



# H3ABioNet

Pan African Bioinformatics Network for H3Africa

## Introduction to Bioinformatics Online Course : IBT

### Introduction to Databases and Resources Advanced Literature Searching



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# Learning Objectives

- Advanced Literature Searching
  - Known which are the best tools to search for literature
  - Understand the logic of constructing advanced search queries
  - Know which basic search tags to use for generating specific search queries
  - Conduct more efficient searches using PubMed

# Learning Outcomes

- Advanced Literature Searching
  - Locate and understand the advanced search pages for PubMed
  - Generate complex search queries using provided examples
  - Use some of the major search fields (tags) to generate specific search results
  - Further refine your search results for specific articles

# Literature Searching



- Essential aspect of all research
- Most resources are now electronically available
- Not necessarily easier to retrieve
- Searching the literature can be a challenging task



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# Google

Google can bring you back  
100,000 answers  
A LIBRARIAN can bring you back the  
**RIGHT ONE**  
– Neil Gaiman



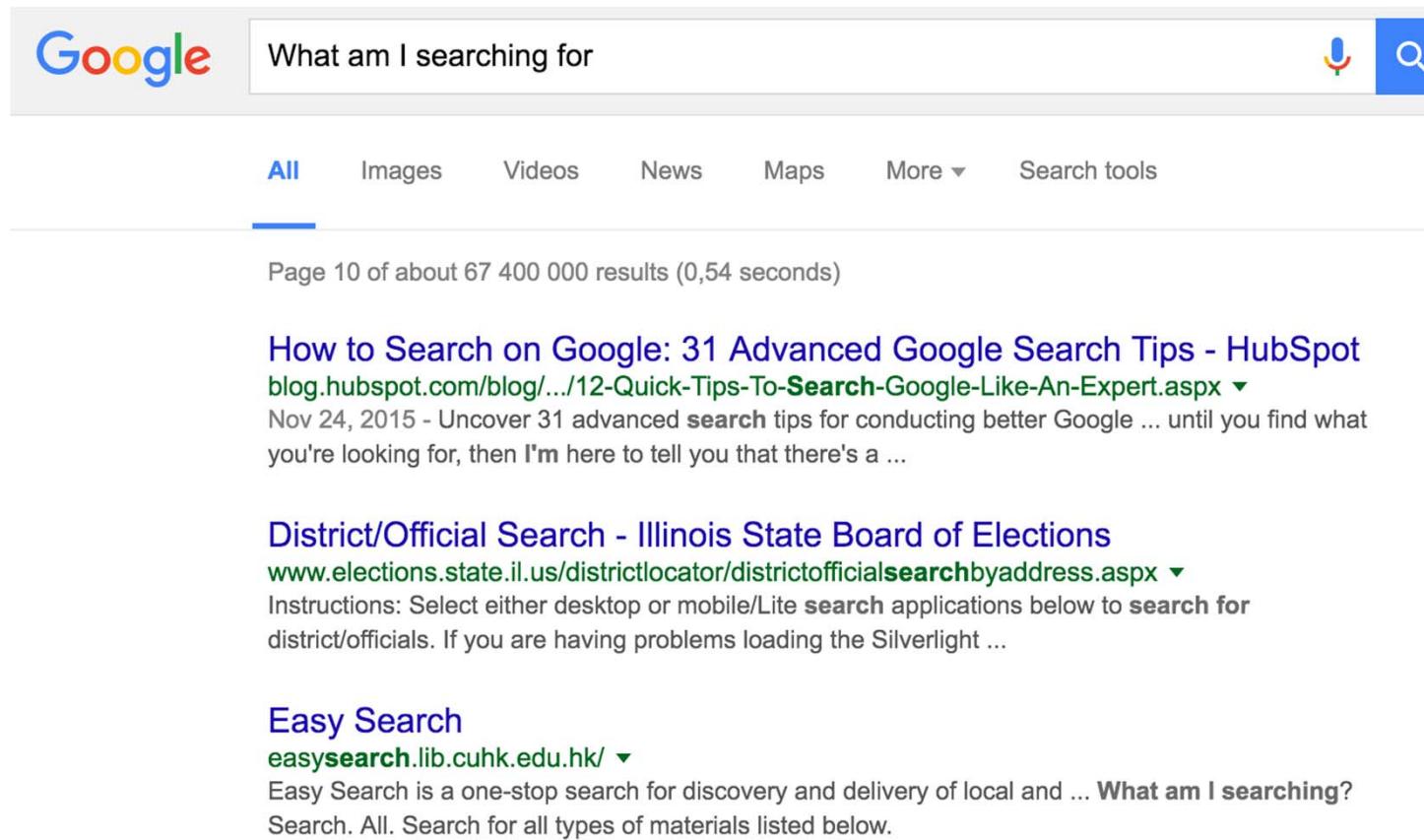
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# Useless Results



Google What am I searching for

All Images Videos News Maps More ▾ Search tools

Page 10 of about 67 400 000 results (0,54 seconds)

**How to Search on Google: 31 Advanced Google Search Tips - HubSpot**  
[blog.hubspot.com/blog/.../12-Quick-Tips-To-Search-Google-Like-An-Expert.aspx](http://blog.hubspot.com/blog/.../12-Quick-Tips-To-Search-Google-Like-An-Expert.aspx) ▾  
Nov 24, 2015 - Uncover 31 advanced **search** tips for conducting better Google ... until you find what you're looking for, then I'm here to tell you that there's a ...

**District/Official Search - Illinois State Board of Elections**  
[www.elections.state.il.us/districtlocator/districtofficialsearchbyaddress.aspx](http://www.elections.state.il.us/districtlocator/districtofficialsearchbyaddress.aspx) ▾  
Instructions: Select either desktop or mobile/Lite **search** applications below to **search for** district/officials. If you are having problems loading the Silverlight ...

