

Annotation	Abbreviation	Source/URL	Assembly	Citation	Comment
Protein-coding gene annotation					
Coding exons	CDS	Gencode v12	GRCh37/hg19	Gencode [1]	-
Introns	intron	Gencode v12 and UCSC table browser (tracks: UCSC genes, RefSeq genes, Ensembl genes)	GRCh37/hg19	Gencode [1], Ensembl [2], RefSeq [3], UCSC [4]; [5]	Defined as intronic nucleotides which do not overlap any exon of a protein-coding transcript.
Intergenic	intergenic	Gencode v12 and UCSC table browser (tracks: UCSC genes, RefSeq genes, Ensembl genes)	GRCh37/hg19	Gencode [1], Ensembl [2], RefSeq [3], UCSC [4]; [5]	Defined as the complement of all known protein-coding transcripts.
UTRs	UTRs	Gencode v12	GRCh37/hg19	Gencode [1]	-
Non-coding gene annotation					
Long non-coding RNAs	lncRNAs (Gencode)	ftp://ftp.sanger.ac.uk/pub/gencode/release_12/gencode.v12.long_noncoding_RNAs.gtf.gz	GRCh37/hg19	Gencode [1]	The original set of long non-coding RNAs as annotated in Gencode was reduced to a set of <i>bona fide</i> non-coding RNAs without any evidence for functional short ORFs (see descriptions above).
Large intergenic non-coding RNAs	lincRNAs	http://www.broadinstitute.org/genome_bio/human_lincrnas/sites/default/files/lincRNA_catalog/lincRNAs_transcripts.bed	GRCh37/hg19	[6]	-
Transcripts of uncertain coding potential	TUCP	http://www.broadinstitute.org/genome_bio/human_lincrnas/sites/default/files/TUCP_transcripts_catalog/TUCP_transcripts.gtf	GRCh37/hg19	[6]	-
Chromatin associated RNAs	CARs	-	NCBI36/hg18	[7]	Mapped to GRCh37/hg19 using liftOver [5].

Annotation	Abbreviation	Source/URL	Assembly	Citation	Comment
lncRNAs	lncRNAdb	http://lncrnadb.com	-	[8]	Coordinates in GRCh37/hg19 have been derived by BLAT [9] with parameters <code>-trimHardA -minIdentity=95</code> .
Short RNAs	miRNAs, snoRNAs, scaRNAs	UCSC table browser (track: sno/miRNA)	GRCh37/hg19	[10, 11, 5]	-
Intronic non-coding RNAs	TINs, PINs	UCSC Genome Browser mirror for functional RNA (http://www.ncrna.org/global/cgi-bin/hgGateway)	NCBI36/hg18	[12]	Mapped to GRCh37/hg19 using liftOver [5]. The original set of human intronic non-coding RNAs [12] was reassessed according to gene annotation in hg19 (see descriptions above).
Regions of conserved secondary structure					
RNAZ	RNAZ	-	GRCh37/hg19	[13]	-
SISSIZ	SISSIZ	-	GRCh37/hg19	[13]	-
EvoFold	EvoFold	UCSC table browser (track: EvoFold)	GRCh37/hg19	[14, 5]	-
Regulation Tracks					
H3K4 trimethylation	H3K4me3	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	Chromatin-mark associated with promoter sites [16, 17]. Mapped to GRCh37/hg19 using liftOver [5].
CpG islands	CpG	UCSC table browser (track: CpG Islands)	GRCh37/hg19	[18, 5]	Associated with transcription start sites [19, 20].
DNaseI hypersensitive sites	DNaseI	UCSC table browser (track: DNaseI Clusters)	GRCh37/hg19	[15, 5]	Associated with transcription factor binding sites [17, 21].
Transcription factor binding sites (TFBs)	TFBs (Encode)	UCSC table browser (track: Txn Factor ChIP)	GRCh37/hg19	[15, 5]	Binding sites identified by ChIP-seq [15].
PoIII binding sites	POL-II	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	PoIII binding sites derived by ChIP-seq [15]. Mapped to GRCh37/hg19 using liftOver [5].

Annotation	Abbreviation	Source/URL	Assembly	Citation	Comment
H3K36 trimethylation	H3K36me3	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	Chromatin-mark associated with active regions of PolII transcripts [22]. Mapped to GRCh37/hg19 using liftOver [5].
H3K27 trimethylation	H3K27me3	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	Chromatin-mark associated with repressed regions of PolII transcripts [23]. Mapped to GRCh37/hg19 using liftOver [5].
H3K4 monomethylation	H3K4me1	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	Chromatin-mark associated with enhancer regions [24, 17]. Mapped to GRCh37/hg19 using liftOver [5].
H3K27 acetylation	H3K27ac	ftp://hgdownload.cse.ucsc.edu/goldenPath/hg18/encodeDCC/wgEncodeBroadChIPSeq/	NCBI36/hg18	[15, 5]	Chromatin-mark associated with enhancer and promoter sites [15, 25, 26]. Mapped to GRCh37/hg19 using liftOver [5].
Other					
Repeats	-	UCSC table browser (track: Masker)	GRCh37/hg19	[27, 5]	-
Genome gaps	-	UCSC table browser (track: Gap)	GRCh37/hg19	[5]	-

