

Towards Collaborative Publishing in Science

---

# **Towards Collaborative Publishing in Science**

Decentralized Information Group - Cambridge, September 2008.

Robert Hoffmann

### **'Conventional' publications**

- Stone, paper, silicon, ...
- Closed number of authors
- Date of publication
- Final version
- 'static'

### **Scientific discourse**

- Over a series of static publications

### **'Dynamic' publications**

- [Open number of authors]
- Versions but no final date of publication
- Emphasis may shift over time
- Evolve with the subject
- Potentially always up-to-date
- Incorporate scientific discourse

# Towards Collaborative Publishing in Science

**wiki**genes  
evolutionary knowledge

Hello Marta Ramirez-Gaité. [Your account] [sign out]

Gene: **TP53** tumor protein p53 (Li-Fraumeni syndrome) Homo sapiens  
[edit this page]

Barrett, M.T., Alvi, A.J., Berns, E.M., Stankovic, T., Balachandran, R., et al.

**Disease relevance of TP53**

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC proto-oncogenes** and the **TP53 tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

**Psychiatry related information on TP53**

- **TP53** mutations were analyzed in 35 human papillomavirus (HPV) type 16 DNA-positive cancers of the **oral cavity** and **oropharynx** and in 35 HPV DNA-negative cancers matched by subsite, country, sex, age, and tobacco and **alcohol consumption**. [Ref.]
- In order to examine the role of the **TP53** gene in the pathogenesis of **schizophrenic disorders**, we investigated the genetic association between a functional polymorphism rs1042522 and schizophrenia by sequencing the fragment covering 72Pro> Arg in 701 cases and 695 controls in this work. [Ref.]
- The **Huntington's disease** protein interacts with p53 and CREB-binding protein and represses transcription. [Ref.]
- NF-kappaB/rel proteins, **tumor suppressor p53**, and **oncogene c-Myc** are critical transcription factors involved in coordinating cellular **decision-making** events in response to external stimuli. [Ref.]
- **Tauroursodeoxycholic acid** modulates **p53**-mediated **apoptosis** in **Alzheimer's disease** mutant **neuroblastoma** cells. [Ref.]

**High impact information on TP53**

- They show that the **p38**-regulated/activated protein kinase (PRAK) induces **senescence** downstream of oncogenic Ras by directly phosphorylating and activating the tumor-suppressor protein **p53**. [Ref.]
- **Telomere** dysfunction suppresses cancer through the **p53** tumor suppressor pathway but also contributes to aging. [Ref.]
- Therefore, NEFD4-1 is a potential **proto-oncogene** that negatively regulates **PTEN** via ubiquitination. a

**Table of contents**

- Disease relevance of **TP53**
- Psychiatry related information on **TP53**
- High impact information on **TP53**
- Chemical compound and disease context of **TP53**
- Biological context of **TP53**
- Anatomical context of **TP53**
- Associations of **TP53** with chemical compounds
- Physical interactions of **TP53**
- Functional interactions

**Editor**  
Edit this page  
Discussion  
History  
Permanent link

**Special functions**  
Your homepage  
Review process

Print

Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

society  
in  
science  
The Branco Weiss Fellowship

MIT  
Massachusetts  
Institute of Technology

CSAIL/DIG - Decentralized  
Information Group

# Towards Collaborative Publishing in Science

advanced search

**Editor**  
[Edit this page](#)  
[Discussion](#)  
[History](#)  
[Permanent link](#)

**Special functions**  
[Your homepage](#)  
[Review process](#)

[Print](#)

[Tutorial](#)  
[Contact](#)

**Links**  
[NCBI Gene](#)  
[Uniprot](#)  
[SNPedia](#)

**Table of contents**

- Disease relevance of [TP53](#)
- Psychiatry related information on [TP53](#)
- High impact information on [TP53](#)
- Chemical compound and disease context of [TP53](#)
- Biological context of [TP53](#)
- Anatomical context of [TP53](#)
- Associations of [TP53](#) with chemical compounds
- Physical interactions of [TP53](#)
- Enzymatic interactions of [TP53](#)
- Co-localisations of [TP53](#)
- Regulatory relationships of [TP53](#)
- Other interactions of [TP53](#)
- Analytical, diagnostic and therapeutic context of [TP53](#)

**Chemical compound and disease context of TP53**

- We determined the prognostic relevance of high-frequency MSI (MSI-H), [TP53](#) mutations, and [KRAS](#) mutations in a well-defined group of patients with stage III [colon cancer](#) (N = 391), randomly assigned for adjuvant treatment with [fluorouracil](#)-based [chemotherapy](#). [Ref.]
- In conclusion, [TP53](#) gene mutation of the primary tumor is helpful in predicting the response of patients with metastatic [breast disease](#) to [tamoxifen](#) therapy. [Ref.]
- Isogenic HCT116 and HCT116 [TP53](#)-/- [colon cancer](#) cells were exposed to the NO\* donor Sper/NO, H2O2, [hypoxia](#), or [hydroxyurea](#), and their mRNA was analyzed using [oligonucleotide microarrays](#). [Ref.]
- [Methylation](#) of CpG [dinucleotides](#) and/or CCWGG motifs at the promoter of [TP53](#) correlates with decreased [gene expression](#) in a subset of [acute lymphoblastic leukemia](#) patients. [Ref.]
- Two paclitaxel(Ptx)-resistant [ovarian cancer cell lines](#), 1A9/Ptx-10 and 1A9/Ptx-22, isolated from the 1A9 [cell line](#) (a clone of the A2780 line) by continuous exposure to Ptx are characterized by [mutations](#) in their major beta-tubulin gene and in one or both alleles of the [TP53](#) gene. [Methylation of CpG dinucleotides and/or CCWGG motifs at the promoter of TP53 correlates with decreased gene expression in a subset of acute lymphoblastic leukemia patients.](#) Agirre, X., Vizmanos, J.L., Calasanz, M.J., Garcia-Delgado, M., Larráyoz, M.J., Novo, F.J. *Oncogene* (2003)

**Biological context of TP53**

- [EP300 acetylation](#) of [TP53](#) in response to [DNA damage](#) regulates its DNA binding and transcriptional activation functions. [Ref.]
- Here, we determine the evolutionary relationships of non-random [LOH](#), [TP53](#) and [CDKN2A](#) mutations, [CDKN2A CpG-island methylation](#) and [ploidy](#) during neoplastic progression. [Ref.]
- We have previously shown in small numbers of patients that disruption of [TP53](#) and [CDKN2A](#) typically occurs before [aneuploidy](#) and cancer. [Ref.]
- [Diploid](#) cell progenitors with somatic genetic or epigenetic abnormalities in [TP53](#) and [CDKN2A](#) were capable of clonal expansion, spreading to large regions of oesophageal [mucosa](#). [Ref.]
- In this study, four of six myoinvasive TCCs also showed [TP53](#) mutation that associated well with [genome instability](#) (P = 0.001), as previously hypothesized. [Ref.]