**Easy Search**  
[easysearch.lib.cuhk.edu.hk/](http://easysearch.lib.cuhk.edu.hk/) ▾  
Easy Search is a one-stop search for discovery and delivery of local and ... **What am I searching?** Search. All. Search for all types of materials listed below.

## Answer 6 Questions to Reveal Your Life Purpose | SUCCESS

[www.success.com/article/answer-6-questions-to-reveal-your-life-purpose](http://www.success.com/article/answer-6-questions-to-reveal-your-life-purpose) ▾  
Dec 25, 2014 - Get ready for some soul-**searching**. ... I, the President of the U.S., (your name) **am** designed to be a good communicator (insert personal quality) ...



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# Useless Results

How do i delete the trending items on my google search bar history ...

[productforums.google.com/d/topic/websearch/FfSE0mP1qyo](http://productforums.google.com/d/topic/websearch/FfSE0mP1qyo) ▾

Jan 29, 2016 - 100+ posts - 100+ authors

Andy Wiles, 1/30/16 11:54 AM. I really hope somebody finds a way to fix this issue! I keep my **search** history cleared and nice and tidy, except ...

Patent Searching 101: A Patent Search Tutorial - IPWatchdog.com ...

[www.ipwatchdog.com/2015/07/11/patent-searching-101-a...search...2/id=59308/](http://www.ipwatchdog.com/2015/07/11/patent-searching-101-a...search...2/id=59308/) ▾

Jul 11, 2015 - It is quite common for inventors to do a patent **search** and find nothing even when ...

Gene, I am pleased to see that you keep emphasizing the ...

Searching for Eternity - Page 8 - Google Books Result

<https://books.google.co.za/books?isbn=076420372X>

Elizabeth Musser - 2007 - Fiction

Of course I am thinking about him, but at the same time, I am thinking of her, wondering if she is down in Bolivia, if she helped them locate the Butcher. "You may ...

---

Searches related to What am I searching for

searching for **something quotes**

what am i searching for **quiz**

what am i searching for **in life**

i **don't know what i'm looking for lyrics**

what am i searching for **on google**

i'm searching for **something lyrics**

searching for **something more in life**

i have everything but still **something is missing quotes**

---

< Goooooooooooooogle >

Previous

5 6 7 8 9 10 11 12 13 14

Next



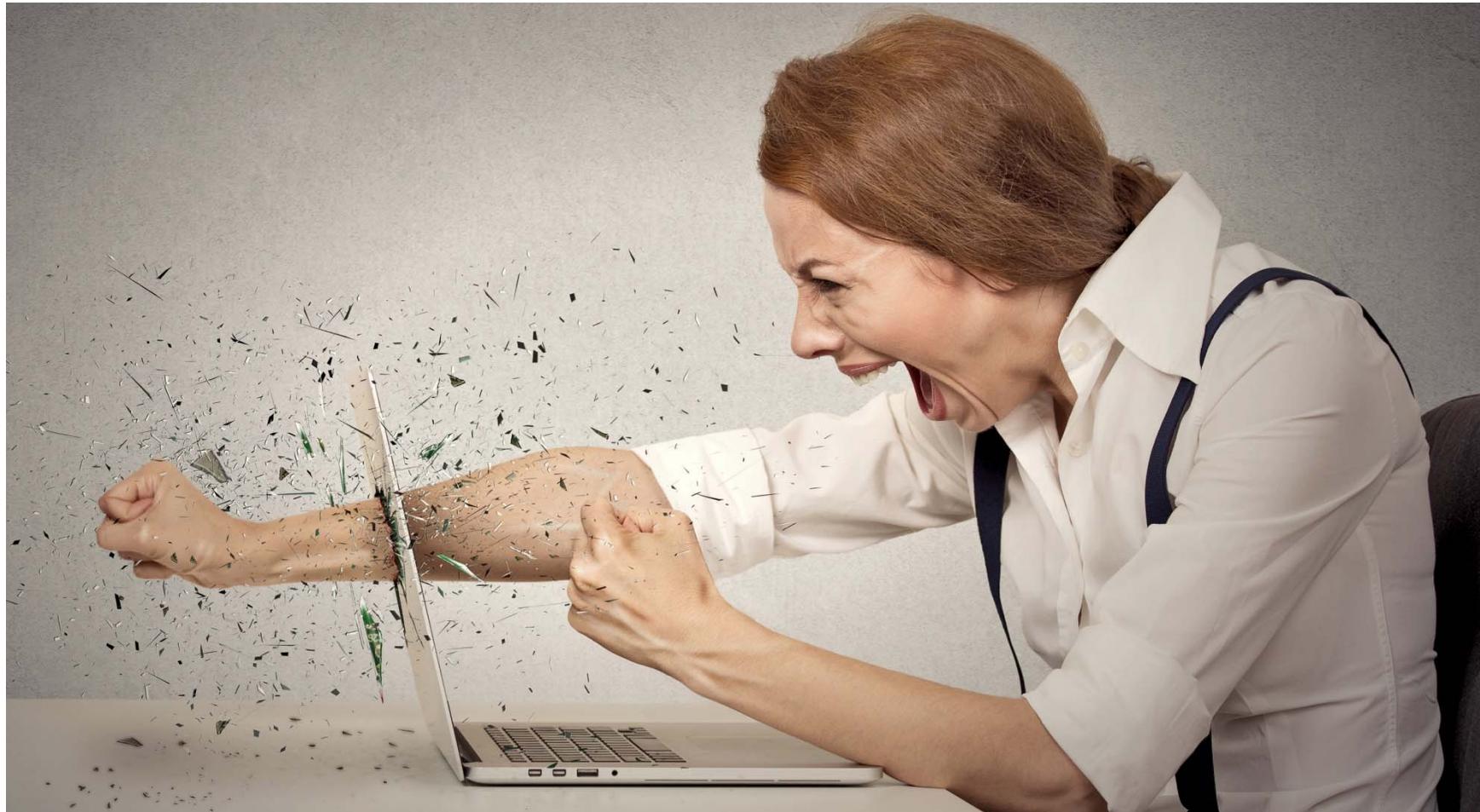
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# This lecture should save you from this!



