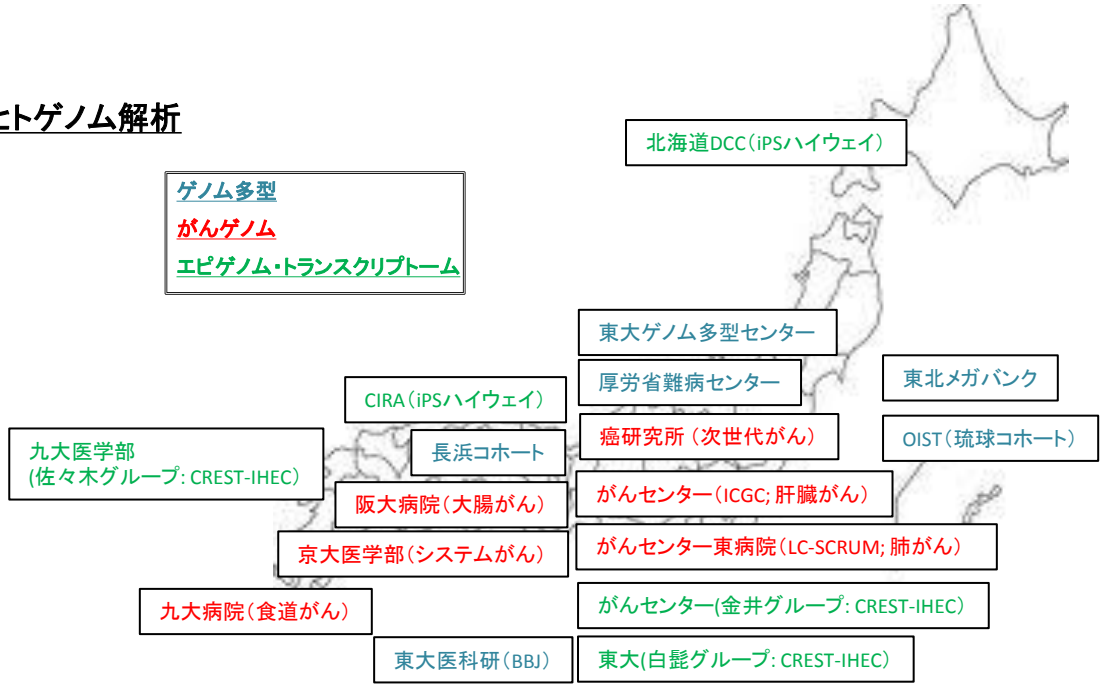
疾患ヒトゲノム変異の生物学的機能注釈を目指した多階層オミクスデータの統合

東京大学新領域創成科学研究科
菅野純夫

背景

全国に展開するヒトゲノム解析



ゲノムデータは急速に蓄積している

ヒトオミクスデータ推定蓄積量

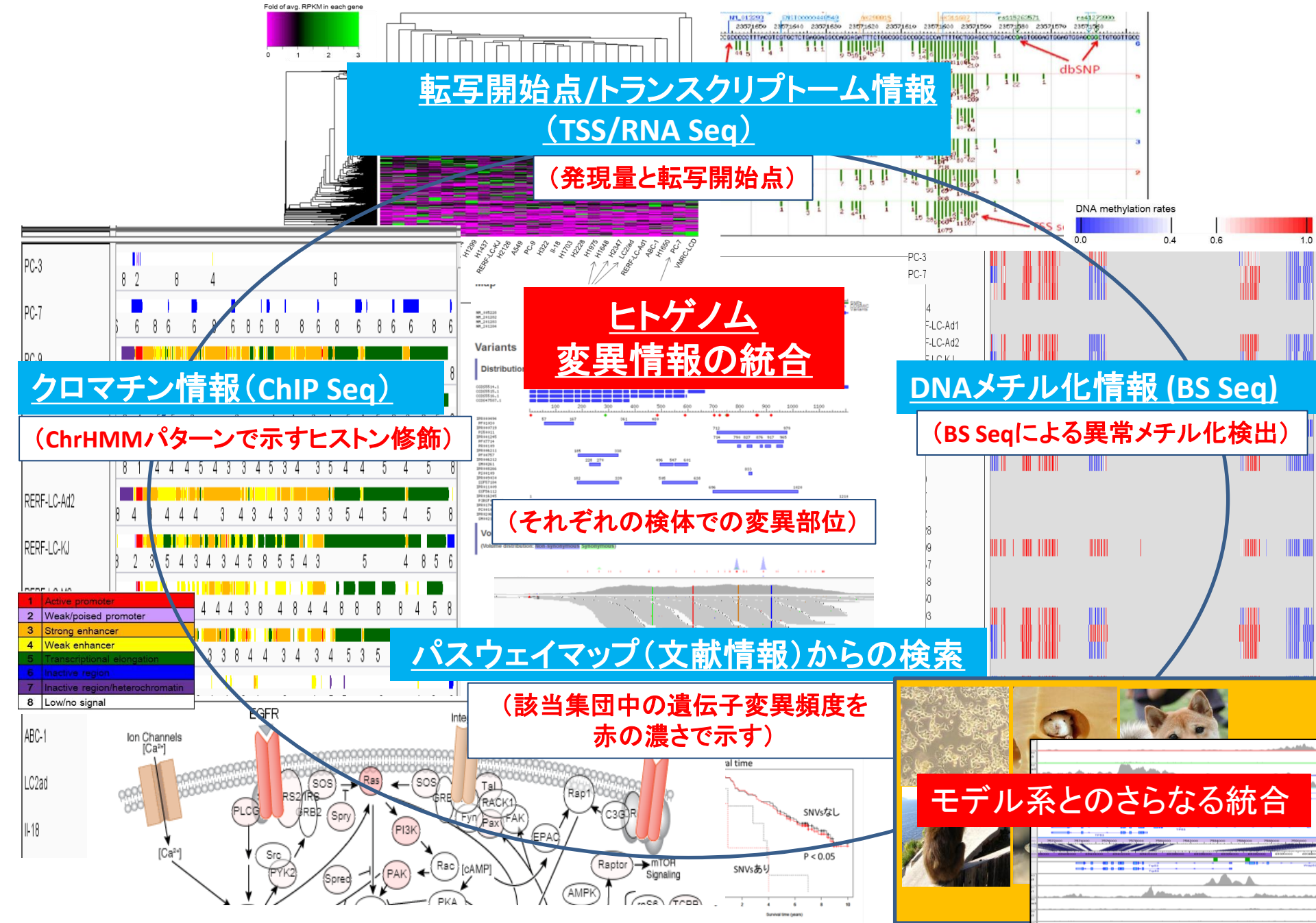
ゲノム多型(WGS/WES）: >2000人
がんゲノム(WGS/WES/Target Seq）: >1000症例
トランスクリプトーム(RNA Seq）: >1000例
エピゲノム(BS/ChIP Seq）: <100例

+培養細胞＋PDX＋モデル系: >5000例
+マウス等モデル生物: ???例

+個別研究者の蓄積するオミクス情報: ???例

構想

ヒト応用研究を志向したオミクス情報の統合（EGFR遺伝子を例に）



研究開発体制と役割

菅野：研究の統括/医科研スパコンの運用

鈴木：データの加工とデータベース設計

土原：ヒト疾患応用研究を志向した検索システムの実装

河野：データベースの構築、オントロジーの整備

＋研究員　3名
＋技術補佐員　2名
（雇用予定）

データ統合対象

	名称	件数（保有しているもの）	概要
1	ヒトがんゲノムデータ	300	肺腺・小細胞がん、食道がん、胃がん、大腸がんの全ゲノム/エキソームデータ
2	ヒト正常ゲノムデータ	500	申請者らの収集した正常日本人全ゲノム/エキソームデータの頻度情報
3	ヒトエピゲノム、トランスクリプトームデータ	300	肺腺がん、大腸がん：正常肝臓（予定）、正常大腸（予定）、iPS/ES細胞
4	ヒト培養細胞オミクスデータ	30 (300計測点)	肺腺がん、大腸がん（予定）、胃がん（予定）、初代培養正常細胞・PDX（予定）
5	マウスオミクスデータ	10 (400計測点)	正常組織、初代培養マクロファージ、B細胞等の免疫担当細胞（FIRST審良P）
6	＊その他のモデル動物データ	100 (500計測点)	「ゲノム支援」で収集、公開予定のデータ