**Anatomical context of TP53**

- Thus, our results showed a relatively high frequency of [TP53](#) mutations (76.8%) in our [cell lines](#), with almost half of the mutations being truncating mutations. [Ref.]
- Inactivation of the [ATM](#) or [TP53](#) gene is frequent in [B-cell lymphocytic leukemia](#) (B-CLL) and leads to aggressive disease. [Ref.]
- Hits identified by screening of a genome-scale siRNA library for [cisplatin enhancers](#) in [TP53](#)-deficient [HeLa cells](#) were significantly enriched for genes with annotated functions in [DNA damage](#) repair as well as poorly characterized genes likely having novel functions in this process. [Ref.]
- Activation of nuclear factor kappaB in radioresistance of [TP53](#)-inactive human [keratinocytes](#). [Ref.]
- There was a nonsignificant trend for association between [TP53](#) mutations and bulky adducts in [lymphocyte](#) DNA (OR, 2.78; 95% CI, 0.64-12.17). [Ref.]

**Associations of TP53 with chemical compounds**

- The gene [TP53](#), encoding [p53](#), has a common sequence polymorphism that results in either [proline](#) or [arginine](#) at amino-acid position 72. [Ref.]
- The [in vitro](#) cytotoxicity of a novel [cyclin-dependent kinase inhibitor](#), [CYC202](#), was evaluated in 26 B-CLLs, 11 with mutations in either the [ATM](#) or [TP53](#) genes, and compared with that induced by [ionizing radiation](#) and [fludarabine](#). [Ref.]
- [CYC202](#) induced [apoptosis](#) within 24 hours of treatment in all 26 analyzed tumor samples independently of [ATM](#) and [TP53](#) gene status, whereas 6 of 26 B-CLLs, mostly [ATM](#) mutant, showed marked [in vitro](#) resistance to [fludarabine](#)-induced [apoptosis](#). [Ref.]

# Towards Collaborative Publishing in Science

advanced search

**Editor**  
[Edit this page](#)  
[Discussion](#)  
[History](#)  
[Permanent link](#)

**Special functions**  
[Your homepage](#)  
[Review process](#)

[Print](#)  
[Tutorial](#)  
[Contact](#)

**Links**  
[NCBI Gene](#)  
[Uniprot](#)  
[SNPedia](#)

**Table of contents**

- Disease relevance of [TP53](#)
- Psychiatry related information on [TP53](#)
- High impact information on [TP53](#)
- Chemical compound and disease context of [TP53](#)
- Biological context of [TP53](#)
- Anatomical context of [TP53](#)
- Associations of [TP53](#) with chemical compounds
- Physical interactions of [TP53](#)
- Enzymatic interactions of [TP53](#)
- Co-localisations of [TP53](#)
- Regulatory relationships of [TP53](#)
- Other interactions of [TP53](#)
- Analytical, diagnostic and therapeutic context of [TP53](#)

**Analytical, diagnostic and therapeutic context of TP53**

- **Point mutations** within the **TP53** gene were detected by use of **polymerase chain reaction (PCR)** in combination with constant denaturant gel electrophoresis. [Ref.]
- To determine the **MDM2** and **TP53** mRNA levels, **Northern-blot** analysis was performed. [Ref.]
- Identification of mutations in the **tumor suppressor gene TP53** has implications for the **molecular epidemiology** and for the molecular pathology of human cancer. [Ref.]
- Results of **gene-targeting** studies have demonstrated that **p63**, a homologue of the **cell-cycle** regulator **TP53**, plays a critically important role in regulation of the formation and differentiation of the AER. [Ref.]
- **Microarray analysis** reveals that **TP53**- and **ATM**-mutant B-CLLs share a defect in activating proapoptotic responses after **DNA damage** but are distinguished by major differences in activating prosurvival responses. [Ref.]

**References**

- **Frequent somatic mutations in PTEN and TP53 are mutually exclusive in the stroma of breast carcinomas.** Kurose, K., Gilley, K., Matsumoto, S., Watson, P.H., Zhou, X.P., Eng, C. *Nat. Genet.* (2002)
- **Augmentation of tumor angiogenesis by a Myc-activated microRNA cluster.** Dews, M., Homayouni, A., Yu, D., Murphy, D., Sevignani, C., Wentzel, E., Furth, E.E., Lee, W.M., Enders, G.H., Mendell, J.T., Thomas-Tikhonenko, A. *Nat. Genet.* (2006)
- **Nf1;Trp53 mutant mice develop glioblastoma with evidence of strain-specific effects.** Reilly, K.M., Loisel, D.A., Bronson, R.T., McLaughlin, M.E., Jacks, T. *Nat. Genet.* (2000)
- **Heterozygous germ line hCHK2 mutations in Li-Fraumeni syndrome.** Bell, D.W., Varley, J.M., Szydio, T.E., Kang, D.H., Wahrer, D.C., Shannon, K.E., Lubratovich, M., Verselis, S.J., Isselbacher, K.J., Fraumeni, J.F., Birch, J.M., Li, F.P., Garber, J.E., Haber, D.A. *Science* (1999)
- **The tumor suppressors Ink4c and p53 collaborate independently with Patched to suppress medulloblastoma formation.** Uziel, T., Zindy, F., Xie, S., Lee, Y., Forget, A., Magdaleno, S., Reh, J.E., Calabrese, C., Solecki, D., Eberhart, C.G., Sherr, S.E., Plimner, S., Clifford, S.C., Hatten, M.E., McKinnon, P.J., Gilbertson, R.J., Curran, T., Sherr, C.J., Roussel, M.F. *Genes Dev.* (2005)
- **Human papillomavirus type 16 and TP53 mutation in oral cancer: matched analysis of the IARC multicenter study.** Dai, M., Clifford, G.M., le Calvez, F., Castellsagué, X., Snijders, P.J., Pawlita, M., Herrero, R., Hainaut, P., Franceschi, S. *Cancer Res.* (2004)
- **Tumor suppressor gene TP53 is genetically associated with schizophrenia in the Chinese population.** Yang, Y., Xiao, Z., Chen, W., Sang, H., Guan, Y., Peng, Y., Zhang, D., Gu, Z., Qian, M., He, G., Qin, W., Li, D., Gu, N., He, L. *Neurosci. Lett.* (2004)
- **The Huntington's disease protein interacts with p53 and CREB-binding protein and represses transcription.** Steffan, J.S., Kazantsev, A., Spasic-Boskovic, O., Greenwald, M., Zhu, Y.Z., Gohler, H., Wanker, E.E., Bates, G.P., Housman, D.E., Thompson, L.M. *Proc. Natl. Acad. Sci. U.S.A.* (2000)
- **Synergistic and opposing regulation of the stress-responsive gene IEX-1 by p53, c-Myc, and multiple NF-kappaB/rel complexes.** Huang, Y.H., Wu, J.Y., Zhang, Y., Wu, M.X. *Oncogene* (2002)
- **Tauroursodeoxycholic acid modulates p53-mediated apoptosis in Alzheimer's disease mutant neuroblastoma cells.** Ramalho, R.M., Borralho, P.M., Castro, R.E., Solá, S., Steer, C.J., Rodrigues, C.M. *J. Neurochem.* (2006)