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# Popular Literature Resources

- Google Scholar – Specialised google search engine for academic literature
- PubMed – bibliographical database run by the NCBI covering a range of biomedical literature including MEDLINE
- PubMed Clinical Queries – A specialised PubMed search for searching for clinical data

# Other literature resources

- Scopus – largest abstract and citation database of peer-reviewed literature across all scientific domains
- ScienceDirect – Linked to Scopus and Elsevier, search across a number of journals from all scientific domains



# MEDLINE



- The US National Library of Medicine (NLM) is the world's largest medical library
- NLM has been indexing biomedical literature since 1897 to help provide easy access to literature
- The printed index to articles, *Index Medicus*, was converted into the database known today as MEDLINE



# MEDLINE

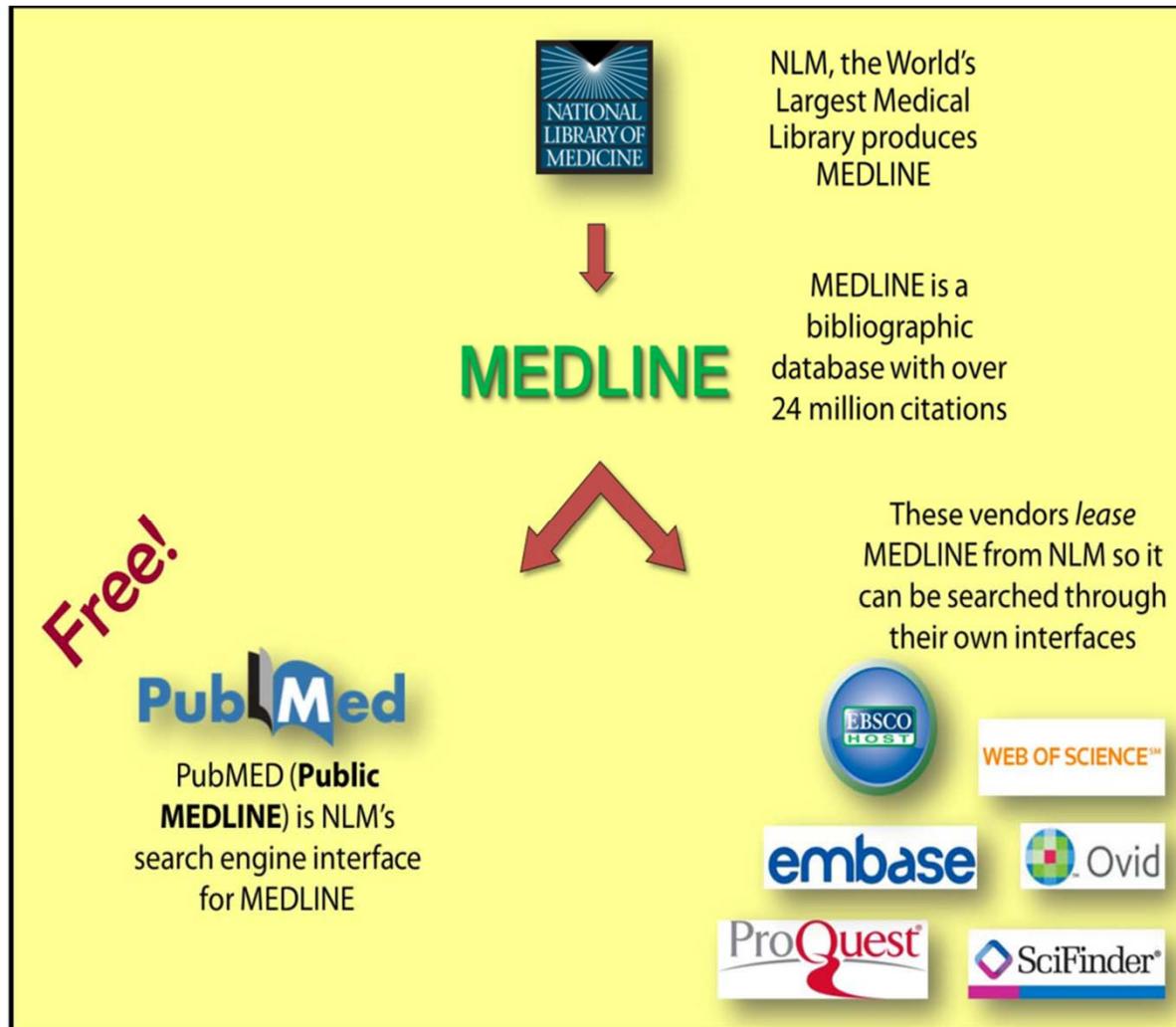


- Since 1996, free access to MEDLINE has been made available to the public via PubMed at NCBI
- In addition to MEDLINE PubMed contains citations from additional biomedical journals and books.
- PubMed provides access to more than 26 million citations for biomedical literature

# MEDLINE



U.S. National Library  
of Medicine



<http://research.library.gsu.edu/pubmed>



# Why use PubMed/MEDLINE

- Comprehensive, current up-to-date information
- Literature from credible sources
- Controlled vocabulary for indexing and searching
- Abstract and/or full text often provided
- Can filter search results to narrow query



<http://www.ncbi.nlm.nih.gov/pubmed/>

# ADVANCED SEARCHING USING PUBMED

# Literature Searching Steps

- First step is to clearly know what information you are looking for
- Break down your search question into keywords
- Identify the main sources for quality data
- Decide if it is best to combine your keywords or search for them separately