用いるすべてのデータは申請者らがデータ産出、1次データ加工を実際に行ったものである。

EPOC 早期・探索臨床研究センター
LC-SCRUM-Japan
大阪大学大学院医学系研究科 外科系臨床医学専攻 外科学講座消化器外科学
九州大学病院別府病院
CREST, PRESTO 国際ヒトエピゲノムコンソーシアム 日本チーム
難病情報センター
文部科学省科学研究費新学術領域研究 ゲノム支援
最先端研究開発支援プログラム 免疫ダイナミズムの統合的理解と免疫制御法の確立

ゲノム支援
(DDBJより公開になり次第、収録許可をデータ所有者に打診)
■支援課題数
129
■ライブラリ内訳
ChIP :753
BS : 256
RNA :1705
smallRNA : 40
その他（DNA, TSSなど）: 164
■生物種内訳
human : 185
mouse :1179
その他 動物 : 790
植物・微生物・細菌 : 764

設備

メインサーバー：医科研ヒトゲノムセンタースパコンシステム

構成名	機種名	Rpeak (Gflops)	CPU	OS	CPUコア数	メモリ/node	ディスク容量
分散メモリ型 Type-A	Hitachi HA8000/RS210	150,141	Intel Xeon X5675 (3.06GHz)	Red Hat Enterprise Linux	144	144 GB	–
分散メモリ型 Type-B	Hitachi HA8000-tc/HT225		AMD Opteron 6276 (2.3GHz)		16,128	32 GB	–
共有メモリ型	SGI UV 100	1,361	Intel Xeon E7 8837 (2.66GHz)		128	2 TB	58 TB
ディスクアレイ	–	–	–	–	–	–	2 PB

既存のサーバーの利用：最低限の設備投資での計画遂行

＊モデル系から得られたオミクスデータに焦点。

特に培養細胞40種類：
ゲノム-エピゲノム-トランスクリプトームのオミクスデータが同一の材料から得られたもの

比較ゲノムブラウザーによりヒトゲノムと対応づけ → データ統合

＋各サイトに購入予定のサーバー

構成名	機種名	Rpeak (Gflops) /node	CPU	OS	CPUコア数 (1nodeあたり)	メモリ/node	ディスク容量/node
大規模データ解析サーバ	DELL PowerEdge R720XD	256	インテル® Xeon® プロセッサー E5-2650 (2.00GHz、20M キャッシュ、8.0GT/s QPI、ターボ、8C、95W) x2	RedHat Enterprise Linux	16core	128GB	27TB

平成26年度の成果

・データベースの公開、論文化
（ヒト培養細胞の多階層オミクスデータに焦点）

・ヒトゲノム多型データの充実
（特に日本人5000人分）

- ToMMo
- PGx
- ExAc

データベースの公開

- DataBase of Transcriptional Start Sites -
DBTSS

Release 9.0 Updated (September 15 2014
Based on UCSC hg38, mm1

About this database

Welcome to **DBTSS (DataBase of Transcriptional Start Sites)**.

To support transcriptional regulation studies, we have constructed the DBTSS (DataBase of Transcriptional Start Sites), which represents exact positions of transcriptional start sites (TSSs) in the genome based on our unique experimentally validated TSS sequencing method, TSS-seq.

This database includes TSS data of a major part of human adult and embryonic tissues are covered. DBTSS now contains 491 million TSS tag sequences for collected from a total of 20 tissues and 7 cell cultures. We also integrated our newly generated RNA-seq data of subcellular- fractionated RNAs and ChIP-seq data of histone modifications, RNA polymerase II and several transcriptional regulatory factors in cultured cell lines. We also included recently accumulating external epigenomic data, such as chromatin map of the ENCODE project.

In this update, we further associated those TSS information with public and original SNV

News

- 15 Sep. 2014: New DBTSS opened.