# Towards Collaborative Publishing in Science

The screenshot shows the Nature Genetics journal website. The main article is a commentary titled "Recommendations of the 2006 Human Variome Project meeting" by Richard G.H. Cotton and participants of the 2006 Human Variome Project meeting. The article lists 43 recommendations from various international institutions, including the Victorian Partnership for Advanced Computing, The Rockefeller University, National Institute on Aging, Wellcome Trust, and many others. The article also discusses the challenges of standardizing genomic data and the need for a universally accessible system, leading to the World Health Organization-sponsored meeting in Melbourne, Australia, which launched the Human Variome Project.

**Journal content**  
Journal home  
Advance online publication  
Current issue  
Archive  
Focuses and Supplements  
Press releases  
Free Association (blog)

**Journal information**  
Guide to authors  
Online submission  
Permissions  
For referees  
Free online issue  
Contact the journal  
Subscribe  
About this site

**NPG services**  
Authors & Referees  
Advertising  
work@npg  
natureprints  
For librarians

**NPG resources**  
Nature  
Nature Biotechnology  
Nature Cell Biology  
Nature Medicine  
Nature Methods  
Nature Reviews Cancer  
Nature Reviews Genetics  
Nature Reviews Molecular Cell Biology  
Nature Reports Stem Cells  
Nature Precedings  
Nature News  
Nature Conferences

**NPG Journals**  
By Subject Area  
Chemistry  
Chemistry  
Drug discovery  
Biotechnology  
Materials  
Methods & Protocols  
Clinical Practice & Research  
Cancer  
Cardiovascular

**Commentary**  
Nature Genetics **39**, 433–436 (2007)  
Published online: 28 March 2007 | doi:10.1038/ng2024

**Recommendations of the 2006 Human Variome Project meeting**  
Richard G.H. Cotton<sup>1,2</sup> & participants of the 2006 Human Variome Project meeting

1. Victorian Partnership for Advanced Computing, Melbourne, Victoria, Australia.
2. The Rockefeller University, New York, New York, USA.
3. National Institute on Aging, Bethesda, Maryland, USA.
4. Weatherall Institute of Molecular Medicine, Oxford, UK.
5. National Center for Health Marketing, US Centers for Disease Control and Prevention, Atlanta, Georgia, USA.
6. World Health Organization, Geneva, Switzerland.
7. Institute of Genomics and Integrative Biology, Delhi, India.
8. National Human Genome Research Institute, Bethesda, Maryland, USA.
9. University of Leicester, Leicester, UK.
10. Medical Research Council, Human Genetics Unit, Edinburgh, Scotland, UK.
11. University of Washington, Seattle, Washington, USA.
12. Universidad de Guadalajara, Guadalajara, Mexico.
13. Center for Human Genetics, University of Leuven, Leuven, Belgium.
14. CNRS de Montpellier, Montpellier, France.
15. Benaroya Research Institute at Virginia Mason, Seattle, Washington, USA.
16. Genomic Disorders Research Centre, Melbourne, Victoria, Australia.
17. Leiden University Medical Center, Leiden, The Netherlands.
18. European Molecular Biology Laboratory, European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge, UK.
19. Baylor College of Medicine, Baylor, Texas, USA.
20. University of British Columbia, Vancouver, British Columbia, Canada.
21. Natural Sciences Sector, United Nations Educational, Scientific and Cultural Organization (UNESCO), Paris, France.
22. March of Dimes Birth Defects Foundation, Mamaroneck, New York, USA.
23. University of California San Francisco, San Francisco, California, USA.
24. Faculté de Pharmacie, Monastir, Tunisia.
25. Karolinska Institute, Stockholm, Sweden.
26. National Center for Biotechnology Information (NCBI), US National Institutes of Health (NIH), Bethesda, Maryland, USA.
27. Anthony Nolan Research Institute, UK.
28. African Institute of Biomedical Science and Technology, Harare, Zimbabwe.
29. Hamamatsu University School of Medicine, Hamamatsu, Japan.
30. Centro de Estudio de las Metabólicas Congénitas (CEMECO), National University of Córdoba and Santísima Trinidad Children's Hospital, Córdoba, Argentina.
31. University of Washington, Seattle, Washington, USA.
32. University of Cape Town, Cape Town, South Africa.
33. Royal Perth Hospital, Perth, Western Australia, Australia.
34. Oregon Health & Science University, Portland, Oregon, USA.
35. Cedars-Sinai Medical Center, Los Angeles, California, USA.
36. NIH Pharmacogenetics Research Network, Bethesda, Maryland, USA.
37. Montreal Children's Hospital Research Institute, Montreal, Quebec, Canada.
38. Keio University School of Medicine, Tokyo, Japan.
39. Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, USA.
40. Centre for Arab Genomic Studies, Dubai, United Arab Emirates.
41. St. James' University Hospital, Leeds, UK.
42. American College of Medical Genetics, Bethesda, Maryland, USA.
43. Richard G.H. Cotton is at the Genomic Disorders Research Centre, St. Vincent's Hospital Melbourne, 25 Victoria Parade, Melbourne, Victoria 3065, Australia. The complete list of the authors appears at the end of the paper. e-mail: rcotton@unimelb.edu.au

Lists of variations in genomic DNA and their effects have been kept for some time and have been used in diagnostics and research. Although these lists have been carefully gathered and curated, there has been little standardization and coordination, complicating their use. Given the myriad possible variations in the estimated 24,000 genes in the human genome, it would be useful to have standard criteria for databases of variation. Incomplete collection and ascertainment of variants demonstrates a need for a universally accessible system. These and other problems led to the World Health Organization–cosponsored meeting on June 20–23, 2006 in Melbourne, Australia, which launched the Human Variome Project. This meeting addressed all areas of human genetics relevant to collection of information on variation and its effects. Members of each of eight sessions (the clinic and phenotype, the diagnostic laboratory, the research laboratory, curation and collection, informatics, relevance to the emerging world, integration and federation and funding and sustainability) developed a number of recommendations that were then organized into a total of 96 recommendations to act as a foundation for future work worldwide. Here we summarize the background of the project, the meeting and its recommendations.