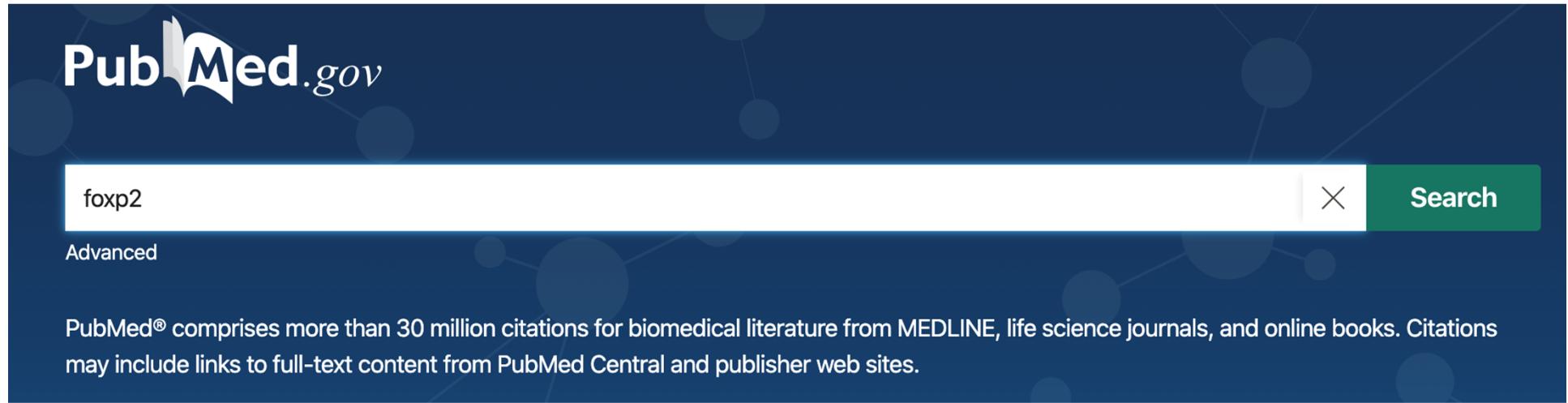
## Example: FOXP2

- You are interested in studying genetic variation in *FOXP2* and its impact of speech development
- You would like to review all studies that have linked mutations in *FOXP2* to some sort of speech development disorder
- You would also like to find the original case study that suggested that *FOXP2* is responsible for speech development in humans

# Breaking down the search query

- You are interested in studying **genetic variation** in ***FOXP2*** and its impact of **speech development**
- You would like to review all studies that have linked **mutations** in ***FOXP2*** to a **speech development disorder**
- You would also like to find the original study by Lai *et. al.* in 2001 that suggested that ***FOXP2*** is responsible for **speech development** in **humans**

# Simplest Search



The screenshot shows the PubMed.gov search interface. At the top left is the PubMed logo. Below it is a search bar containing the query "foxp2". To the right of the search bar is a green "Search" button. Below the search bar is a link labeled "Advanced". A descriptive text block below the search bar states: "PubMed® comprises more than 30 million citations for biomedical literature from MEDLINE, life science journals, and online books. Citations may include links to full-text content from PubMed Central and publisher web sites."



foxp2

Search

Advanced Create alert Create RSS

User Guide

Save

Email

Send to

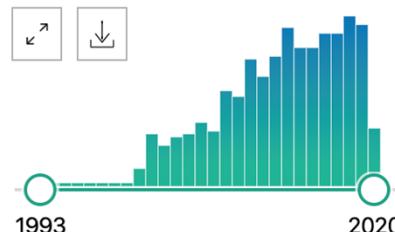
Sorted by: Best match

Display options

MYNCBI FILTERS

604 results

RESULTS BY YEAR



TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

ARTICLE ATTRIBUTE

- Associated data

ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial

[The FOXP2-Driven Network in Developmental Disorders and Neurodegeneration.](#)

1 Oswald F, Klöble P, Ruland A, Rosenkranz D, Hinz B, Butter F, Ramljak S, Zechner U, Herlyn H.

Front Cell Neurosci. 2017 Jul 26;11:212. doi: 10.3389/fncel.2017.00212. eCollection 2017.

PMID: 28798667 [Free PMC article.](#)

RNA-seq led to identification of 27 genes with differential regulation under the control of human **FOXP2**, which were previously reported to have **FOXP2**-driven and/or songbird song-related expression regulation. ...These genes may be directly regulated by **FOXP2** considering numerous matches of established **FOXP2**-binding motifs as well as publicly available **FOXP2**-ChIP-seq reads within their putative promoters. ...

[Cite](#) [Share](#)

[The untold stories of the speech gene, the \*\*FOXP2\*\* cancer gene.](#)

2 Herrero MJ, Gitton Y.

Genes Cancer. 2018 Jan;9(1-2):11-38. doi: 10.18632/genescancer.169.

PMID: 29725501 [Free PMC article.](#) Review.