Genome

Transcriptome

Epigenome

DNA methylation

Histone modifications

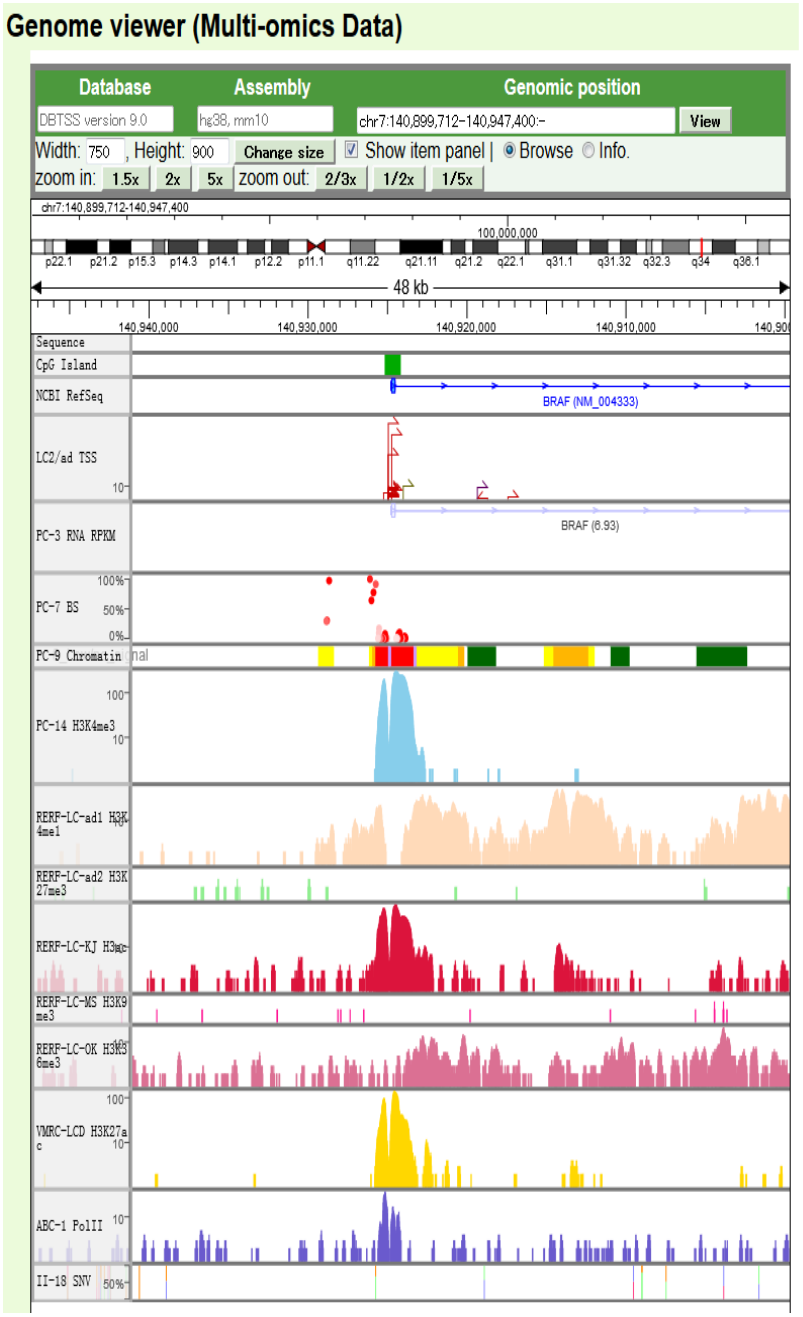
Database for multi-omics analysis

Further integration with

- Image data
- Clinical information

Search for novel/more useful biomarkers at “bulk” level

C



Viewer control

Gene model

TSS-Seq

RNA-Seq

BS-Seq

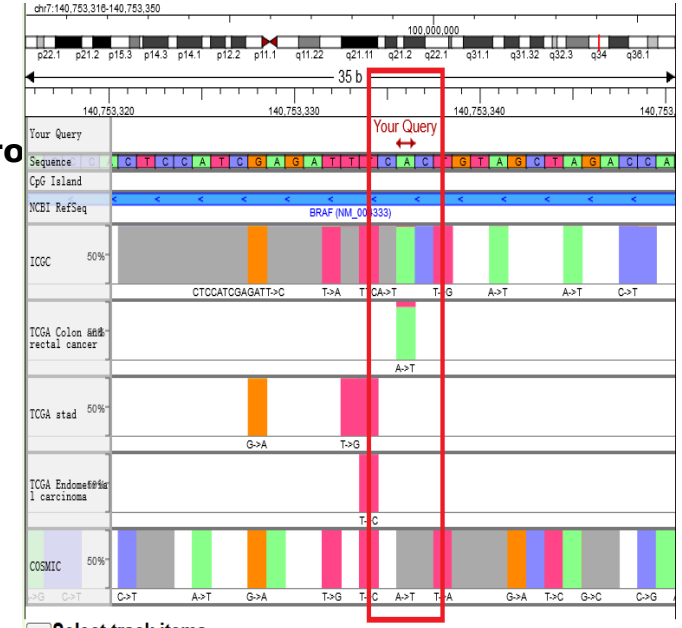
ChromHMM

ChIP-Seq

H3K4me3 H3K27me3 H3K9Ac H3K27Ac H3K4me H3K9me3 H3K36me3 Pol II

SNV

D



Database Search

Keyword Search

Species:
H. sapiens

Keyword:
NM_*

Search

Lift over:
hg38

chr1:99,950,000-100,050,000

Search

Human Chromatin Features

Search

Search from Genomic Position:
chr1:75,787,000

Search

Search from SNP (dbSNP rsID):
rs375229869

Search

Search from SNV (COSMIC: somatic mutation):
BRAF

Search

Search from SNV-enriched Gene in Cancers:
Lung adenocarcinoma ...

Search

MAPK/Erk in Growth and Differentiation

Cancer type: Lung adenocarcinoma 26 cell lines (each) | Cell: LC2/ad | Unit: RPKM

RPKM
RPKM log2 fold against normal cell
H3K4me3 (proximal)
H3K4me1 (distal)
H3K27ac (distal)
PolII (proximal)
H3K36me (gene body)
H3K27me3 (gene body)
H3K27me3 (distal)

Tint control:light deep

Coloring: RPKM: 0 255<

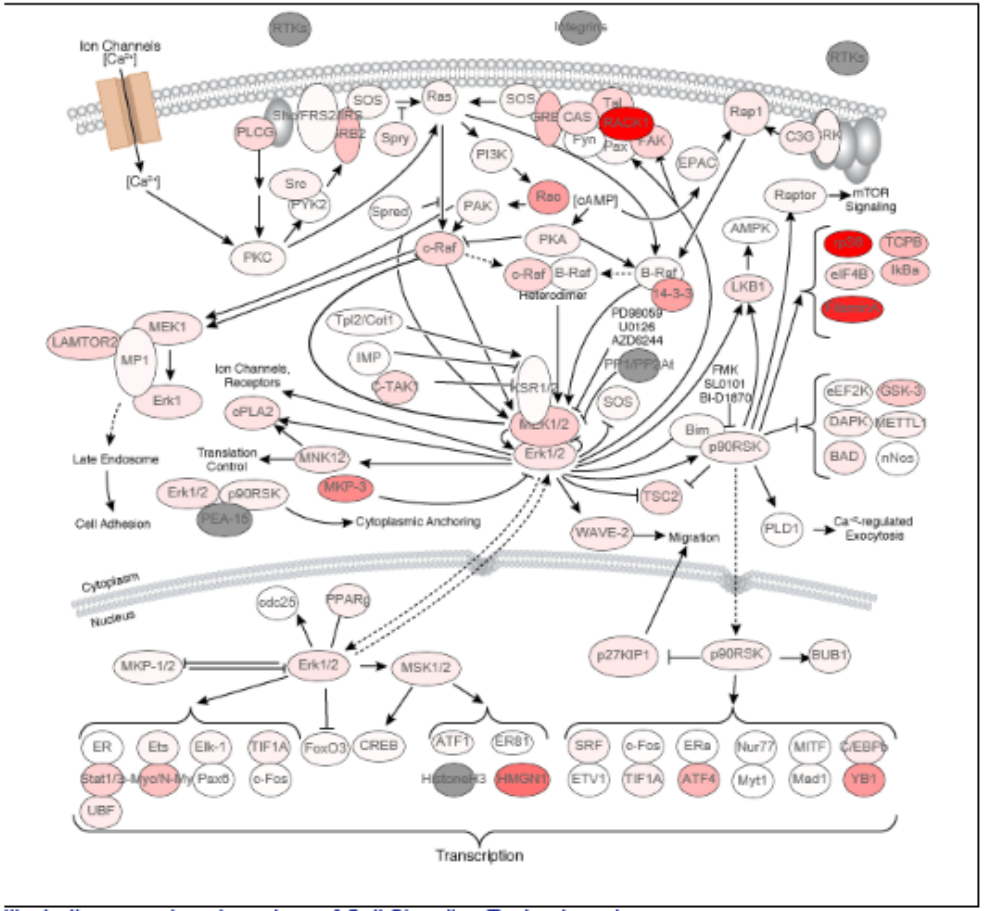


Illustration reproduced courtesy of Cell Signaling Technology, Inc.

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Nucleic Acids Research, 2014 1
doi: 10.1093/nar/gku1080

DBTSS as an integrative platform for transcriptome, epigenome and genome sequence variation data