**Subscribe to Nature Genetics**  
Subscribe

**This issue**  
Table of contents  
Previous article  
Next article

**Article tools**  
Download PDF  
Send to a friend  
Articles in CrossRef citing this article  
Export citation  
Export references  
Rights and permissions  
Order commercial reprints  
Save this link

**Article navigation**  
Acknowledgments  
Supplementary info  
References

**Search PubMed for**  
Richard G.H. Cotton  
William Applebe  
Arielen O Auerbach  
Kevin Becker  
Walter Bodmer  
D Joe Boone  
more authors of this article

**naturejobs**  
Postdoctoral Research Officer  
University of Surrey  
Guildford United Kingdom  
Home-Based Clinical Research  
Associate (CRA) — Colorado  
— IMMEDIATE NEED!!  
KSR  
Staring Heights, MI, USA  
More science jobs

**natureproducts**  
search buyers guide

ADVERTISEMENT  
WWW.TIGM.ORG

# Towards Collaborative Publishing in Science

The image shows a screenshot of the Nature Genetics journal website. A large red question mark is overlaid on the page, with red lines pointing to various sections and text. The main content area features a 'Commentary' by Richard G.H. Cotton et al. titled 'Recommendations of the 2006 Human Variome Project meeting'. The text discusses the challenges of genomic DNA variation and the need for a standardized system. The left sidebar contains navigation links like 'Journal content', 'Journal information', and 'NPG journals'. The right sidebar includes 'Subscribe to Nature Genetics', 'This issue', 'Article tools', and 'Search PubMed for'. A red box highlights a paragraph in the main text that discusses the need for a standardized system for genomic DNA variation.

**Commentary**  
Nature Genetics 39, 433–436 (2007)  
Published online: 28 March 2007 | doi:10.1038/ng2024

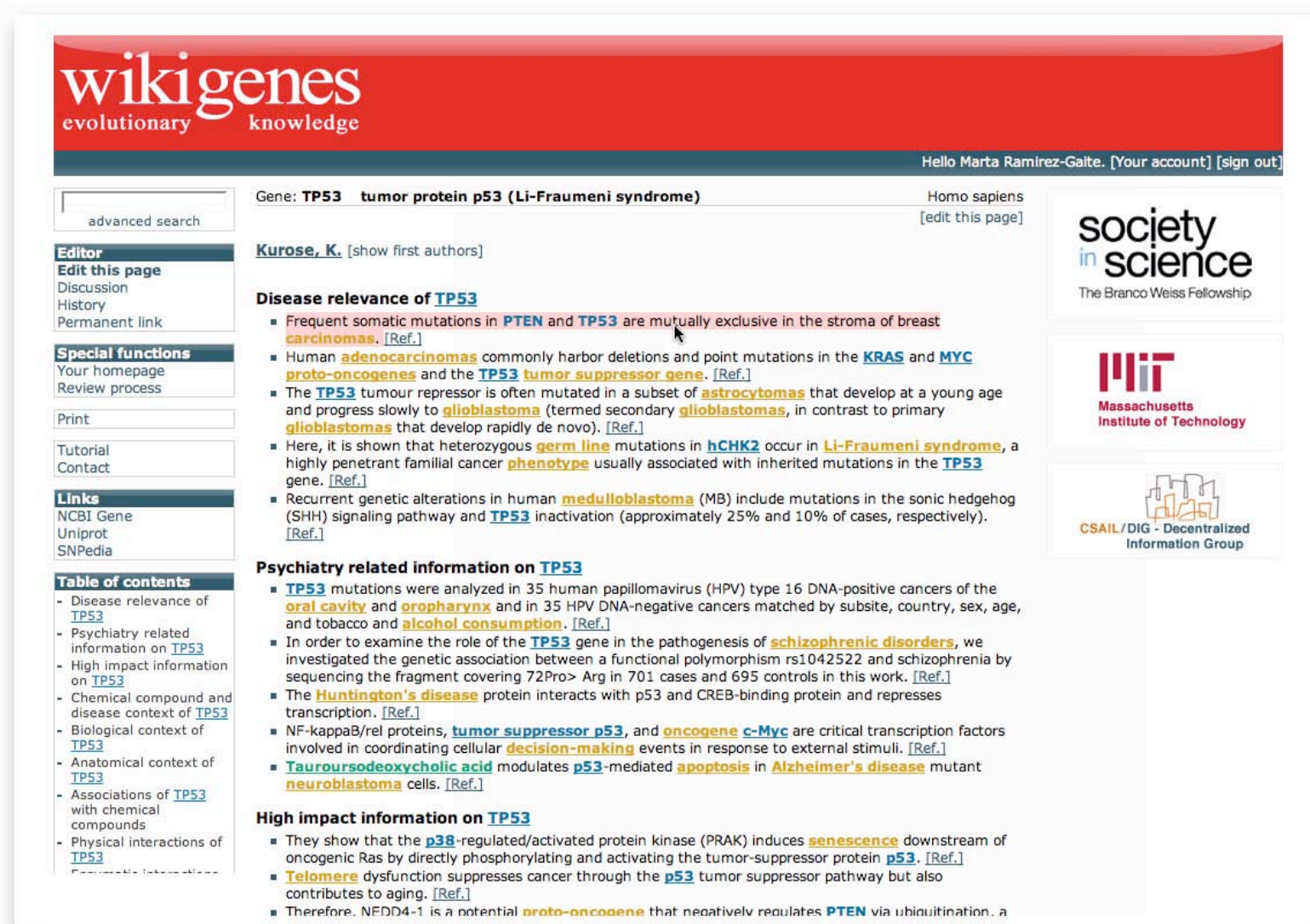
**Recommendations of the 2006 Human Variome Project meeting**  
Richard G.H. Cotton<sup>1,2</sup> & participants of the 2006 Human Variome Project meeting