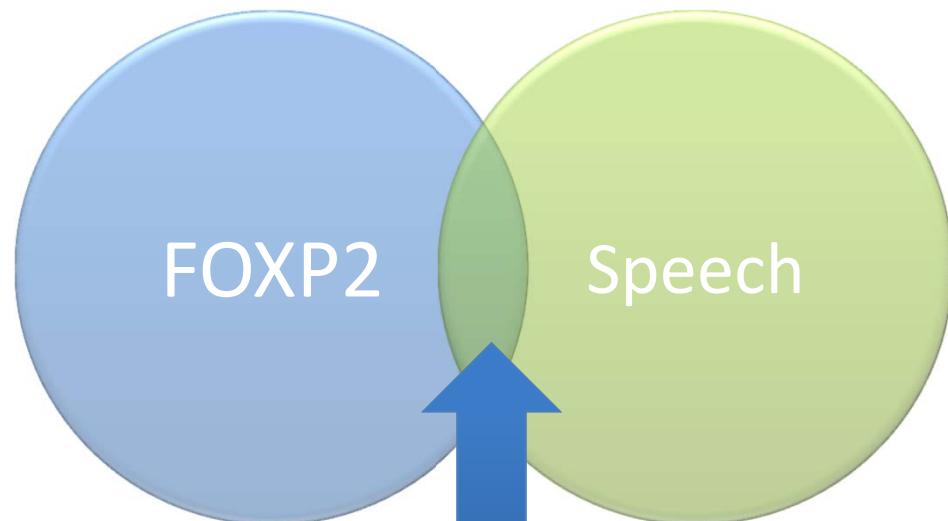
**FOXP2** encodes a transcription factor involved in speech and language acquisition. Growing evidence now suggests that dysregulated **FOXP2** activity may also be instrumental in human oncogenesis, along the lines of other cardinal developmental transcription factors such as DLX5 and DLX6 [1-4]. ...Whether **FOXP2** may further become a therapeutic target is an actively explored lead. Knowledge reviewed here may help improve our understanding of **FOXP2** roles during oncogenesis and provide cues for diagnostic, prognostic and therapeutic analyses....

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# Combining Search Terms – Boolean Operators

**FOXP2 – 604**

↓ OR ↓  
**124 578**



NOT  
319

**AND 285**



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# Other search operators

- Truncation
  - Child\* = Child and children
- Quotations
  - “copy number variation”



# Advanced



National Library of Medicine  
National Center for Biotechnology Information

## PubMed Advanced Search Builder

Add terms to the query box

All Fields



Enter a search term

### Query box

Enter / edit your search query here

### History and Search Details

Your history is currently empty! As you use Pub

- All Fields
- Author
- Author - Corporate
- Author - First
- Author - Identifier
- Author - Last
- Book
- Conflict of Interest Statements
- Date - Completion
- Date - Create
- Date - Entry
- Date - MeSH
- Date - Modification
- Date - Publication
- EC/RN Number
- Editor
- Filter
- Grant Number
- ISBN
- Investigator
- Issue
- Journal
- Language
- Location ID
- MeSH Major Topic
- MeSH Subheading
- MeSH Terms
- Other Term
- Pagination
- Pharmacological Action
- Publication Type
- Publisher
- Secondary Source ID
- Subject - Personal Name
- Supplementary Concept
- Text Word
- Title
- Title/Abstract
- Transliterated Title
- Volume

# ry Builder

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User Guide

ADD

Show Index

Search

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FIELD NAME	[TAG]	SEARCHES
Affiliation	[AD]	First listed author's institutional affiliation and address
Author	[AU]	Author name
Corporate Name as Author	[CN]	Corporate names as authors
Full Author Name	[FAU]	Full author name (2002 onward, if available)
Issue	[IP]	Journal issue number
Journal Title	[TA]	Title of journal, journal title abbreviation, ISSN
Language	[LA]	Language of the article (not the abstract)
Last Author Name	[LASTAU]	Last personal author in a citation
MeSH Major Topic	[MAJR]	Main topic of an article
MeSH Subheading	[SH]	MeSH Subheading
MeSH Terms	[MH]	MeSH Term
Pagination	[PG]	First page number of a journal article
Personal Name as Subject	[PS]	Person as the subject of an article, not as an author
Publication Date	[PD]	Date an article was published
Publication Type	[PT]	Format of an article (letter, clinical trial, etc) rather than content
Substance Name	[NM]	Chemical and substance names discussed in an article
Text Word	[TW]	Textual fields of PubMed records
Title	[TI]	Article title
Title or Abstract	[TIAB]	Words in an article title or an abstract
Volume	[VI]	Volume number of a particular journal
Year	[DP]	The year when a journal article was published. Search a date range, type in 2000:2009[DP]



# Advanced Search Query



National Library of Medicine  
National Center for Biotechnology Information

Log in

## PubMed Advanced Search Builder

Add terms to the query box

All Fields



Enter a search term

ADD



Show Index

## Query box

Enter / edit your search query here

Search



## History and Search Details

Your history is currently empty! As you use PubMed your recent searches will appear here.



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# Breaking down the search query

- You are interested in studying **genetic variation** in ***FOXP2*** and its impact of **speech development**
- You would like to review all studies that have linked **mutations** in ***FOXP2*** to a **speech development disorder**
- You would also like to find the original study by Lai *et. al.* in 2001 that suggested that ***FOXP2*** is responsible for **speech development in humans**



# FOXP2 Query Building



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National Center for Biotechnology Information

Log in

## PubMed Advanced Search Builder

PubMed.gov

User Guide

Add terms to the query box

All Fields



Enter a search term

ADD



Show Index

Query box

Enter / edit your search query here

Search



## History and Search Details

Download Delete

Search	Actions	Details	Query	Results	Time
#3	...	>	Search: <b>mutation</b>	1,093,123	04:58:47
#2	...	>	Search: <b>speech development</b>	30,109	04:58:34
#1	...	>	Search: <b>foxp2</b>	604	04:58:05

Showing 1 to 3 of 3 entries



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# FOXP2 Query Building



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National Center for Biotechnology Information

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PubMed Advanced Search Builder

PubMed.gov

User Guide

Add terms to the query box

All Fields ▾ Enter a search term

AND ▾ Show Index

Query box

((foxp2) AND (speech development)) AND (mutation)

Search

History and Search Details

Download Delete

Search	Actions	Details	Query	Results	Time
#5	...	>	Search: ((foxp2) AND (speech development)) AND (mutation)	92	05:03:54
#3	...	>	Search: mutation	1,093,123	04:58:47
#2	...	>	Search: speech development	30,109	04:58:34
#1	...	>	Search: foxp2	604	04:58:05

Showing 1 to 4 of 4 entries



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Email

Send to

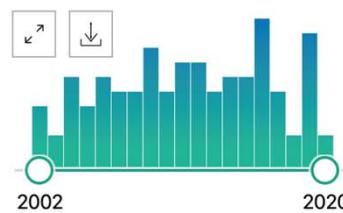
Sorted by: Best match

Display options

## MYNCBI FILTERS

92 results

## RESULTS BY YEAR



## TEXT AVAILABILITY

- Abstract
- Free full text
- Full text

## ARTICLE ATTRIBUTE

- Associated data

## ARTICLE TYPE

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Reviews

- [The Key Regulator for Language and Speech Development, FOXP2, is a Novel Substrate for SUMOylation.](#)

1 Meredith LJ, Wang CM, Nascimento L, Liu R, Wang L, Yang WH.

J Cell Biochem. 2016 Feb;117(2):426-38. doi: 10.1002/jcb.25288.

