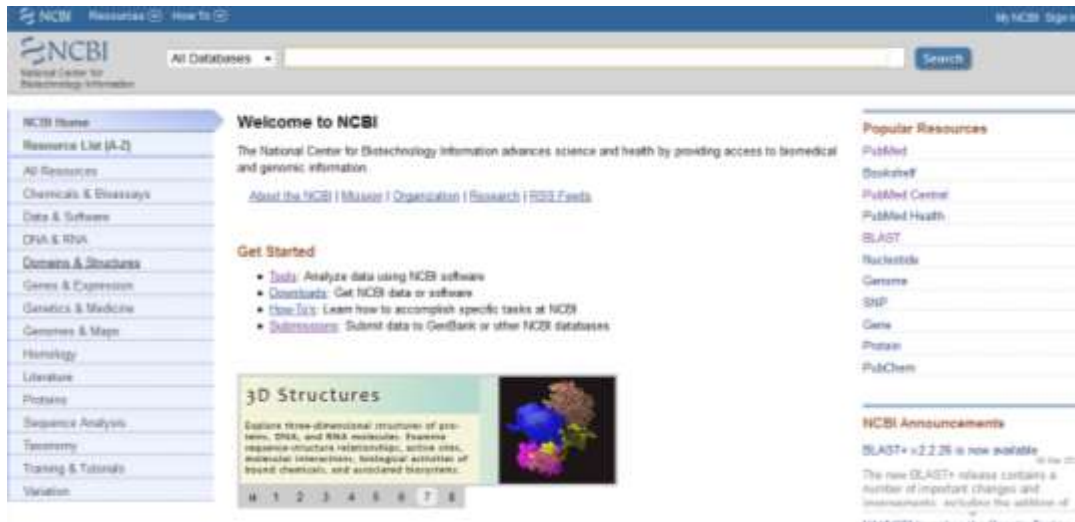
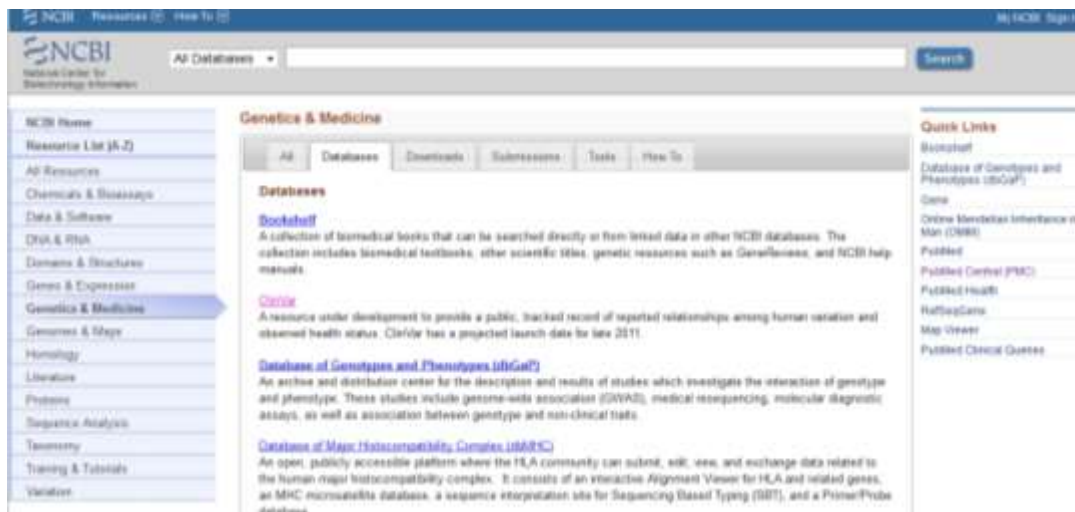


## Pengenalan NCBI untuk Klinik

National Center for Biotechnology Information (NCBI) memiliki data base yang berisi, senyawa kimia termasuk obat-obatan, Gen, DNA dan protein. Salah satu database yang ada di NCBI yang berkaitan langsung dengan klinik adalah Gene & Medicine. Untuk mengunjungi data base dapat menuju halaman utama NCBI dengan alamat: [www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov) seperti tampilan dibawah ini.



Genetic & Medicine memiliki beberapa database diantaranya adalah CLINVAR yang berisi hubungan antara gen dengan fenotip terutama terkait status kesehatan manusia.