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ABSTRACT

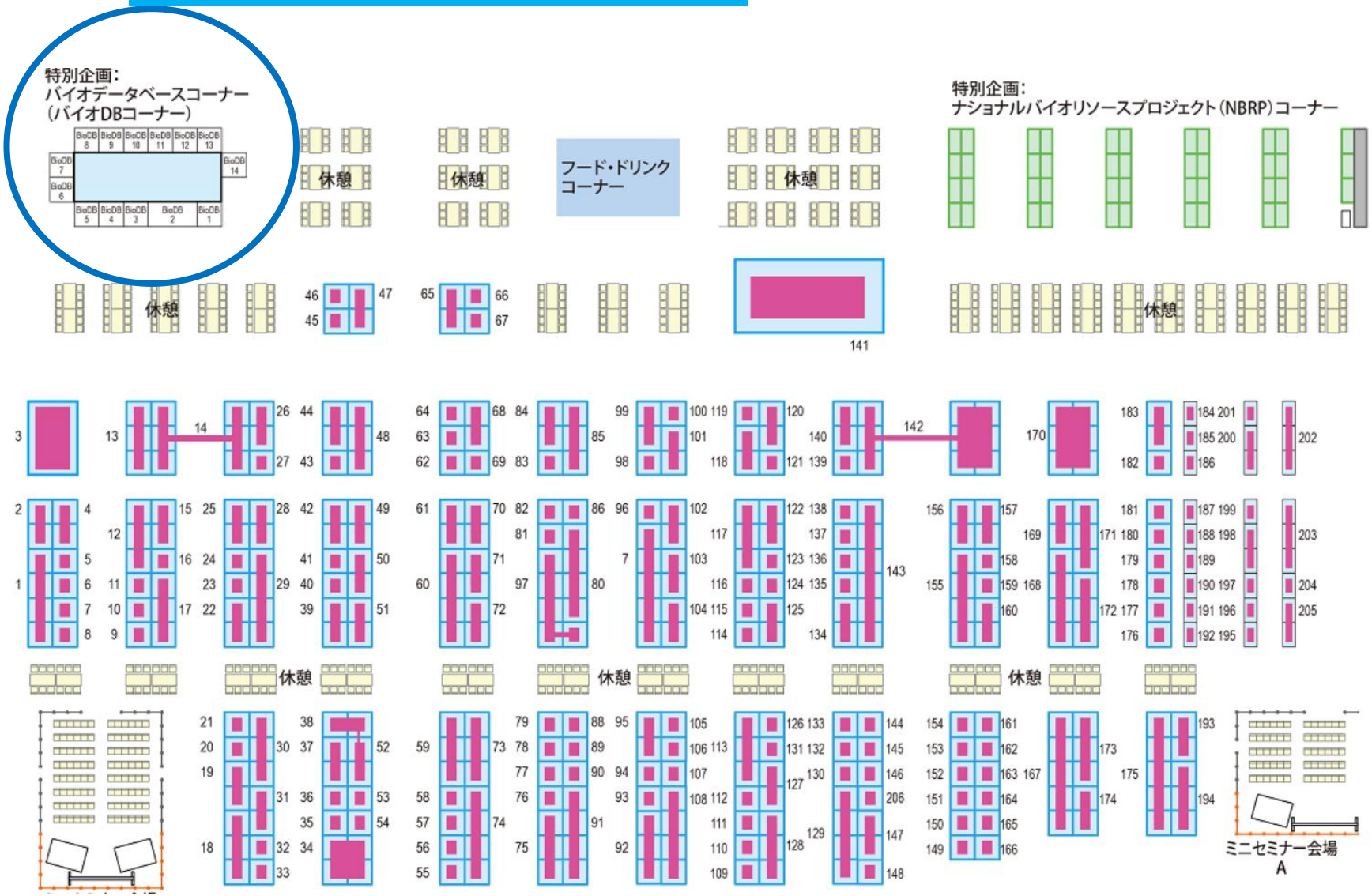
DBTSS (http://dbtss.hgc.jp/) was originally constructed as a collection of uniquely determined transcriptional start sites (TSSs) in humans and some other species in 2002. Since then, it has been regularly updated and in recent updates epigenetic information has also been incorporated because such information is useful for characterizing the biological relevance of these TSSs/downstream genes. In the newest release, Release 9, we further integrated public and original single nucleotide variation (SNV) data into our database. For our original data, we generated SNV data from genomic analyses of various cancer types, including 97 lung adenocarcinomas and 57 lung small cell carcinomas from Japanese patients as well as 26 cell lines of lung cancer origin. In addition, we obtained publically available SNV data from other cancer types and germline variations in total of 11,322 individuals. With these updates, users can examine the association between sequence variation pattern in clinical lung cancers with its corresponding TSS-seq, RNA-seq, ChIP-seq and BS-seq data. Consequently, DBTSS is no longer a mere storage site for TSS information but has evolved into an integrative platform of a variety of genome activity data.

To know accurate TSS positions is valuable as it could lead to more accurate characterization of its upstream transcriptional regulatory region. Thus, we constructed a database containing such information of mostly human genes in 2002. Since then, its updates have been regularly reported in the *Nucleic Acids Research* database issues (2004, 2006, 2008, 2010 and 2012 (4)). With the advances in sequencing technologies, we have developed TSS-seq, where the oligo-capping technique is applied to next generation sequencing (NGS), allowing even more accurate genome-wide determination of TSSs (5). The NGS sequencers are not only suited for determining genomic DNA sequences but also for transcriptome analysis (RNA-seq (6)) and epigenome analysis (ChIP-seq (7) and bisulfite sequencing (BS-seq; (8))) Since such additional data enable further biological characterization of transcriptional regulatory regions, we have also incorporated transcriptomic/epigenomic data of various tissues/cell cultures in DBTSS. In the latest update, Release 9, it contains 1257 million TSS tag sequences collected from 24 tissues and 33 cell cultures (see Table 1). It also contains the data of subcellularly fractionated RNAs as well as the ChIP-seq data of various histone modifications, binding sites of RNA polymerase II and several transcription factors, mainly, in cultured cell lines.

In this report, we introduce Release 9 of DBTSS, where we significantly enlarged the number of incorporated single nucleotide variation (SNV) data, which were both collected from publically available databases and generated from our own experiments (see below). The association of such large-

NBDCバイオDBコーナー

データベースのデモをやっています。



Data contents (ChIP Seq and others)

Cell lines — Data contents

Cell line			General information*						Cell culture		Sequencing dataset					Memo
Name	Type	in-house analysis number	Distributor	Catalogue number	Ethnicity	Gender	Age	Smoking status	Medium	Dish	Whole-genome	RNA-Seq	BS-Seq	ChIP-Seq**	TSS-Seq	
SAEC (control)	Small airway epithelial cell	s_35	TAKARA (Lonza)	-	-	M	-	-	-	collagen Type I-coated	-	♦	-	♦	♦	Normal
PC-9	lung adenocarcinoma	s_9	RIKEN BRC	RCB4455	Japanese	-	-	-	RPMI	-	♦	♦	♦	♦	♦	-
PC-14	lung adenocarcinoma	s_10	IBL	-	Japanese	-	-	-	RPMI	-	♦	♦	♦	♦	♦	-
RERF-LC-KJ	lung adenocarcinoma	s_13	RIKEN BRC	RCB1313	Japanese	M	78	-	RPMI	-	♦	♦	♦	♦	♦	-
RERF-LC-Ad1	lung adenocarcinoma	s_11	JCRB	JCRB1020	Japanese	M	70	-	RPMI	-	♦	♦	♦	♦	♦	-
RERF-LC-Ad2	lung adenocarcinoma	s_12	JCRB	JCRB1021	Japanese	M	-	-	RPMI	-	♦	♦	♦	♦	♦	-
LC2/ad	lung adenocarcinoma	s_18	RIKEN BRC	RCB0440	Japanese	F	51	-	DMEM	collagen Type I-coated	♦	♦	♦	♦	♦	-
RERF-LC-MS	lung adenocarcinoma	s_14	JCRB	JCRB0081	Japanese	-	-	-	EMEM	-	♦	♦	♦	♦	♦	-
VMRC-LCD	lung adenocarcinoma	s_16	JCRB	JCRB0814	Japanese	M	-	-	EMEM	-	♦	♦	♦	♦	♦	-
ABC-1	lung adenocarcinoma	s_17	JCRB	JCRB0815	Japanese	M	47	-	EMEM	-	♦	♦	♦	♦	♦	-
PC-7	lung adenocarcinoma	s_8	IBL	-	Japanese	-	-	-	RPMI	-	♦	♦	♦	♦	♦	Non-adherent
PC-3	lung adenocarcinoma	s_7	JCRB	JCRB0077	Japanese	F	48	-	RPMI	collagen Type I-coated	♦	♦	♦	♦	♦	-
II-18	lung adenocarcinoma	s_19	RIKEN BRC	RCB2093	Japanese	-	-	-	RPMI	-	♦	♦	♦	♦	♦	-
RERF-LC-OK	lung adenocarcinoma	s_15	JCRB	JCRB0811	Japanese	-	-	-	RPMI	-	♦	♦	♦	♦	♦	-
A549	lung adenocarcinoma	s_1	ATCC	CCL-185	Caucasian	M	58	-	DMEM	-	♦	♦	♦	♦	♦	-
A427	lung adenocarcinoma	s_20	ATCC	HTB-53	Caucasian	M	52	-	RPMI	-	♦	♦	♦	♦	♦	-
H322	lung adenocarcinoma	s_21	ATCC	CRL-5806	Caucasian	-	-	-	RPMI	-	♦	♦	♦	♦	♦	-
H1648	lung adenocarcinoma	s_25	ATCC	CRL-5882	Black	M	39	Y	RPMI	collagen Type I-coated	♦	♦	♦	♦	♦	-
H1650	lung adenocarcinoma	s_26	ATCC	CRL-5883	Caucasian	M	27	Y	RPMI	-	♦	♦	♦	♦	♦	-