1. Victorian Partnership for Advanced Computing, Melbourne, Victoria, Australia.
2. The Rockefeller University, New York, New York, USA.
3. National Institute of Biomedical Genomics, Bethesda, Maryland, USA.
4. Weatherall Institute of Molecular Medicine, Oxford, UK.
5. National Center for Health Marketing, US Centers for Disease Control and Prevention, Atlanta, Georgia, USA.
6. World Health Organization, Geneva, Switzerland.
7. Institute of Genomics and Integrative Biology, Delhi, India.
8. National Human Genome Research Institute, Bethesda, Maryland, USA.
9. University of Leicester, Leicester, UK.
10. Medical Research Council, Human Genetics Unit, Edinburgh, Scotland, UK.
11. University of Washington, Seattle, Washington, USA.
12. Universidad de Guadalajara, Guadalajara, Mexico.
13. Center for Human Genetics, University of Leuven, Leuven, Belgium.
14. CNRS de Montpellier, Montpellier, France.
15. Benaroya Research Institute at Virginia Mason, Seattle, Washington, USA.
16. Genomic Disorders Research Centre, Melbourne, Victoria, Australia.
17. Leiden University Medical Center, Leiden, The Netherlands.
18. European Molecular Biology Laboratory, European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge, UK.
19. Baylor College of Medicine, Baylor, Texas, USA.
20. University of British Columbia, Vancouver, British Columbia, Canada.
21. Natural Sciences Sector, United Nations Educational, Scientific and Cultural Organization (UNESCO), Paris, France.
22. March of Dimes Birth Defects Foundation, Mamaroneck, New York, USA.
23. University of California San Francisco, San Francisco, California, USA.
24. Faculté de Pharmacie, Monastir, Tunisia.
25. Karolinska Institutet, Stockholm, Sweden.
26. National Center for Biotechnology Information (NCBI), US National Institutes of Health (NIH), Bethesda, Maryland, USA.
27. Anthony Nolan Research Institute, UK.
28. African Institute of Biomedical Science and Technology, Harare, Zimbabwe.
29. Hamamatsu University School of Medicine, Hamamatsu, Japan.
30. Centro de Estudio de las Metabólicas Congénitas (CEMECO), National University of Córdoba and Santísima Trinidad Children's Hospital, Córdoba, Argentina.
31. University of Washington, Seattle, Washington, USA.
32. University of Cape Town, Cape Town, South Africa.
33. Royal Perth Hospital, Perth, Western Australia, Australia.
34. Oregon Health & Science University, Portland, Oregon, USA.
35. Cedars-Sinai Medical Center, Los Angeles, California, USA.
36. NIH Pharmacogenetics Research Network, Bethesda, Maryland, USA.
37. Montreal Children's Hospital Research Institute, Montreal, Quebec, Canada.
38. Keio University School of Medicine, Tokyo, Japan.
39. Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, USA.
40. Centre for Arab Genomic Studies, Dubai, United Arab Emirates.
41. St. James' University Hospital, Leeds, UK.
42. American College of Medical Genetics, Bethesda, Maryland, USA.
43. Richard G.H. Cotton is at the Genomic Disorders Research Centre, St. Vincent's Hospital Melbourne, 25 Victoria Parade, Melbourne, Victoria 3065, Australia. The complete list of the authors appears at the end of the paper. e-mail: [cotton@unimelb.edu.au](mailto:cotton@unimelb.edu.au)

Lists of variations in genomic DNA and their effects have been kept for some time and have been used in diagnostics and research. Although these lists have been carefully gathered and curated, there has been little standardization and coordination, complicating their use. Given the myriad possible variations in the estimated 24,000 genes in the human genome, it would be useful to have standard criteria for databases of variation. Incomplete collection and ascertainment of variants demonstrates a need for a universally accessible system. These and other problems led to the World Health Organization–cosponsored meeting on June 20–23, 2006 in Melbourne, Australia, which launched the Human Variome Project. This meeting addressed all areas of human genetics relevant to collection of information on variation and its effects. Members of each of eight sessions (the clinic and phenotype, the diagnostic laboratory, the research laboratory, curation and collection, informatics, relevance to the emerging world, integration and federation and funding and sustainability) developed a number of recommendations that were then organized into a total of 96 recommendations to act as a foundation for future work worldwide. Here we summarize the background of the project, the meeting and its recommendations.



# Towards Collaborative Publishing in Science



**wiki**genes  
evolutionary knowledge

Hello Marta Ramirez-Gaité. [Your account] [sign out]

Gene: **TP53** tumor protein p53 (Li-Fraumeni syndrome) Homo sapiens  
[edit this page]

**Kurose, K.** [show first authors]

**Disease relevance of TP53**

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast carcinomas. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC** proto-oncogenes and the **TP53** tumor suppressor gene. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

**Psychiatry related information on TP53**

- TP53** mutations were analyzed in 35 human papillomavirus (HPV) type 16 DNA-positive cancers of the **oral cavity** and **oropharynx** and in 35 HPV DNA-negative cancers matched by subsite, country, sex, age, and tobacco and **alcohol consumption**. [Ref.]
- In order to examine the role of the **TP53** gene in the pathogenesis of **schizophrenic disorders**, we investigated the genetic association between a functional polymorphism rs1042522 and schizophrenia by sequencing the fragment covering 72Pro> Arg in 701 cases and 695 controls in this work. [Ref.]
- The **Huntington's disease** protein interacts with p53 and CREB-binding protein and represses transcription. [Ref.]
- NF-kappaB/rel proteins, **tumor suppressor p53**, and **oncogene c-Myc** are critical transcription factors involved in coordinating cellular **decision-making** events in response to external stimuli. [Ref.]
- Tauroursodeoxycholic acid** modulates **p53**-mediated **apoptosis** in **Alzheimer's disease** mutant **neuroblastoma** cells. [Ref.]

**High impact information on TP53**

- They show that the **p38**-regulated/activated protein kinase (PRAK) induces **senescence** downstream of oncogenic Ras by directly phosphorylating and activating the tumor-suppressor protein **p53**. [Ref.]
- Telomere** dysfunction suppresses cancer through the **p53** tumor suppressor pathway but also contributes to aging. [Ref.]
- Therefore, NEFD4-1 is a potential **proto-oncogene** that negatively regulates **PTEN** via ubiquitination. a

**Table of contents**

- Disease relevance of **TP53**
- Psychiatry related information on **TP53**
- High impact information on **TP53**
- Chemical compound and disease context of **TP53**
- Biological context of **TP53**
- Anatomical context of **TP53**
- Associations of **TP53** with chemical compounds
- Physical interactions of **TP53**
- Evolutionary interactions

**Editor**  
Edit this page  
Discussion  
History  
Permanent link

**Special functions**  
Your homepage  
Review process

Print

Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

society  
in  
science  
The Branco Weiss Fellowship

MIT  
Massachusetts  
Institute of Technology

CSAIL/DIG - Decentralized  
Information Group

# Towards Collaborative Publishing in Science

**wiki**genes  
evolutionary knowledge

Hello Marta Ramirez-Gaité. [Your account] [sign out]

Gene: **TP53** tumor protein p53 (Li-Fraumeni syndrome) Homo sapiens  
[edit this page]

Reilly, K.M. [show first authors]

**Disease relevance of TP53**

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC** **proto-oncogenes** and the **TP53** **tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

**Psychiatry related information on TP53**

- **TP53** mutations were analyzed in 35 human papillomavirus (HPV) type 16 DNA-positive cancers of the **oral cavity** and **oropharynx** and in 35 HPV DNA-negative cancers matched by subsite, country, sex, age, and tobacco and **alcohol consumption**. [Ref.]
- In order to examine the role of the **TP53** gene in the pathogenesis of **schizophrenic disorders**, we investigated the genetic association between a functional polymorphism rs1042522 and schizophrenia by sequencing the fragment covering 72Pro> Arg in 701 cases and 695 controls in this work. [Ref.]
- The **Huntington's disease** protein interacts with p53 and CREB-binding protein and represses transcription. [Ref.]
- NF-kappaB/rel proteins, **tumor suppressor p53**, and **oncogene c-Myc** are critical transcription factors involved in coordinating cellular **decision-making** events in response to external stimuli. [Ref.]
- **Tauroursodeoxycholic acid** modulates **p53**-mediated **apoptosis** in **Alzheimer's disease** mutant **neuroblastoma** cells. [Ref.]

