000673467.1_3	Exon 4 (till stop) out of 4

BTD	Chr3:15,713,117-15,713,140	ENST00000673620.1_5	Exon 4 (till stop) out of 4
BTD	Chr3:15,751,567-15,751,581	ENST00000672141.1_5	Exon 4 (till stop) out of 4
BTD	Chr3:15,751,906-15,751,932	ENST00000672760.1_3	Exon 4 (till stop) out of 4
BTD	Chr3:15,763,277-15,763,356	ENST00000672427.1_4	Exon 5 (till stop) out of 5
CA5A	Chr16:87,915,417-87,915,545	ENST00000649158.1_3	Exon 7 (till stop) out of 7
CTPS1	Chr1:41,469,653-41,469,786	ENST00000463285.7_3	Exon 13 (till stop) out of 13
FBP1	Chr9:97,400,625-97,400,692	ENST00000648117.1_3	Exon 1 (from start) out of 6
MMADHC	Chr2:150,428,744-150,428,845	ENST00000422782.2_3	Exon 7 out of 9
PGM1	Chr1:64,120,661-64,120,782	ENST00000650546.1_1	Exon 11 out of 12
SLC19A3	Chr2:228,550,866-228,550,885	ENST00000409287.5_3	Exon 4 (till stop) out of 4

WGS

Two genes with low coverage in males

ABCD1 chrX: 152,990,323-153,010,216 forward strand

OTC chrX: 38,211,798-38,280,703 forward strand

One gene with low coverage overall in both sexes (due to pseudogene)

CBS chr21: 44,473,301-44,497,053 reverse strand