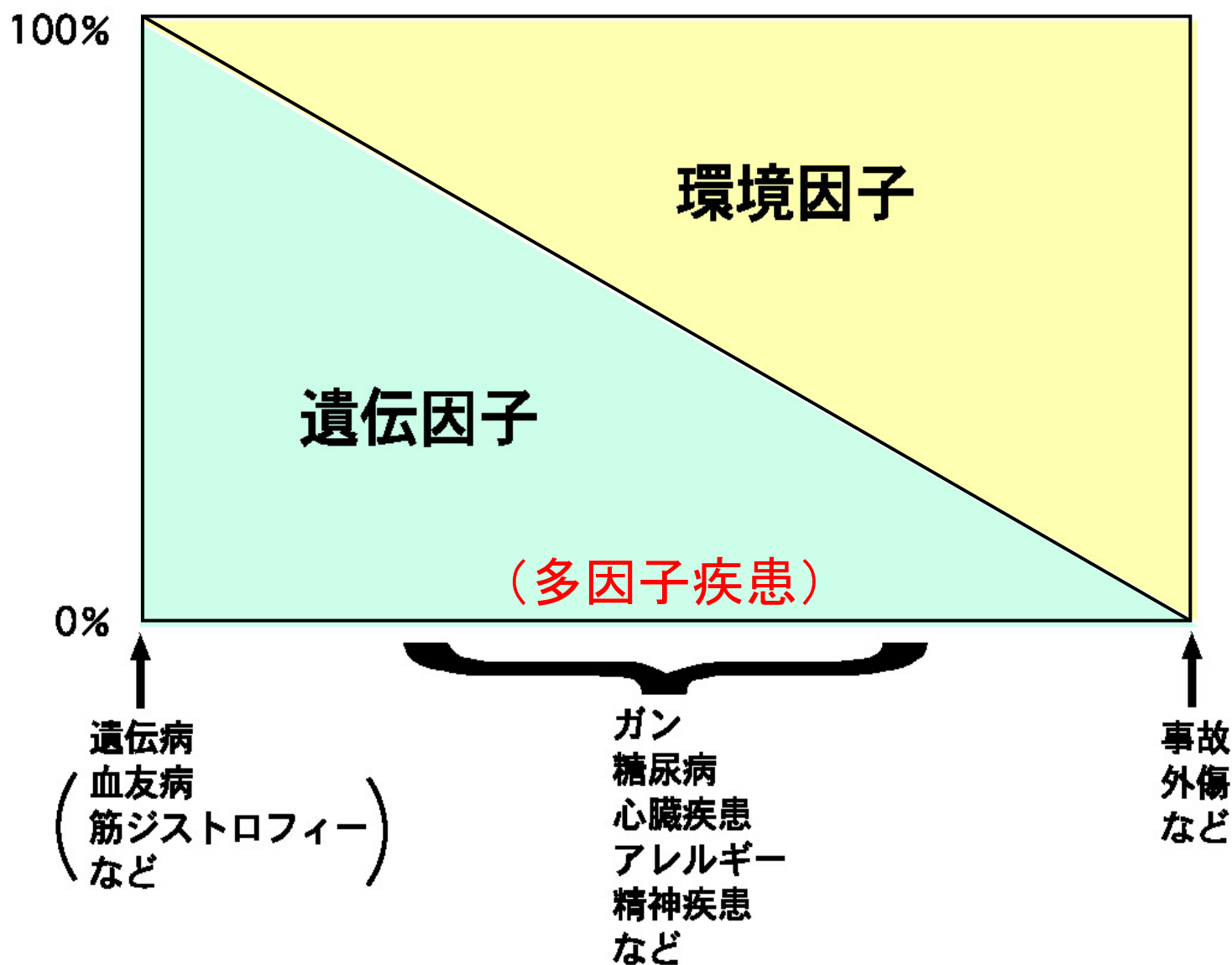


ゲノム医学研究の現状と 改正個人情報保護法への懸念

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疾患に関わる遺伝因子と環境因子



ゲノム全域から疾患遺伝子を 探索する研究の潮流

単一遺伝子疾患 (遺伝病、難病)

連鎖解析法
(パラメトリック)



全エクソーム配列解析
(全ゲノム配列解析)
&
Clinical sequencing

遺伝病・難病の原因変異の同定、
データベース情報共有、
臨床検査への導入

多因子疾患

連鎖解析法
(ノンパラメトリック)



ゲノム全域関連解析法
(GWAS) &
全ゲノムimputation



(全ゲノム配列解析)