**High impact information on TP53**

- They show that the **p38**-regulated/activated protein kinase (PRAK) induces **senescence** downstream of oncogenic Ras by directly phosphorylating and activating the tumor-suppressor protein **p53**. [Ref.]
- **Telomere** dysfunction suppresses cancer through the **p53** tumor suppressor pathway but also contributes to aging. [Ref.]
- Therefore, NEFD4-1 is a potential **proto-oncogene** that negatively regulates **PTEN** via ubiquitination. a

**Table of contents**

- Disease relevance of **TP53**
- Psychiatry related information on **TP53**
- High impact information on **TP53**
- Chemical compound and disease context of **TP53**
- Biological context of **TP53**
- Anatomical context of **TP53**
- Associations of **TP53** with chemical compounds
- Physical interactions of **TP53**
- Functional interactions

**Editor**  
Edit this page  
Discussion  
History  
Permanent link

**Special functions**  
Your homepage  
Review process

Print

Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

society  
in  
science  
The Branco Weiss Fellowship

MIT  
Massachusetts  
Institute of Technology

CSAIL/DIG - Decentralized  
Information Group

## Towards Collaborative Publishing in Science

---

[Nath, N.](#), [Muranaka, T.](#), [Hardie, D.G.](#), [Palecek, S.P.](#), [Frederick, D.L.](#), et al.

### Disease relevant


- Together, these metabolic alterations mediated by [SNF1](#) are an important component of ir at the inactivation of ADK and [SNF1](#) by the [geminivirus](#) proteins represents a [cellular strategy to counter](#) this defense [\[Ref\]](#).
- In yeast, [SNF1](#) is one of the main regulators in the shift from fermentation to aerobic metabolism; AMPK, its mammalian counterpart, is a master metabolic regulator involved in a variety of metabolic disorders such as diabetes and [obesity](#) [\[Ref\]](#).
- A [snf1](#) null mutant is sensitive to [heat stress](#) and starvation and fails to accumulate glycogen during growth in rich medium [\[Ref\]](#).

Rate this author's contribution



[Author's homepage] 4 stars  
[Contributions][Contact]

# Towards Collaborative Publishing in Science

[sign in]

**Editor**  
Edit this page  
Discussion  
History  
Permanent link

**Special functions**  
Print  
Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

**Table of contents**

- Disease relevance of [TP53](#)
- Psychiatry related information on [TP53](#)
- High impact information on [TP53](#)
- Chemical compound and disease context of [TP53](#)
- Biological context of [TP53](#)
- Anatomical context of [TP53](#)
- Associations of [TP53](#) with chemical compounds
- Physical interactions of [TP53](#)
- Enzymatic interactions of [TP53](#)
- Colocalizations of [TP53](#)

Gene: **TP53** **tumor protein p53 (Li-Fraumeni syndrome)** Homo sapiens  
[\[edit this page\]](#)

**Ramirez-Gaite, M.** [\[show first authors\]](#)

**Disease relevance of TP53**




- Frequent somatic mutations in [PTEN](#) and [TP53](#) are mutually exclusive in the stroma of breast [carcinomas](#). [\[Ref.\]](#)
- Human [adenocarcinomas](#) commonly harbor deletions and point mutations in the [KRAS](#) and [MYC proto-oncogenes](#) and the [TP53 tumor suppressor gene](#). [\[Ref.\]](#)
- The [TP53](#) tumour repressor is often mutated in a subset of [astrocytomas](#) that develop at a young age and progress slowly to [glioblastoma](#) (termed secondary [glioblastomas](#), in contrast to primary [glioblastomas](#) that develop rapidly de novo). [\[Ref.\]](#)
- Here, it is shown that heterozygous [germ line](#) mutations in [hCHK2](#) occur in [Li-Fraumeni syndrome](#), a highly penetrant familial cancer [phenotype](#) usually associated with inherited mutations in the [TP53](#) gene. [\[Ref.\]](#)
- Recurrent genetic alterations in human [medulloligodendrocyte](#) [SHH](#) signaling pathway and [TP53](#) inactivation ([\[Hold mouse button down for author info.\]](#) [\[Ref.\]](#)).

**Psychiatry related information on TP53**

- [TP53](#) mutations were analyzed in 35 human papillomavirus (HPV) type 16 DNA-positive cancers of the [oral cavity](#) and [oropharynx](#) and in 35 HPV DNA-negative cancers matched by subsite, country, sex, age, and tobacco and [alcohol consumption](#). [\[Ref.\]](#)
- In order to examine the role of the [TP53](#) gene in the pathogenesis of [schizophrenic disorders](#), we investigated the genetic association between a functional polymorphism rs1042522 and schizophrenia by sequencing the fragment covering 72Pro> Arg in 701 cases and 695 controls in this work. [\[Ref.\]](#)
- The [Huntington's disease](#) protein interacts with p53 and CREB-binding protein and represses transcription. [\[Ref.\]](#)
- NF-kappaB/rel proteins, [tumor suppressor p53](#), and [oncogene c-Myc](#) are critical transcription factors involved in coordinating cellular [decision-making](#) events in response to external stimuli. [\[Ref.\]](#)
- [Tauroursodeoxycholic acid](#) modulates [p53](#)-mediated [apoptosis](#) in [Alzheimer's disease](#) mutant [neuroblastoma](#) cells. [\[Ref.\]](#)

**High impact information on TP53**

- They show that the [p38](#)-regulated/activated protein kinase (PRAK) induces [senescence](#) downstream of oncogenic Ras by directly phosphorylating and activating the tumor-suppressor protein [p53](#). [\[Ref.\]](#)
- [Telomere](#) dysfunction suppresses cancer through the [p53](#) tumor suppressor pathway but also contributes to aging. [\[Ref.\]](#)
- Therefore, NFDD4-1 is a potential [proto-oncogene](#) that negatively regulates [PTEN](#) via ubiquitination. a



# Towards Collaborative Publishing in Science

The image is a collage illustrating the flow of scientific information. On the left, a tablet displays the Wikigenes website, which provides a structured overview of a research paper. In the center, a research paper titled "Genetic architecture of KRAS in human colorectal cancer" is shown, featuring a table of contents and a main text area with a blue arrow pointing from the Wikigenes tablet. On the right, a computer monitor displays the NCBI Entrez Gene database page for the KRAS gene, showing its official name, symbol, and various annotations. The background includes logos for Wikigenes, Society for Science & Public Policy, and MIT.