Human Genomes

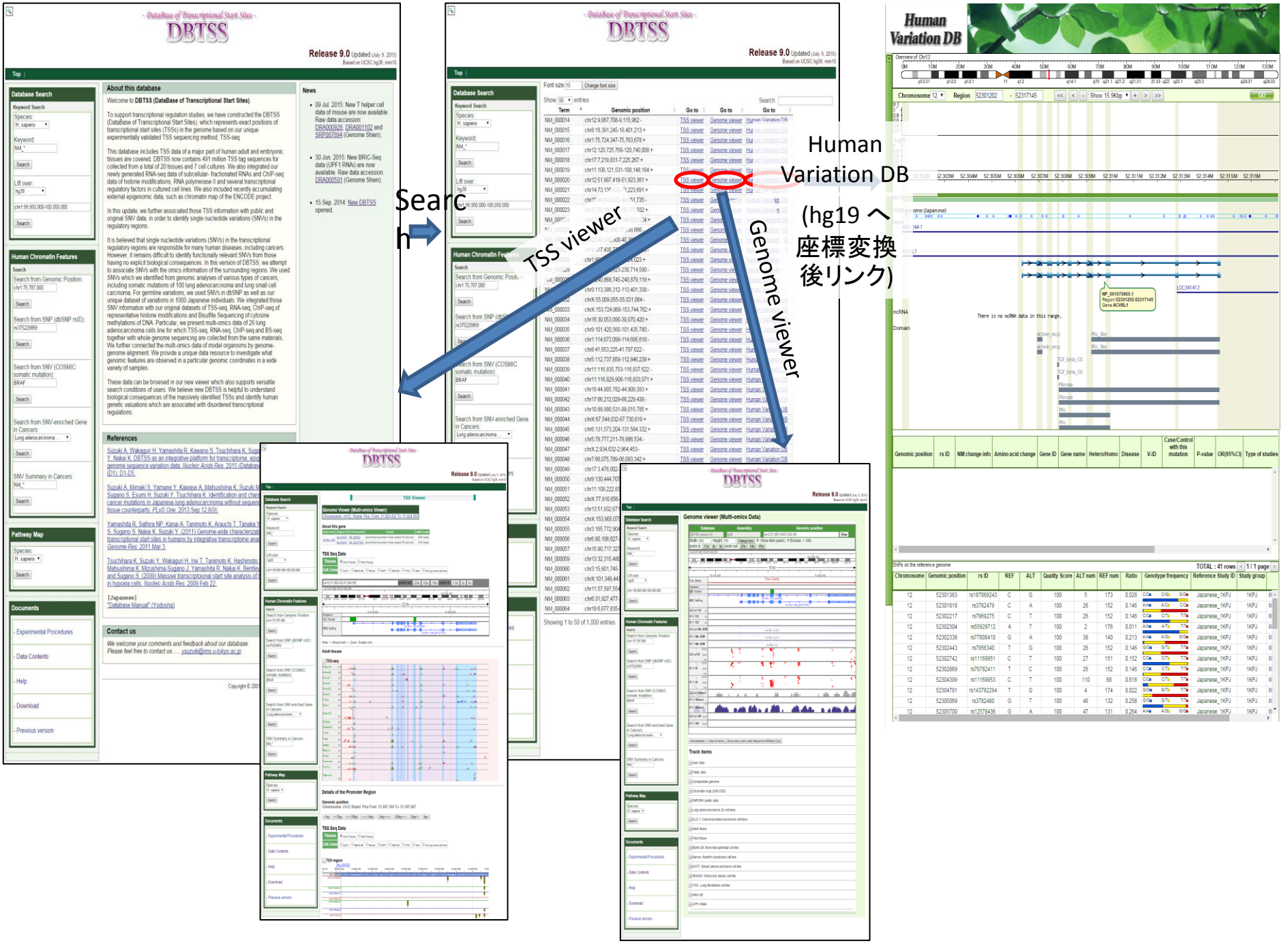
Data provider	Cancer type	Number of samples	Reference
National Cancer Center Hospital East and University of Tokyo	Lung adenocarcinoma	97	PLoS One. 2013 Sep 12;8(9):e73484.
National Cancer Center Hospital East	Small cell lung cancers	57	J Thorac Oncol. 2014 Sep;9(9):1324-31.
ICGC	43 of ICGC DCC Project Codes	6,590	https://dcc.icgc.org/
Dr. Meyerson's Lab.	Lung adenocarcinoma	183	Cell. 2012 Sep 14;150(6):1107-20.
Dr. Ogawa's Lab.	Myelodysplasia	29	Nature. 2011 Sep 11;478(7367):64-9
	Clear-cell renal cell carcinoma	106	Nat Genet. 2013 Aug;45(8):860-7
TCGA	Gastric adenocarcinoma	295	Nature. Published online 23 July 2014
	Urothelial bladder carcinoma	131	Nature. 507 (7492):315-22.
	Glioblastoma	291	Cell. 155 (2):462-477.
	Clear cell renal cell carcinoma	446	Nature. 499 (7456):43-49.
	Endometrial carcinoma	373	Nature. 497 (7447):67-73.
	Acute myeloid leukemia	200	NEJM. 368:2059-2074.
	Breast tumors	507	Nature. 490 (7418):61-70.
	Squamous cell lung cancers	178	Nature. 489 (7417):519:525.
	Colon and rectal cancer	224	Nature. 487 (7407):330-337.
	Ovarian carcinoma	316	Nature. 474 (7353):609-615.
	Glioblastoma	91	Nature. 455 (7216):1061-1068.
HGVD	Normal (Japanese)	1,208	URL: http://www.genome.med.kyoto-u.ac.jp/SnpDB
ToMMo	Normal (Japanese)	1,070	URL: http://humandbs.biosciencedbc.jp/en/hum0015-v1
JPDSC (Japan PGx Data Science Consortium)	Normal (Japanese)	2,994	URL: http://humandbs.biosciencedbc.jp/hum0013-v1
Total		**15,386**	

Exon Archive Project at MGH

ExAC release0.3; Total: 60,706 samples

Population	Male Samples	Female Samples	Total
African/African American (AFR)	1,888	3,315	5,203
Latino (AMR)	2,254	3,535	5,789
East Asian (EAS)	2,016	2,311	4,327
Finnish (FIN)	2,084	1,223	3,307
Non-Finnish European (NFE)	18,740	14,630	33,370
South Asian (SAS)	6,387	1,869	8,256
Other (OTH)	275	179	454
Total	33,644	27,062	60,706

DBに登録されているSNP(SNV)のサイト数は 全人種トータルで10,185,731箇所


DBTSS
Release 9.0
Search
TSS viewer
Genome viewer
Human Variation DB
(hg19 へ
座標変換
後リンク)