PMID: 26212494 [Free PMC article.](#)

Transcription factor forkhead box protein P2 (**FOXP2**) plays an essential role in the **development** of language and **speech**. However, the transcriptional activity of **FOXP2** regulated by the post-translational modifications remains unknown. ...Interestingly, we observed that human etiological **FOXP2** R553H **mutation** robustly reduces its SUMOylation potential as compared to wild-type **FOXP2**. ...

[“ Cite](#) [Share](#)

- [FOXP2 variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum.](#)

2 Reuter MS, Riess A, Moog U, Briggs TA, Chandler KE, Rauch A, Stampfer M, Steindl K, Gläser D, Joset P; DDD Study, Krumbiegel M, Rabe H, Schulte-Mattler U, Bauer P, Beck-Wödl S, Kohlhase J, Reis A, Zweier C.

J Med Genet. 2017 Jan;54(1):64-72. doi: 10.1136/jmedgenet-2016-104094. Epub 2016 Aug 29.

PMID: 27572252

RESULTS: We identified four different truncating **mutations**, two novel missense **mutations** within the forkhead domain and an intragenic deletion in **FOXP2** in 14 individuals from eight unrelated families. ...Motor **development** was normal or only mildly delayed. Mild cognitive impairment was reported for most individuals. CONCLUSIONS: By identifying intragenic deletions or **mutations** in 14 individuals from eight unrelated families with variable developmental delay/cognitive impairment and **speech** and language deficits, we considerably broaden the **mutational** and clinical spectrum associated with aberrations in **FOXP2**....

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# Narrower search

MeSH Terms  
 Other Term  
 Pagination  
 Pharmacological Action  
 Publication Type  
 Publisher  
 Secondary Source ID  
 Subject - Personal Name  
 Supplementary Concept  
 Text Word  
 Title

Search term

**AND**

User Guide

Query box

**History and Search Details**

Search	Actions	Details	Query	Results	Time
#4	...	>	Search: ((foxp2[Title/Abstract]) AND (speech development[Title/Abstract])) AND (mutation[Title/Abstract])	3	05:10:29
#3	...	>	Search: mutation[Title/Abstract]	358,784	05:10:01
#2	...	>	Search: speech development[Title/Abstract]	1,079	05:09:44
#1	...	>	Search: foxp2[Title/Abstract]	555	05:09:27

Showing 1 to 4 of 4 entries



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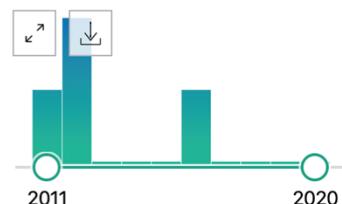


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RESULTS BY YEAR



TEXT AVAILABILITY

Abstract

Free full text

Full text

ARTICLE ATTRIBUTE

Associated data

ARTICLE TYPE

Books and Documents

Clinical Trial

Meta-Analysis

Randomized Controlled Trial

Review

Systematic Reviews

PUBLICATION DATE

1 year

5 years

10 years

[Additional filters](#)

3 results

- 1 [The Key Regulator for Language and Speech Development, FOXP2, is a Novel Substrate for SUMOylation.](#)

Meredith LJ, Wang CM, Nascimento L, Liu R, Wang L, Yang WH.

J Cell Biochem. 2016 Feb;117(2):426-38. doi: 10.1002/jcb.25288.

PMID: 26212494 [Free PMC article.](#)

Here, we demonstrated that **FOXP2** is clearly defined as a SUMO target protein at the cellular levels as **FOXP2** is covalently modified by both SUMO1 and SUMO3. ...Interestingly, we observed that human etiological **FOXP2** R553H **mutation** robustly reduces its SUMOylation potential as compared to wild-type **FOXP2**...

[Cite](#) [Share](#)

- 2 [An aetiological Foxp2 mutation causes aberrant striatal activity and alters plasticity during skill learning.](#)

French CA, Jin X, Campbell TG, Gerfen E, Groszer M, Fisher SE, Costa RM.

Mol Psychiatry. 2012 Nov;17(11):1077-85. doi: 10.1038/mp.2011.105. Epub 2011 Aug 30.

PMID: 21876543 [Free PMC article.](#)

Mutations in the human **FOXP2** gene cause impaired **speech development** and linguistic deficits, which have been best characterised in a large pedigree called the KE family. ...Independent studies in multiple species suggest that the striatum is a key site of **FOXP2** action. Here, we used *in vivo* recordings in awake-behaving mice to investigate the effects of the KE-family **mutation** on the function of striatal circuits during motor-skill learning. ...

[Cite](#) [Share](#)

- 3 [Foxp2 mutations impair auditory-motor association learning.](#)

Kurt S, Fisher SE, Ehret G.

PLoS One. 2012;7(3):e33130. doi: 10.1371/journal.pone.0033130. Epub 2012 Mar 7.