# Towards Collaborative Publishing in Science

The screenshot displays the WikiGenes website interface. At the top, the logo reads "wikigenes evolutionary knowledge". A user notification in the top right corner says "Hello Marta Ramirez-Gaité. [Your account] [sign out]".

On the left sidebar, there are sections for "Save this page" (with links for "Cancel changes" and "Preferences"), "Tutorial Contact", and "Links" (with links for "NCBI Gene", "Uniprot", and "SNPedia"). Below this is a "watch the WikiGenes Intro guided tour" link with a filmstrip icon.

The main content area is titled "Gene: TP53 tumor protein p53 (Li-Fraumeni syndrome)" and "Homo sapiens". It features a section "Disease relevance of TP53" with a bulleted list of scientific findings:

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC proto-oncogenes** and the **TP53 tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

Below the text is a section "Psychiatry related information on TP53".

At the bottom of the page, there is a navigation bar with icons for "AA", "I", "gene", "chemical", "MeSH", and "PubMed reference". Below this is an "Edit summary:" text box and two buttons: "[Save this page]" and "[Cancel changes]".

On the right side of the page, there are three logos: "society in science The Branco Weiss Fellowship", "MIT Massachusetts Institute of Technology", and "CSAIL/DIG - Decentralized Information Group".

# Towards Collaborative Publishing in Science

The screenshot displays the WikiGenes website interface. At the top, the logo reads "wikigenes evolutionary knowledge". A user notification says "Hello Marta Ramirez-Gaité. [Your account] [sign out]". The main content area is for the gene "TP53 tumor protein p53 (Li-Fraumeni syndrome)" in "Homo sapiens".

**Save this page**  
Cancel changes  
Preferences

Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

watch the  
**WikiGenes Intro**  
guided tour

**Disease relevance of TP53**

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC proto-oncogenes** and the **TP53 tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

**Psychiatry related information on TP53**

AA / [edit icons] X<sub>2</sub> X<sup>2</sup> Ω [share icons]

gene [DNA helix icon] chemical [molecule icon] MeSH [tree icon] PubMed reference [PubMed logo]

Define this term as a gene

Cur summary:

[Save this page]  
[Cancel changes]

society in science  
The Branco Weiss Fellowship

MIT  
Massachusetts  
Institute of Technology

CSAIL/DIG - Decentralized  
Information Group

# Towards Collaborative Publishing in Science

The screenshot shows the WikiGenes database search interface. At the top, the 'wikigenes' logo is visible. Below it, a search bar contains the text 'KRAS'. To the right of the search bar are dropdown menus for 'genes and proteins', 'everywhere', and 'every organism'. The search results are displayed in a table-like format with columns for gene names, descriptions, and organisms. The results include entries for **Kras** (Mus musculus), **KRAS** (Homo sapiens), **Kras** (Rattus norvegicus), **HRAS** (Homo sapiens), **Hras1** (Mus musculus), and **TP53** (Homo sapiens). Each entry includes a brief description and synonyms. On the right side of the search results, there are three logos: 'society in science The Branco Weiss Fellowship', 'MIT Massachusetts Institute of Technology', and 'CSAIL/DIG - Decentralized Information Group'. On the left side, there are navigation links such as 'Save', 'Cancel', 'Preferences', 'Tutorial', 'Contact', 'Links', 'NCBI', 'Uniprot', and 'SNPedia'.

Gene Name	Description	Organism
<b>Kras</b>	v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog <b>Gene:</b> ...after irradiation. Other interactions of <b>Kras</b> There appears to be selectivity in the activated <b>gene</b> ...Disease relevance of <b>Kras</b> Activated <b>Kras</b> and Ink4a/ Arf deficiency cooperate to produce metastatic pancreatic ductal adenocarcinoma. Somatic activation of oncogenic <b>Kras</b> in hematopoietic cells initiates a... / Synonyms: -K-rasGTPase <b>KRAS</b> ; GTPase <b>KRAS</b> precursor; -Ras <b>K-ras</b>	Mus musculus
<b>KRAS</b>	v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog <b>Gene:</b> ... <b>KRAS</b> mutations cause Noonan syndrome. Furthermore, we found that whereas a <b>gene</b> - expression signature...alone, integrating mouse and human data, we discovered a <b>gene</b> - expression signature of <b>KRAS2</b> mutation in human lung cancer. An oncogenic <b>KRAS2</b> expression signature identified by cross- species <b>gene</b> - expression... / Synonyms: -K-RASGTPase <b>KRAS</b> ; GTPase <b>KRAS</b> precursor; -RAS <b>KRAS1</b>	Homo sapiens
<b>Kras</b>	v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog <b>Gene:</b> ...- ras <b>gene</b> . Anatomical context of <b>Kras</b> The period of maximal differentiation between days 9 to 13...Disease relevance of <b>Kras</b> 3- Hydroxy- 3- methylglutaryl CoA reductase inhibitors prevent high...into the rat pheochromocytoma( PC12) neural cell line. High impact information on <b>Kras</b> Although p21... / Synonyms: -K-rasGTPase <b>KRAS</b> ; GTPase <b>KRAS</b> precursor; -Ras <b>Kras2</b>	Rattus norvegicus
<b>HRAS</b>	v-Ha-ras Harvey rat sarcoma viral oncogene homolog <b>Gene:</b> ...and by immunoblotting using monoclonal antibodies recognizing the HRAS and <b>KRAS gene</b> products. We have tested...guanine nucleotide bound to p21ras. Enzymatic interactions of HRAS We then determined whether the <b>KRAS</b> ...protein and calmodulin are phosphorylated by the receptor kinase in the presence of the <b>KRAS</b> basic domain... / Synonyms: <b>K-ras</b>	Homo sapiens
<b>Hras1</b>	Harvey rat sarcoma virus oncogene 1 <b>Gene:</b> ...with the HPV- 16 E7 <b>gene</b> and the activated c- H- ras <b>gene</b> fall into two distinct phenotypic classes. Finally...or the polyoma virus middle T <b>gene</b> increases cell permissiveness to parvovirus minute- virus- of- mice. High...in the IRF- 1 <b>gene</b> ( IRF- 1 -/- mice) can be transformed by expression of an activated c- Ha- ras oncogene... / Synonyms: <b>Kras2</b>	Mus musculus
<b>TP53</b>	tumor protein p53 (Li-Fraumeni syndrome) <b>Gene:</b> ...in the stroma of breast carcinomas. Human adenocarcinomas commonly harbor mutations in the <b>KRAS</b> and MYC proto- oncogenes and the TP53 tumor suppressor <b>gene</b> . The TP53 tumour suppressor is often mutated in a subset...- frequency MSI( MSI- H ), TP53 mutations, and <b>KRAS</b> mutations in a well- defined group of patients with stage...	Homo sapiens



# Towards Collaborative Publishing in Science

The screenshot displays the WikiGenes website interface. At the top, the logo reads "wikigenes evolutionary knowledge". A user notification in the top right corner says "Hello Marta Ramirez-Gaité. [Your account] [sign out]".