平成27年度の計画（経過）

- ゲノム支援データ（マウス）の収載
- RDF化の推進（TSSを端緒に）
- 臨床ゲノムデータとのリンク方法の開発

Other species

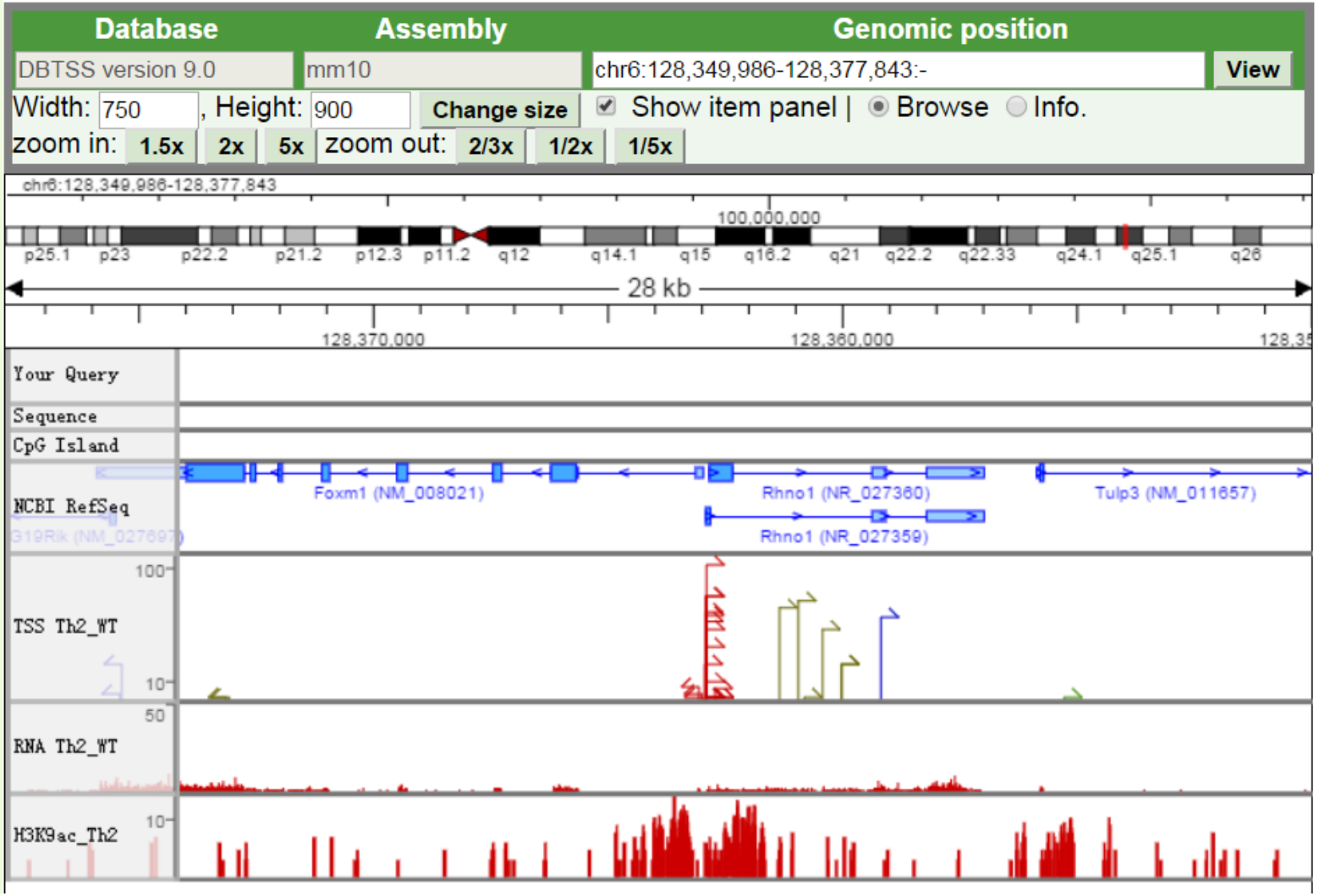
	Human	Mouse	Malaria	Chyzon	Rat	Chimp	Macaque
TSS-seq	73	7	1	1	1	1	2
RNA-seq	42	0	0	0	0	0	0
ChIP-seq	255	0	0	0	0	0	0
RIP-seq	12	0	0	0	0	0	0
BS-seq	26	0	0	0	0	0	0
ChromHMM	36	0	0	0	0	0	0
SNV	49	0	0	0	0	0	0

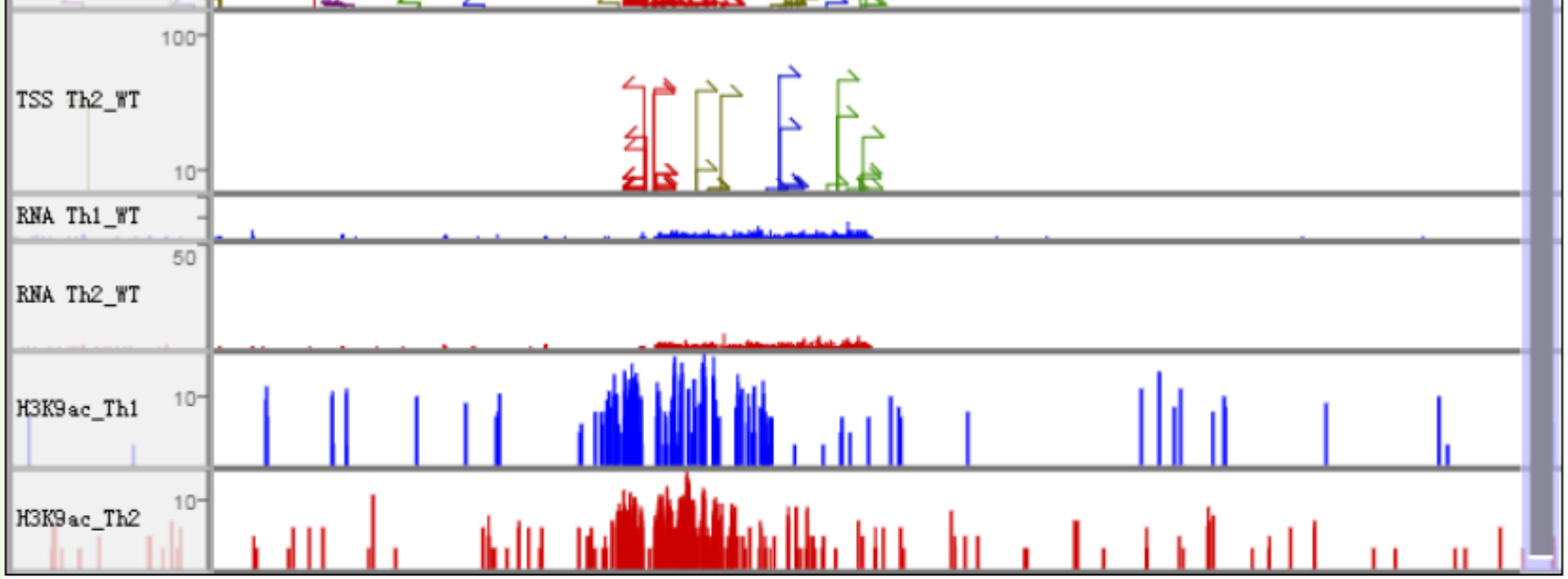
Integrating mouse data (ゲノム支援)