PMID: 22412993 [Free PMC article.](#)

The magnitude of impairments in association learning, however, depends on the nature of the **mutation**. Mice with a missense **mutation** in the DNA-binding domain are able to learn, but at a much slower rate than wild type animals, while mice carrying an early nonsense **mutation** learn very little. ...Given the importance of such networks for the acquisition of human spoken language, and the fact that similar mutations in human **FOXP2** cause problems with **speech development**, this work opens up a new perspective on the use of mouse models for understanding pathways underlying speech and language disorders....



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# MeSH Terms

- Medical Subject Headings
  - Set of controlled vocabulary terms used by the NLM
  - Articles are assigned MeSH terms by subject specialists who read over the articles and determine the subject contents
  - Ensures that articles on the same subject are grouped together regardless of the terms used by the authors

# MeSH Terms

- May be useful to look up the MeSH term for your keywords in order to run a more specific search

# MeSH Terms

MeSH      MeSH      speech development      [Create alert](#)   [Limits](#)   [Advanced](#)      [X](#)   [Search](#)

Summary      Send to:

## Search results

Items: 2

[Language Development Disorders](#)

1. Conditions characterized by language abilities (comprehension and expression of **speech** and writing) that are below the expected level for a given age, generally in the absence of an intellectual impairment. These conditions may be associated with DEAFNESS; BRAIN DISEASES; MENTAL DISORDERS; or environmental factors.

Year introduced: 1986(1980)

[Speech Development, Delayed, With Facial Asymmetry, Strabismus, And Transverse Earlobe Crease \[Supplementary Concept\]](#)

2. Date introduced: November 5, 2012



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# MeSH Terms

MeSH

MeSH speech development

Create alert  Limits  Advanced  Search

[All MeSH Categories](#)

[Diseases Category](#)

[Nervous System Diseases](#)

[Neurologic Manifestations](#)

[Neurobehavioral Manifestations](#)

[Communication Disorders](#)

[Language Disorders](#)

**Language Development Disorders**

[Specific Language Disorder](#)



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# FOXP2 + MeSH

**PubMed Advanced Search Builder**

Add terms to the query box

All Fields ▼ Enter a search term ▼

**ADD** ▼ Show Index

**Query box**

Enter / edit your search query here ▼

**Search** ▼

**History and Search Details**

▼ Download ▼ Delete

Search	Actions	Details	Query	Results	Time
#6	...	>	Search: ((foxp2[Title/Abstract]) AND (language development disorders[Title/Abstract])) AND (mutation[Title/Abstract])	13	05:32:40
#4	...	>	Search: ((foxp2[Title/Abstract]) AND (speech development[Title/Abstract])) AND (mutation[Title/Abstract])	3	05:23:41
#3	...	>	Search: mutation[Title/Abstract]	358,784	05:10:01
#2	...	>	Search: speech development[Title/Abstract]	1,079	05:09:44
#1	...	>	Search: foxp2[Title/Abstract]	555	05:09:27

Showing 1 to 5 of 5 entries



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13 results

Your search was processed without automatic term mapping because it retrieved zero results.

- Equivalent missense variant in the **FOXP2** and **FOXP1** transcription factors causes distinct neurodevelopmental disorders.

1 Sollis E, Deriziotis P, Saitsu H, Miyake N, Matsumoto N, Hoffer M JV, Ruivenkamp CAL, Alders M, Okamoto N, Bijlsma EK, Plomp AS, Fisher SE.

Hum Mutat. 2017 Nov;38(11):1542-1554. doi: 10.1002/humu.23303. Epub 2017 Aug 14.

PMID: 28741757

The closely related paralogues **FOXP2** and **FOXP1** encode transcription factors with shared functions in the **development** of many tissues, including the brain. ...We present here for the first time a direct comparison of the molecular and clinical consequences of the same **mutation** affecting the equivalent residue in **FOXP1** and **FOXP2**. ...

“ Cite Share

- Early neuroimaging markers of **FOXP2** intragenic deletion.

2 Liégeois FJ, Hildebrand MS, Bonthrone A, Turner SJ, Scheffer IE, Bahlo M, Connelly A, Morgan AT.

Sci Rep. 2016 Oct 13;6:35192. doi: 10.1038/srep35192.

PMID: 27734906 Free PMC article.

**FOXP2** is the major gene associated with severe, persistent, developmental speech and **language disorders**. While studies in the original family in which a **FOXP2 mutation** was found showed volume reduction and reduced activation in core **language** and speech networks, there have been no imaging studies of different **FOXP2** mutations. ...The hippocampus is implicated for the first time in **FOXP2** diseases. We conclude that **FOXP2** anomaly is either directly or indirectly associated with atypical **development** of widespread subcortical networks early in life....

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- FOXP genes, neural development, speech and language disorders.**

3 Takahashi H, Takahashi K, Liu FC.

Adv Exp Med Biol. 2009;665:117-29. doi: 10.1007/978-1-4419-1599-3\_9.

PMID: 20429420 Review.

Thus these genes appear to be involved in the **development** control of the central nervous system.

Recently, **FOXP2**, a member of the Foxp subfamily, was identified as the first gene to be linked to an inherited form of **language** and speech disorder. The discovery of a **mutation** in **FOXP2** in a family with a speech and **language** disorder opened a new window to understanding the genetic cascades and neural circuits that underlie speech and **language** via molecular approaches



# Breaking down the search query

- You are interested in studying **genetic variation** in ***FOXP2*** and its impact of **speech development**
- You would like to review all studies that have linked **mutations** in ***FOXP2*** to a **speech development disorder**
- You would also like to find the original study by Lai *et. al.* in 2001 that suggested that ***FOXP2*** is responsible for **speech development** in **humans**

# FOXP2 Query Building

PubMed Advanced Search Builder

Pub**Med**.gov

User Guide

Add terms to the query box

All Fields **AND** Show Index

Enter a search term

Query box

((lai[Author]) AND (foxp2[Title/Abstract])) AND (("2001"[Date - Publication] : "2001"[Date - Publication]))

Search

History and Search Details

Download Delete

Search	Actions	Details	Query	Results	Time
#1	...	>	Search: ((lai[Author]) AND (foxp2[Title/Abstract])) AND (("2001"[Date - Publication] : "2001"[Date - Publication]))	1	05:38:19

Showing 1 to 1 of 1 entries

# FOXP2 Query Building

> [Nature](#). 2001 Oct 4;413(6855):519-23. doi: 10.1038/35097076.