On the left sidebar, there are sections for "Save this page" (with links for "Cancel changes" and "Preferences"), "Tutorial Contact", and "Links" (with links for "NCBI Gene", "Uniprot", and "SNPedia"). Below this is a "watch the WikiGenes Intro guided tour" link with a filmstrip icon.

The main content area is titled "Gene: TP53 tumor protein p53 (Li-Fraumeni syndrome) Homo sapiens". It features a "Disease relevance of TP53" section with a bulleted list of findings:

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC proto-oncogenes** and the **TP53 tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** that develop at a young age and progress slowly to **glioblastoma** (termed secondary **glioblastomas**, in contrast to primary **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

Below this is a section for "Psychiatry related information on TP53" with a link to "Selected gene: KRAS- v-Ki-ras2 Kirsten rat sarcoma viral onco... (Homo sapiens) [NCBI Gene]".

At the bottom of the main content area, there is a navigation bar with icons for "AA", "I", "gene" (with a DNA helix icon), "chemical" (with a molecular structure icon), "MeSH", and "PubMed reference".

At the very bottom, there is an "Edit summary:" text box, a "[Save this page]" button, and a "[Cancel changes]" link.

On the right side of the page, there are three logos: "society in science The Branco Weiss Fellowship", "MIT Massachusetts Institute of Technology", and "CSAIL/DIG - Decentralized Information Group".

# Towards Collaborative Publishing in Science

**wiki**genes  
evolutionary knowledge

[sign in]

Gene: **TP53 tumor protein p53 (Li-Fraumeni syndrome)** Homo sapiens  
[edit this page]

Barrett, M.T., Alvi, A.J., Berns, E.M., Stankovic, T., Balachandran, R., et al.

**Disease relevance of TP53**

- Frequent somatic mutations in **PTEN** and **TP53** are mutually exclusive in the stroma of breast **carcinomas**. [Ref.]
- Human **adenocarcinomas** commonly harbor deletions and point mutations in the **KRAS** and **MYC proto-oncogenes** and the **TP53 tumor suppressor gene**. [Ref.]
- The **TP53** tumour repressor is often mutated in a subset of **astrocytomas** and progress slowly to **glioblastoma** (termed secondary **glioblastomas** and **glioblastomas** that develop rapidly de novo). [Ref.]
- Here, it is shown that heterozygous **germ line** mutations in **hCHK2** occur in **Li-Fraumeni syndrome**, a highly penetrant familial cancer **phenotype** usually associated with inherited mutations in the **TP53** gene. [Ref.]
- Recurrent genetic alterations in human **medulloblastoma** (MB) include mutations in the sonic hedgehog (SHH) signaling pathway and **TP53** inactivation (approximately 25% and 10% of cases, respectively). [Ref.]

**Psychiatry related information on TP53**

- TP53** mutations were analyzed in 35 human papillomavirus (HPV) type 16 DNA-positive cancers of the **oral cavity** and **oropharynx** and in 35 HPV DNA-negative cancers matched by subsite, country, sex, age, and tobacco and **alcohol consumption**. [Ref.]
- In order to examine the role of the **TP53** gene in the pathogenesis of **schizophrenic disorders**, we investigated the genetic association between a functional polymorphism rs1042522 and schizophrenia by sequencing the fragment covering 72Pro> Arg in 701 cases and 695 controls in this work. [Ref.]
- The **Huntington's disease** protein interacts with p53 and CREB-binding protein and represses transcription. [Ref.]
- NF-kappaB/rel proteins, **tumor suppressor p53**, and **oncogene c-Myc** are critical transcription factors involved in coordinating cellular **decision-making** events in response to external stimuli. [Ref.]
- Tauroursodeoxycholic acid** modulates **p53**-mediated **apoptosis** in **Alzheimer's disease** mutant **neuroblastoma** cells. [Ref.]

**High impact information on TP53**

- They show that the **p38**-regulated/activated protein kinase (PRAK) induces **senescence** downstream of oncogenic Ras by directly phosphorylating and activating the tumor-suppressor protein **p53**. [Ref.]
- Telomere** dysfunction suppresses cancer through the **p53** tumor suppressor pathway but also contributes to aging. [Ref.]
- Therefore, NFDD4-1 is a potential **proto-oncogene** that negatively regulates **PTEN** via ubiquitination. a

advanced search

**Editor**  
Edit this page  
Discussion  
History  
Permanent link

**Special functions**  
Print  
Tutorial  
Contact

**Links**  
NCBI Gene  
Uniprot  
SNPedia

**Table of contents**  
- Disease relevance of **TP53**  
- Psychiatry related information on **TP53**  
- High impact information on **TP53**  
- Chemical compound and disease context of **TP53**  
- Biological context of **TP53**  
- Anatomical context of **TP53**  
- Associations of **TP53** with chemical compounds  
- Physical interactions of **TP53**  
- Enzymatic interactions of **TP53**  
- Colocalizations of **TP53**

Gene: **KRAS - v-Ki-ras2 Kirsten rat sarcoma viral... (Homo sapiens)**  
[Hold mouse button down for author info.]

society in science  
The Branco Weiss Fellowship

MIT  
Massachusetts Institute of Technology

CSAIL/DIG - Decentralized Information Group

## Towards Collaborative Publishing in Science

The screenshot shows a web browser window displaying a gene page for **SNF1** (AMP-activated serine/threonine protein...) in *Saccharomyces cerevisiae*. A "Relationship editor (RDF)" dialog box is open, showing a relationship between **gene: sN\_35758** and **gene: sN\_32484**. The relationship is labeled "suppress" with a sub-label "- switch direction". The dialog includes an "[add new]" button, "[save]", "[delete]", and "[cancel]" buttons. A dropdown menu is open over the "gene: sN\_35758" field, listing several gene identifiers: ATG13, ATG1, ATIC, CAT8, CTK1, CYC8, ELM1, GAL83, GCN5, and GLC7. At the bottom of the dialog are "[Save]" and "[Cancel]" buttons. The background page shows a snippet of text: "...inhibits the" and "relative". The browser's address bar shows "wiki for the life sciences where authorship matters. Nature Genetics (2008)". The user's name "Hello John Os" is visible in the top right corner. The browser's toolbar at the bottom includes icons for text formatting (bold, italic, list, link), mathematical symbols (x<sub>2</sub>, x<sup>2</sup>), and other navigation tools.

## **Acknowledgements**

Branco Weiss, Olaf Kuebler  
Tim Berners-Lee, Danny Weitzner

Chris Sander, Ben Gross  
Marta Ramirez, Thomas Pfeiffer

Alfonso Valencia  
Rolf Apweiler  
Peter Schuster

## Towards Collaborative Publishing in Science

---