国内外大規模共同研究、
データベース利活用

- データベースによる情報共有の例 -

米国NCBI HP → Genetics & Medicine

NCBI Home	Genetics & Medicine						Quick Links
Resource List (A-Z)	All	Databases	Downloads	Submissions	Tools	How To	Bookshelf
All Resources							Database of Genotypes and Phenotypes (dbGaP)
Chemicals & Bioassays							Gene
Data & Software							Online Mendelian Inheritance in Man (OMIM)
DNA & RNA							PubMed
Domains & Structures							PubMed Central (PMC)
Genes & Expression							PubMed Health
Genetics & Medicine							RefSeqGene
Genomes & Maps							Map Viewer
Homology							PubMed Clinical Queries
Literature							
Proteins							
Sequence Analysis							
Taxonomy							
Training & Tutorials							
Variation							

Databases

Bookshelf
A collection of biomedical books that can be searched directly or from linked data in other NCBI databases. The collection includes biomedical textbooks, other scientific titles, genetic resources such as *GeneReviews*, and NCBI help manuals.

ClinVar
A resource to provide a public, tracked record of reported relationships between human variation and observed health status with supporting evidence. Related information in the [NIH Genetic Testing Registry \(GTR\)](#), [MedGen](#), [Gene](#), [OMIM](#), [PubMed](#) and other sources is accessible through hyperlinks on the records.

Database of Genotypes and Phenotypes (dbGaP)
An archive and distribution center for the description and results of studies which investigate the interaction of genotype and phenotype. These studies include genome-wide association (GWAS), medical resequencing, molecular diagnostic assays, as well as association between genotype and non-clinical traits.

Database of Major Histocompatibility Complex (dbMHC)
An open, publicly accessible platform where the HLA community can submit, edit, view, and exchange data related to the human major histocompatibility complex. It consists of an interactive Alignment Viewer for HLA and related genes, an MHC microsatellite database, a sequence interpretation site for Sequencing Based Typing (SBT), and a Primer/Probe database.

Gene
A searchable database of genes, focusing on genomes that have been completely sequenced and that have an active research community to contribute gene-specific data. Information includes nomenclature, chromosomal localization, gene products and their attributes (e.g., protein interactions), associated markers, phenotypes, interactions, and links to citations, sequences, variation details, maps, expression reports, homologs, protein domain content, and external databases.

GeneReviews

遺伝病原因遺伝子変異のデータベース：統計値

ClinVar submissions

This page summarizes submissions and properties of submissions ([assertion criteria](#), genes, unique variation records), represented in ClinVar.

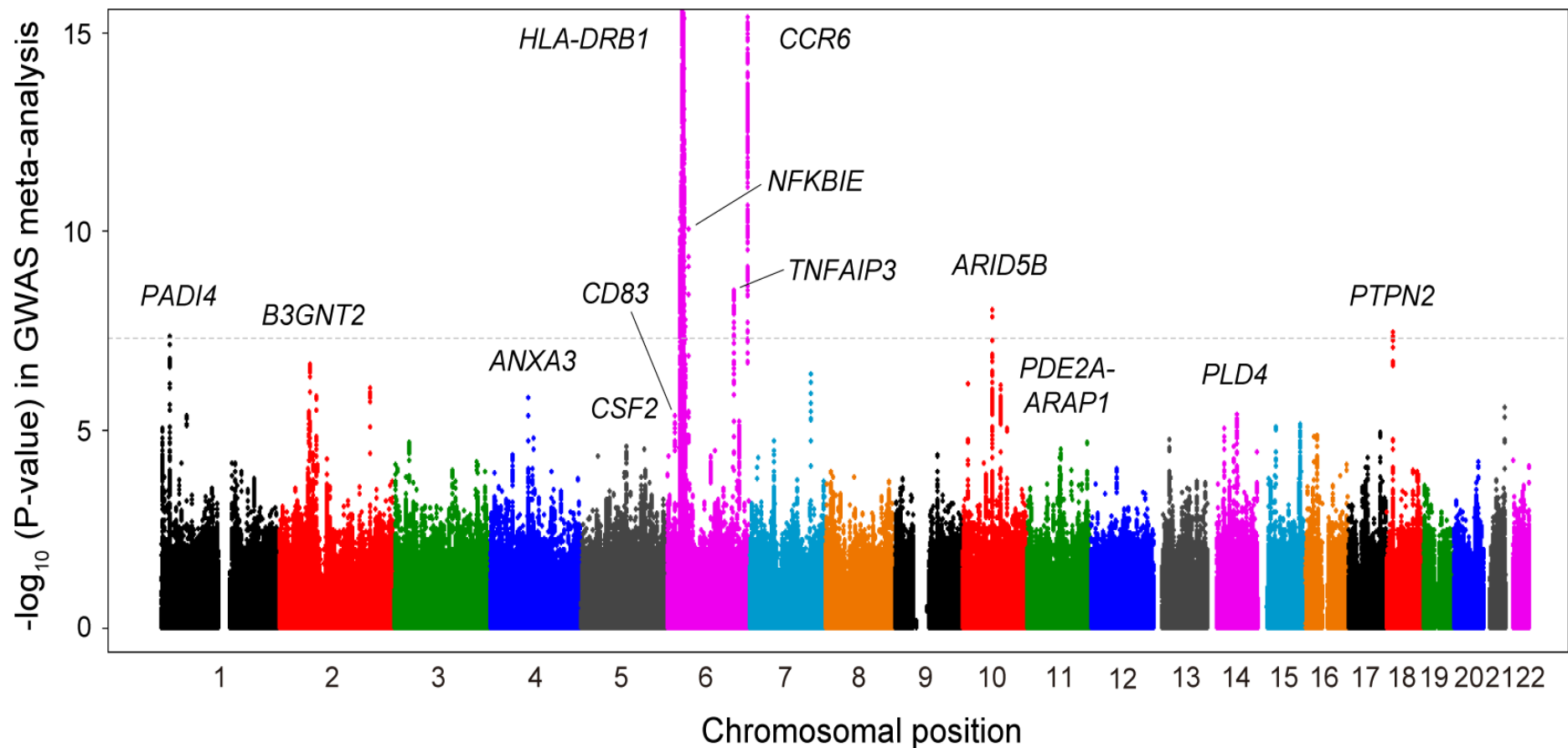
Follow [this link](#) to review all submitters and the summary of their contributions. We acknowledge their support.