References

- [1] Harrow J, Frankish A, Gonzalez JM, Tapanari E, Diekhans M, et al. (2012) GENCODE: the reference human genome annotation for The ENCODE Project. *Genome Res* 22: 1760–1774.
- [2] Hubbard T, Barker D, Birney E, Cameron G, Chen Y, et al. (2002) The Ensembl genome database project. *Nucleic Acids Res* 30: 38–41.
- [3] Pruitt KD, Tatusova T, Maglott DR (2007) NCBI reference sequences (RefSeq): a curated non-redundant sequence database of genomes, transcripts and proteins. *Nucleic Acids Res* 35: D61–D65.
- [4] Meyer LR, Zweig AS, Hinrichs AS, Karolchik D, Kuhn RM, et al. (2013) The UCSC genome browser database: extensions and updates 2013. *Nucleic Acids Res* : D64-9.
- [5] Kuhn RM, Haussler D, Kent WJ (2012) The UCSC genome browser and associated tools. *Brief Bioinformatics* .
- [6] Cabili MN, Trapnell C, Goff L, Koziol M, Tazon-Vega B, et al. (2011) Integrative annotation of human large intergenic noncoding RNAs reveals global properties and specific subclasses. *Genes Dev* 25: 1915–1927.
- [7] Mondal T, Rasmussen M, Pandey GK, Isaksson A, Kanduri C (2010) Characterization of the RNA content of chromatin. *Genome Res* 20: 899–907.
- [8] Amaral P, Clark M, Gascoigne D, Dinger M, Mattick J (2011) lncRNADB: a reference database for long noncoding RNAs. *Nucleic Acids Res* 39: D146–D151.
- [9] Kent WJ (2002) BLAT—the BLAST-like alignment tool. *Genome Res* 12: 656–664.
- [10] Lestrade L, Weber MJ (2006) snoRNA-LBME-db, a comprehensive database of human H/ACA and C/D box snoRNAs. *Nucleic Acids Res* 34: D158–D162.
- [11] Griffiths-Jones S (2004) The microRNA Registry. *Nucleic Acids Res* 32: D109–D111.
- [12] Nakaya HI, Amaral PP, Louro R, Lopes A, Fachel AA, et al. (2007) Genome mapping and expression analyses of human intronic noncoding RNAs reveal tissue-specific patterns and enrichment in genes related to regulation of transcription. *Genome Biol* 8: R43.
- [13] Smith MA, Gesell T, Stadler PF, Mattick JS (2013) Widespread purifying selection on RNA structure in mammals. *Nucleic Acids Res* 41: 8220-8236.
- [14] Pedersen JS, Bejerano G, Siepel A, Rosenbloom K, Lindblad-Toh K, et al. (2006) Identification and classification of conserved RNA secondary structures in the human genome. *PLoS Comput Biol* 2: e33.
- [15] Birney E, Stamatoyannopoulos J, Dutta A, Guigó R, Gingeras T, et al. (2007) Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. *Nature* 447: 799–816.
- [16] Roh TY, Cuddapah S, Cui K, Zhao K (2006) The genomic landscape of histone modifications in human T cells. *Proc Natl Acad Sci U S A* 103: 15782–15787.

- [17] Bulger M, Groudine M (2010) Enhancers: the abundance and function of regulatory sequences beyond promoters. *Dev Biol* 339: 250–257.
- [18] Gardiner-Garden M, Frommer M (1987) CpG islands in vertebrate genomes. *J Mol Biol* 196: 261–282.
- [19] Deaton AM, Bird A (2011) CpG islands and the regulation of transcription. *Genes Dev* 25: 1010–1022.
- [20] Guenther MG, Levine SS, Boyer LA, Jaenisch R, Young RA (2007) A chromatin landmark and transcription initiation at most promoters in human cells. *Cell* 130: 77–88.
- [21] Xi H, Shulha HP, Lin JM, Vales TR, Fu Y, et al. (2007) Identification and characterization of cell type-specific and ubiquitous chromatin regulatory structures in the human genome. *PLoS Genet* 3: e136.
- [22] Mikkelsen TS, Ku M, Jaffe DB, Issac B, Lieberman E, et al. (2007) Genome-wide maps of chromatin state in pluripotent and lineage-committed cells. *Nature* 448: 553–560.
- [23] Barski A, Cuddapah S, Cui K, Roh TY, Schones DE, et al. (2007) High-resolution profiling of histone methylations in the human genome. *Cell* 129: 823–837.
- [24] Heintzman ND, Stuart RK, Hon G, Fu Y, Ching CW, et al. (2007) Distinct and predictive chromatin signatures of transcriptional promoters and enhancers in the human genome. *Nat Genet* 39: 311–318.
- [25] Terrenoire E, McRonald F, Halsall JA, Page P, Illingworth RS, et al. (2010) Immunostaining of modified histones defines high-level features of the human metaphase epigenome. *Genome Biol* 11: R110.
- [26] Shin JH, Li RW, Gao Y, Baldwin R 6th, Li Cj (2012) Genome-wide ChIP-seq mapping and analysis reveal butyrate-induced acetylation of H3K9 and H3K27 correlated with transcription activity in bovine cells. *Funct Integr Genomics* 12: 119–130.
- [27] Smit A, Hubley R, Green P (2010) RepeatMasker Open-3.0. URL <http://www.repeatmasker.org>.