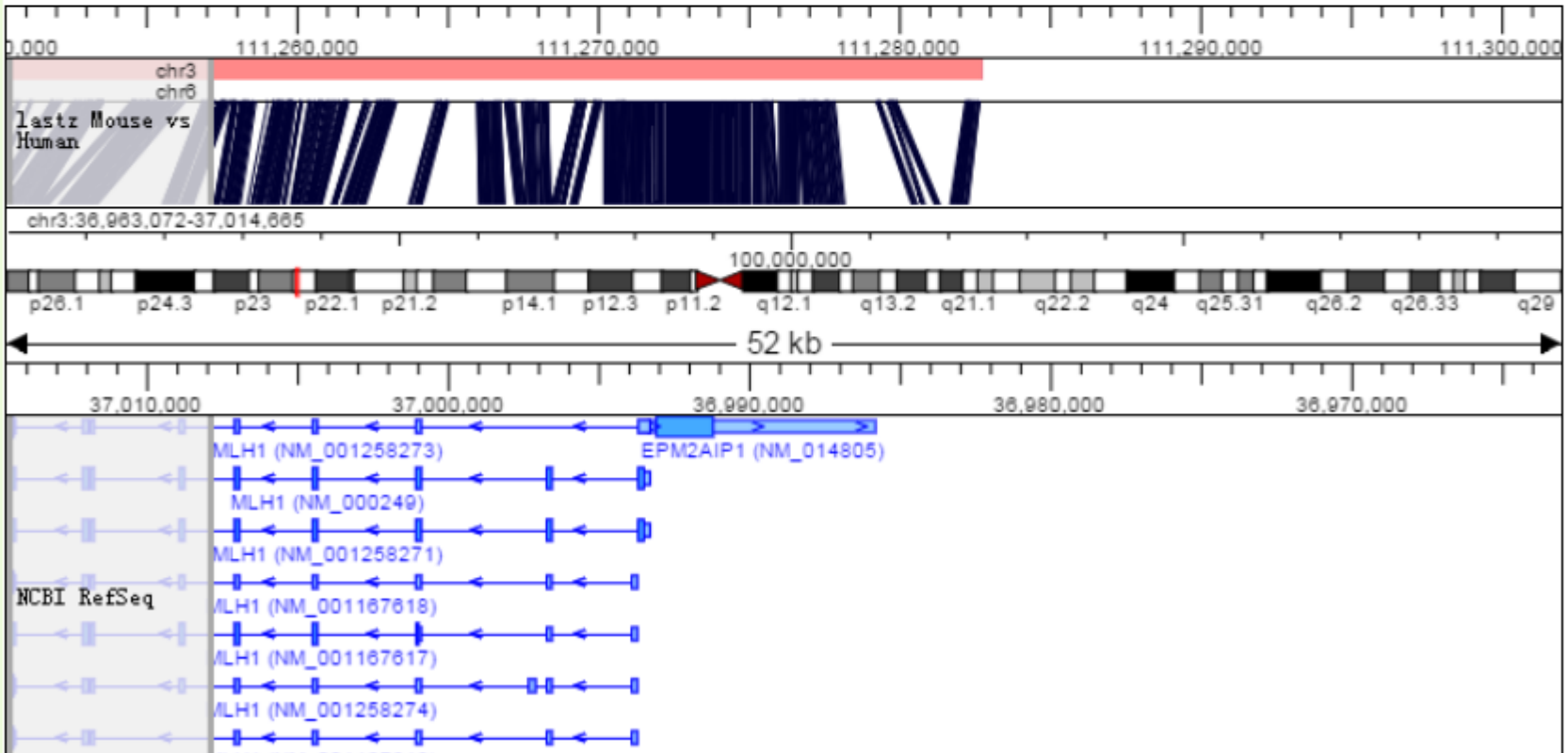
名前	所属	課題	Acc	データセット	配列数
田中 知明	千葉大学	転写因子p53による新たな代謝調節機能と代謝環境応答のエピジェネティクス制御	DRA000928 SRP007894 DRA001102	26	515,880,048
秋光 信佳	東京大学アイソトープ総合センター	RNA分解を介した核局在型高分子非コードRNA作用マシナリーの制御機構の解明	DRA000591	25	943,054,680

田中先生データ(Mouse)

Genome viewer (Multi-omics Data)




100
TSS Th2_WT
10
RNA Th1_WT
50
RNA Th2_WT
H3K9ac_Th1
10
H3K9ac_Th2
10

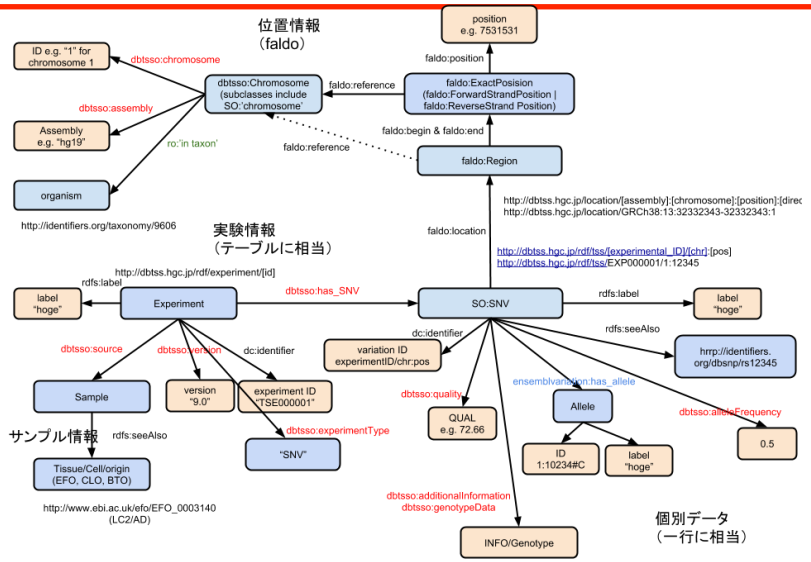

111,260,000
111,270,000
111,280,000
111,290,000
111,300,000
chr3
chr6
lastz Mouse vs Human
chr3:36,963,072-37,014,665
100,000,000
p26.1
p24.3
p23
p22.1
p21.2
p14.1
p12.3
p11.2
q12.1
q13.2
q21.1
q22.2
q24
q25.31
q26.2
q26.33
q29
52 kb
37,010,000
37,000,000
36,990,000
36,980,000
36,970,000
MLH1 (NM_001258273)
EPM2AIP1 (NM_014805)
MLH1 (NM_000249)
MLH1 (NM_001258271)
NCBI RefSeq
MLH1 (NM_001167618)
MLH1 (NM_001167617)
MLH1 (NM_001258274)

Integrating mouse data (ゲノム支援;作業中)

名前	所属	課題	Acc	データセット	配列数
三野 享史	京都大学ウイルス研究所感染防御研究分野	Regnase-1の作用機構から探る自然免疫応答における転写後調節の解明	DRA003234	8	161,938,914
岩間 厚志	千葉大学大学院医学研究院先端応用医学講座細胞分子医学	エピジェネティック異常による造血器腫瘍の発症機構の解明	DRA000485 DRA000486 DRA000488 DRA000858	20	600,000,000
河野 友宏	東京農業大学生物応用科学部	次世代シークエンサーを用いた生殖系列のエピゲノム修飾とトランスクリプトーム解析	DRA000484 DRA000607	133	8,900,000,000
深田 吉孝	東京大学大学院理学系研究科	生存戦略としての体内時計システムの分子解剖	DRP001092 DRP001093 DRP001094 DRP001349	54	1,350,000,000
小野寺 淳	千葉大学・大学院医学研究院・免疫発生学	ポリコーム／トライソラックス複合体による免疫記憶維持機構の解明	GSE51079	6	180,000,000
西中村 隆一	熊本大学	胎児型腎臓幹細胞の成体腎での再活性化	DRA000957	6	150,000,000

RDF化

- Variationデータ用オントロジー
  - variation: ENSEMBL Ontology
  - location: FALDO
  - 不足分は独自オントロジー定義
- DBTSSオントロジー
- RDF化スクリプト




```
#################################################################
#
#    Data properties
#
#################################################################


###  http://dbtss.hgc.jp/rdf/ontology/dbtss.owl#additionalInformation

:additionalInformation rdf:type owl:DatatypeProperty ;

                       rdfs:label "Additional Information" ;

                       rdfs:comment "Values from INFO tag" .


###  http://dbtss.hgc.jp/rdf/ontology/dbtss.owl#alleleFrequency

:alleleFrequency rdf:type owl:DatatypeProperty ;

                 rdfs:label "Allele Frequency" ;

                 rdfs:comment "Allele Frequency, for each ALT allele, in the same order as listed" ;

                 rdfs:range xsd:float .


###  http://dbtss.hgc.jp/rdf/ontology/dbtss.owl#allelicDepths

:allelicDepths rdf:type owl:DatatypeProperty ;

               rdfs:label "Allelic Depths" ;
```