Submission overview

Category of analysis	Current total (Mar 21, 2016)
Records submitted	179845
Records with assertion criteria	90796
Total genes represented	26868
Unique variation records	139791
Unique variation records with interpretations	126247
Unique variation records with assertion criteria	73919
Unique variation records with practice guidelines (4 stars)	23
Unique variation records from expert panels (3 stars)	3626
Unique variation records with assertion criteria, multiple submitters, and no conflicts (2 stars)	8243
Unique variation records with assertion criteria (1 star)	58955
Genes with variants specific to one gene	4666
Genes with variants specific to one protein-coding gene	4579
Genes included in a variant spanning more than one gene	26763
Variants affecting overlapping genes	9098
Total submitters	482

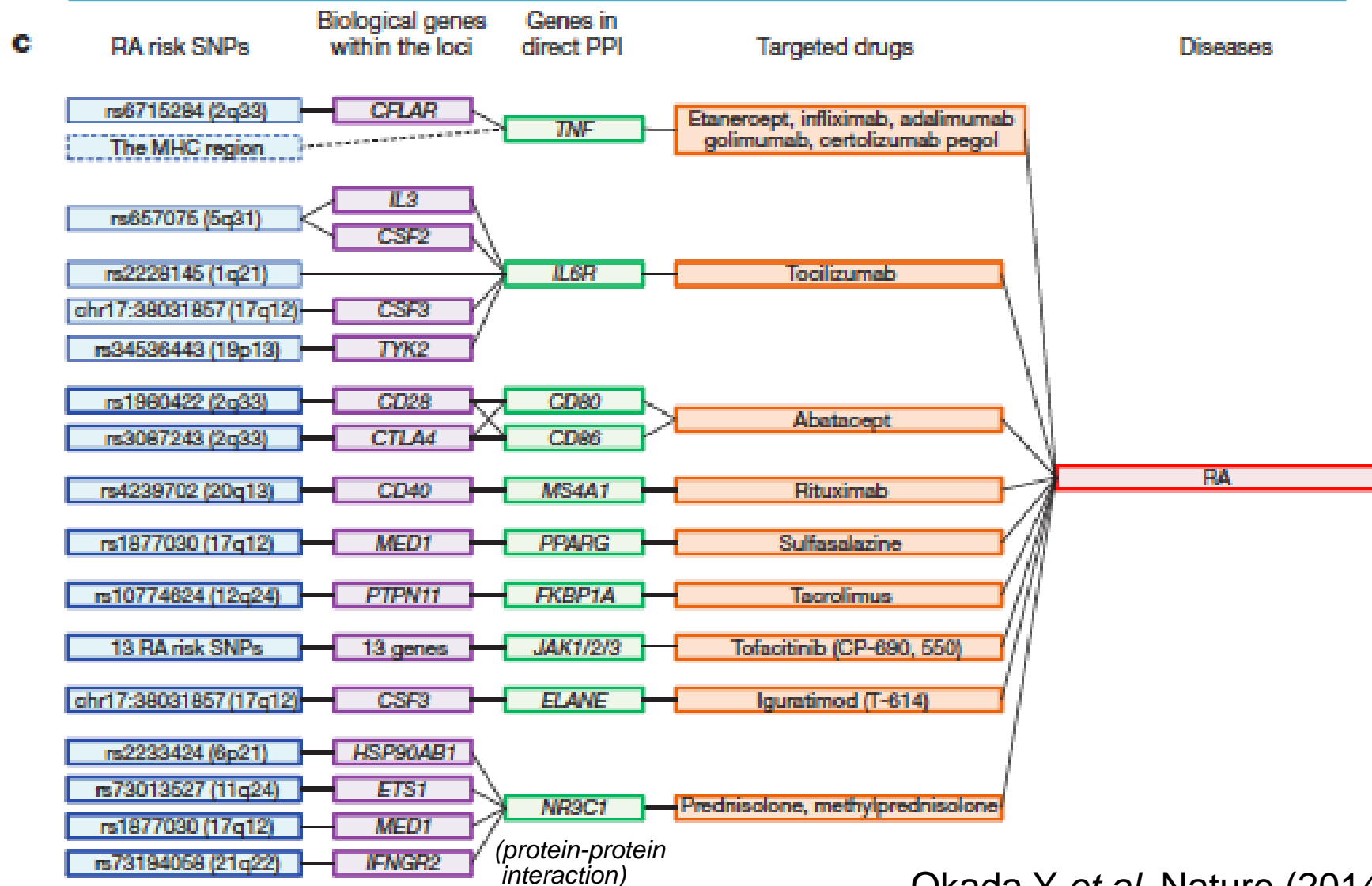
日本人 대규모 GWAS メタ解析による 関節リウマチの新規感受性遺伝子の同定

(GWAS in 4,074 / 16,891; replication in 5,277 / 21,684)



新規遺伝子 : *B3GNT2*, *ANXA3*, *CSF2*, *CD83*, *ARID5B*, *PDE2A-ARAP1*, *PLD4*, *PTPN2*

関節リウマチに関する**国際大規模GWAS**メタ解析 (患者3万人、対照者7.4万人)により、 101個の感受性座位が検出され、**治療標的**が示された



統合失調症に関する国際大規模GWASメタ解析
(患者3.7万人/対照者11.3万人; 108個の感受性座位
→ 病因について新たな示唆)

ARTICLE

doi:10.1038/nature13595

Biological insights from 108 schizophrenia-associated genetic loci

Schizophrenia Working Group of the Psychiatric Genomics Consortium*