CLINVAR memiliki data base dan tool yang sangat berguna untuk klinik diantaranya adalah Clinical Queries, dbGap, Variation, GeneReviews, dan OMIM.

Clinical Queries berisi database Publikasi yang terkait dengan penyakit yang dapat dikelompokkan menjadi 5 kategori yaitu therapy, Diagnosis, Pronosis, Etiology dan clinical guide.

GeneReviews, merupakan sumber informasi/buku yang berisi kaitan gen dengan status kesehatan manusia seperti dibawah ini

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GeneReviews™  
 Edited by Roberta A Pagan, Editor-in-chief, Thomas D Bird, Cynthia R Dalen, Karen Stephens, and Margaret P Adam  
 Seattle (WA): University of Washington, Seattle; 1995.  
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GeneReviews are expert-authored, peer-reviewed disease descriptions that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions. Published exclusively online, each GeneReviews entry is: (1) peer reviewed for accuracy by (a) editorial staff experts in clinical genetics, laboratory genetics, and genetic counseling and by (b) acknowledged international subject experts, (2) updated by the author(s) in a formal comprehensive process every two to three years or as needed, and (3) revised by the author(s) or editorial staff whenever significant changes in clinically relevant information occur. GeneReviews are part of the GeneTests Web site, which also includes: a Laboratory Directory of US and international laboratories offering molecular genetic testing, specialized cytogenetic testing, and biochemical testing for inherited disorders, a Clinic Directory of US and international genetics clinics providing genetic evaluation and genetic counseling, Resources that are consumer health-oriented organizations and disease registries, Educational materials, and an Illustrated Glossary of genetic counseling and testing terms.

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GeneTests-GeneClinics: genetic testing information for a growing audience. [Hum Mutat. 2002]  
 GeneClinics: a hybrid text/data electronic publishing model using XML, applied to clinical genetic test. [Am J Med Inform Assoc. 2002]  
 Good laboratory practices for biochemical genetic testing and newborn screening for inherited metabolic disorders. [Pediatrics. 2002]  
 Genome: Evaluation of prenatally diagnosed structural congenital anomalies. [J Clinet Genet Hum Genet. 2001]  
 Genome: Hereditary nonpolyposis colorectal cancer: diagnostic strategies and their use. [Genet Test Mol Diagn. 2001]

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dbSNP berisi SNP yang telah ditemukan yang dapat memiliki kaitan dengan suatu fenotip/kondisi klinik.

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As: 797 | Cited in PubMed: 75 | ClinicalLSD Submissions: 219 | Human (Homo sapiens): 228

Items 1 - 20 of 219 Page 1 of 11 First

Recent activity: Diabetes (797), 1p36 Deletion Syndrome - GeneReviews™, diabetes (16297), gene(s) (27024), diabetes.MED.1a\_diagnosis... (110)

1: rs122467175 [Homo sapiens] Links  
 GGAGCCTCANTGGATCTACACTAG (G/T)TGAGGSCATGTTTGCTTCTCGAG  
 Allele Origin: C-Germine T-Germine  
 Clinical Significance: probable pathogenic  
 HGVS Names: [NC\_000022.10:g.491081724+0][[AG\_007362 r.g.181577+0][[AG\_021371 r.g.212964+0][[AG\_01114277 r.g.18947+0][[NM\_014009.3:c.10897+0][[AP\_01107842 r.g.Pw03323a][[AP\_034726.2:g.Pw0307a]

2: rs104894761 [Homo sapiens] Links  
 AGATGATCTGCGCAGTACGCTGAG (C/G)T)CCGACCTCCGATCTGCGCTCCAT  
 Allele Origin: C-Germine T-Germine G-Germine  
 Clinical Significance: probable pathogenic  
 HGVS Names: [NC\_000022.10:g.102171390+0][[AG\_009697 r.g.20420+0][[AG\_013220 r.g.255460+0][[AG\_000054 r.g.4090+0][[NM\_01142101 r.g.490+0][[AP\_000045 r.g.Asp1373a][[AP\_01129623 r.g.Asp1373a][[AG\_021371 r.g.880-1042+0]

3: rs104894760 [Homo sapiens] Links

OMIM merupakan data base yang berisi tentang gen dan penyakit genetik pada manusia dapat di buka di link dibawah ini