- DBTSS Variation RDF

```
<http://dbtss.hgc.jp/rdf/experiment/TSE000086> <http://dbtss.hgc.jp/rdf/ontology/has_SNV> <http://dbtss.hgc.jp/rdf/data/TSE0

<http://dbtss.hgc.jp/rdf/data/TSE000086/1:10234> a <http://purl.obolibrary.org/obo/SO_0001483>;
   <http://www.w3.org/2000/01/rdf-schema#label> "variation on chr1:10234 from TSE000086";
   <http://biohackathon.org/resource/faldo#location> <http://dbtss.hgc.jp/rdf/location/chromosome:GRCh38:1:10234-10234:1>;
   <http://dbtss.hgc.jp/rdf/ontology/additionalInfomation> "AC=1;AF=0.50;AN=2;BaseQRankSum=-0.777;DB;DP=53;Dels=0.02;FS=1.862
   <http://dbtss.hgc.jp/rdf/ontology/alleleFrequency> 0.5;
   <http://dbtss.hgc.jp/rdf/ontology/allelicDepths> "42,10";
   <http://dbtss.hgc.jp/rdf/ontology/dbsnpID> "rs145599635";
   <http://dbtss.hgc.jp/rdf/ontology/genotype> "0/1";
   <http://dbtss.hgc.jp/rdf/ontology/genotypeData> "GT:AD:DP:GQ:PL|0/1:42,10:52:61.94:103,0,62";
   <http://dbtss.hgc.jp/rdf/ontology/genotypeQuality> 61.94;
   <http://dbtss.hgc.jp/rdf/ontology/quality> 72.66;
   <http://purl.org/dc/terms/identifier> "1:10234";
   <http://www.w3.org/2000/01/rdf-schema#seeAlso> <http://info.identifiers.org/dbsnp/rs145599635>,
     <http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs145599635>;
   <https://github.com/simonjupp/ensembl-rdf/blob/master/ontology/ensembl_variation_ontology.owl#has_allele> <http://dbtss.hg
     <http://dbtss.hgc.jp/rdf/data/TSE000086/1:10234#T> .

<http://dbtss.hgc.jp/rdf/data/TSE000086/1:10234#C> a <https://github.com/simonjupp/ensembl-rdf/blob/master/ontology/ensembl_v
     <https://github.com/simonjupp/ensembl-rdf/blob/master/ontology/ensembl_variation_ontology.owl#ancestral_allele>;
   <http://www.w3.org/2000/01/rdf-schema#label> "TSE000086 chr1:10234 allele C";
   <http://purl.org/dc/terms/identifier> "1:10234#C" .
```

がん“最適化創薬”の実現に向けた臨床情報・ゲノム情報管理システムの構築

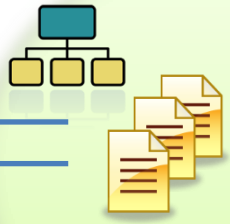
担当医師

臨床検体

ISO 15189

遺伝子検査

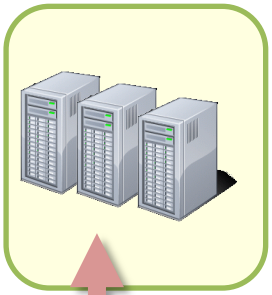
電子カルテ

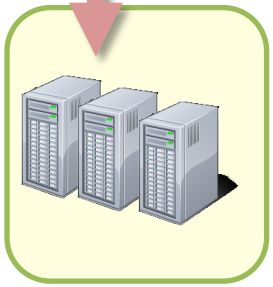


患者背景
治療情報

プライバシーの保護

ゲノム情報





エキスパート
パネル

担当医師

患者毎のQueryに
対し、臨床情報と
ゲノム情報を連結

治療選択

患者

承認薬

治験

救済的使用

適応外使用

分子疫学情報の共有

臨床試験情報の共有

既存のバイオバンク
データベース

既存の臨床試験
データベース

新しい診断・治療法
の開発

ヒト疾患ゲノム統合DB (DBTSSの拡張):

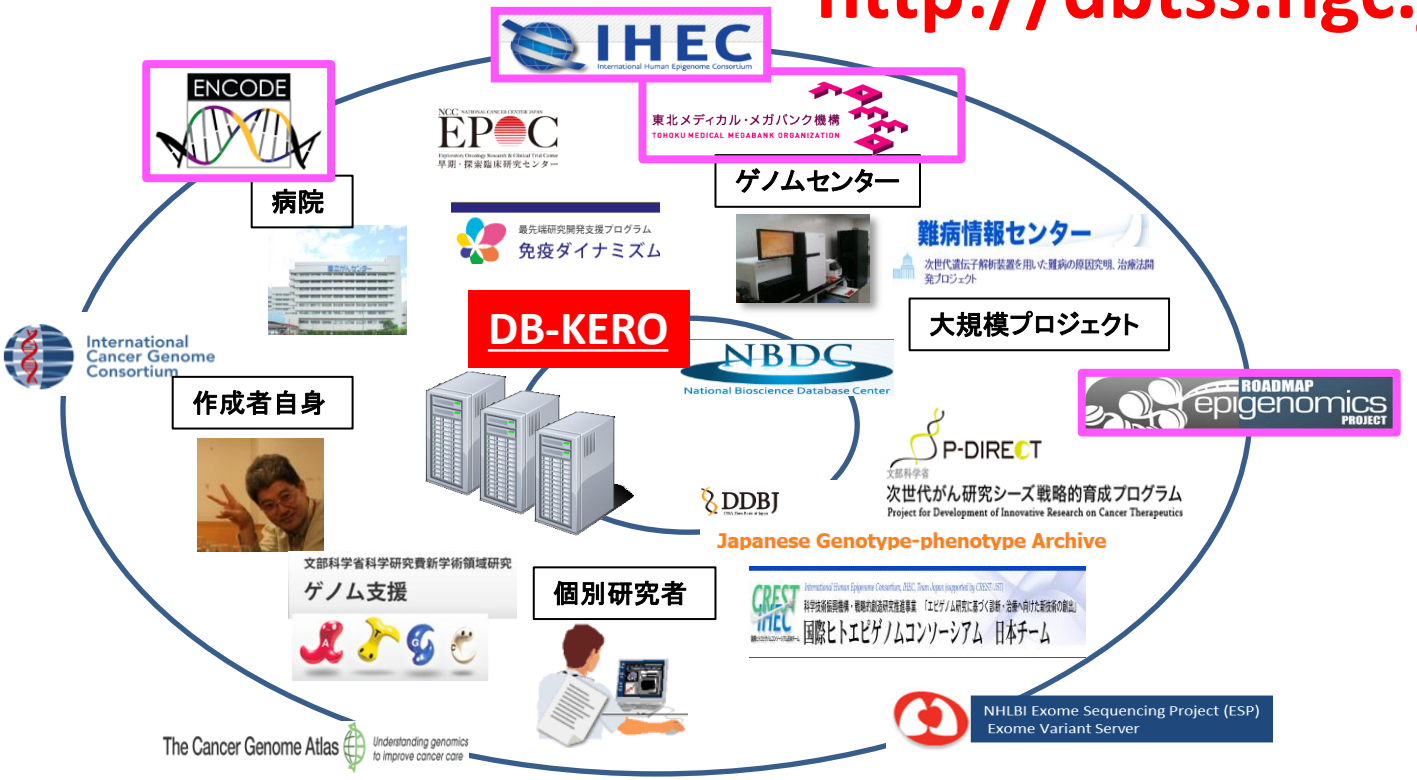
KERO(Kashiwa Encyclopedia of Regulatory Omics)

ヒトゲノム・エピゲノム・トランスクリプトームデータの統合

ヒト疾患ゲノム変異への機能的注釈

パターン検索システムの開発と実装

http://dbtss.hgc.jp/



オミクスデータ統合が加速するヒトゲノム臨床応用研究

＝疾患ゲノムのその座標で“何が起きているのか”を網羅的に検索