Schizophrenia is a highly heritable disorder. Genetic risk is conferred by a large number of alleles, including common alleles of small effect that might be detected by genome-wide association studies. Here we report a multi-stage schizophrenia genome-wide association study of up to 36,989 cases and 113,075 controls. We identify 128 independent associations spanning 108 conservatively defined loci that meet genome-wide significance, 83 of which have not been previously reported. Associations were enriched among genes expressed in brain, providing biological plausibility for the findings. Many findings have the potential to provide entirely new insights into aetiology, but associations at *DRD2* and several genes involved in glutamatergic neurotransmission highlight molecules of known and potential therapeutic relevance to schizophrenia, and are consistent with leading pathophysiological hypotheses. Independent of genes expressed in brain, associations were enriched among genes expressed in tissues that have important roles in immunity, providing support for the speculated link between the immune system and schizophrenia.

個人情報保護法の改正に関する懸念

ゲノム情報は、個人名などの情報と共に登録されていて利用できるという条件がないと個人の特定につながらない。顔や指紋との大きな違い。

目的・用途による違い

鑑別(親子鑑定、個人識別)のためのDNA検査:個人識別性

研究(疾患遺伝子探索など)のためのゲノム解析:そのままでは個人識別性低い(意図的に解析しない限り、個人識別性低い)

憲法:学問研究の自由との関係

国内・国際大規模共同研究を阻害

バイオバンク、公的データベースの利活用に制限、情報共有、ゲノム医療・適切な医療サービスの実現への障害

既収集試料の利用に制限、連結不可能・可能匿名化が無効(研究活動大幅ダウン)?

ゲノム領域によって識別性異なる(連鎖不平衡)、集団差の問題、ゲノム(germ line, somatic mutation)・エピゲノム配列情報・配列解析エラーの扱い

むしろ、研究目的のデータから個人識別を試みる行為を禁止すべき(バイオバンク、データベースでは、利用の際に覚書署名)