## A Forkhead-Domain Gene Is Mutated in a Severe Speech and Language Disorder

C S Lai <sup>1</sup>, S E Fisher, J A Hurst, F Vargha-Khadem, A P Monaco

Affiliations + expand

PMID: 11586359 DOI: [10.1038/35097076](https://doi.org/10.1038/35097076)

### Abstract

Individuals affected with developmental disorders of speech and language have substantial difficulty acquiring expressive and/or receptive language in the absence of any profound sensory or neurological impairment and despite adequate intelligence and opportunity. Although studies of twins consistently indicate that a significant genetic component is involved, most families segregating speech and language deficits show complex patterns of inheritance, and a gene that predisposes individuals to such disorders has not been identified. We have studied a unique three-generation pedigree, KE, in which a severe speech and language disorder is transmitted as an autosomal-dominant monogenic trait. Our previous work mapped the locus responsible, SPCH1, to a 5.6-cM interval of region 7q31 on chromosome 7 (ref. 5). We also identified an unrelated individual, CS, in whom speech and language impairment is associated with a chromosomal translocation involving the SPCH1 interval. Here we show that the gene FOXP2, which encodes a putative transcription factor containing a polyglutamine tract and a forkhead DNA-binding domain, is directly disrupted by the translocation breakpoint in CS. In addition, we identify a point mutation in affected members of the KE family that alters an invariant amino-acid residue in the forkhead domain. Our findings suggest that FOXP2 is involved in the developmental process that culminates in speech and language.



**H3ABioNet**

Pan African Bioinformatics Network for H3Africa

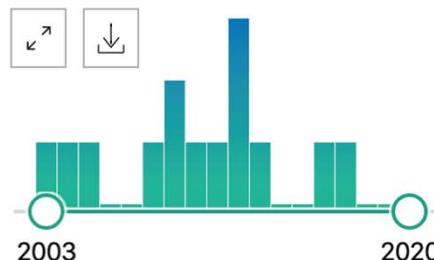


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13 results

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**RESULTS BY YEAR****TEXT AVAILABILITY**

- Abstract
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- Full text

**ARTICLE ATTRIBUTE**

- Associated data

**ARTICLE TYPE**

- Books and Documents
- Clinical Trial
- Meta-Analysis
- Randomized Controlled Trial
- Review
- Systematic Reviews

Your search was processed without automatic term mapping because it retrieved zero results.

- 1 [Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders.](#)

Sollis E, Deriziotis P, Saitsu H, Miyake N, Matsumoto N, Hoffer MJV, Ruivenkamp CAL, Alders M, Okamoto N, Bijlsma EK, Plomp AS, Fisher SE.  
Hum Mutat. 2017 Nov;38(11):1542-1554. doi: 10.1002/humu.23303. Epub 2017 Aug 14.  
PMID: 28741757

The closely related paralogues **FOXP2** and **FOXP1** encode transcription factors with shared functions in the **development** of many tissues, including the brain. ...We present here for the first time a direct comparison of the molecular and clinical consequences of the same **mutation** affecting the equivalent residue in **FOXP1** and **FOXP2**. ...

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- 2 [Early neuroimaging markers of FOXP2 intragenic deletion.](#)

Liégeois FJ, Hildebrand MS, Bonthrone A, Turner SJ, Scheffer IE, Bahlo M, Connelly A, Morgan AT.

Sci Rep. 2016 Oct 13;6:35192. doi: 10.1038/srep35192.

PMID: 27734906 [Free PMC article.](#)

**FOXP2** is the major gene associated with severe, persistent, developmental speech and **language disorders**. While studies in the original family in which a **FOXP2 mutation** was found showed volume reduction and reduced activation in core **language** and speech networks, there have been no imaging studies of different **FOXP2** mutations. ...The hippocampus is implicated for the first time in **FOXP2** diseases. We conclude that **FOXP2** anomaly is either directly or indirectly associated with atypical **development** of widespread subcortical networks early in life....

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- Biography
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- Classical Article
- Clinical Conference
- Clinical Study
- Clinical Trial Protocol
- Clinical Trial, Phase I
- Clinical Trial, Phase II
- Clinical Trial, Phase III
- Clinical Trial, Phase IV
- Clinical Trial, Veterinary
- Comment
- Introductory Journal Article
- Journal Article
- Lecture
- Legal Case
- Legislation
- Letter
- Multicenter Study
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- Observational Study, Veterinary
- Overall
- Patient Education Handout
- Periodical Index
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Review

> *Adv Exp Med Biol.* 2009;665:117-29. doi: 10.1007/978-1-4419-1599-3\_9.

## FOXP Genes, Neural Development, Speech and Language Disorders

Hiroshi Takahashi<sup>1</sup>, Kaoru Takahashi, Fu-Chin Liu

Affiliations + expand

PMID: 20429420 DOI: [10.1007/978-1-4419-1599-3\\_9](https://doi.org/10.1007/978-1-4419-1599-3_9)

FULL TEXT LINKS



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“ Cite

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13 results

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1

Case Reports > Hum Mutat. 2017 Nov;38(11):1542-1554.  
Epub 2017 Aug 14.

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# Sidebar...

- The advanced search pages that I have mentioned as part of PubMed are found on almost all the databases at the NCBI
- These databases can be searched by building up queries in a similar way as in PubMed

# Summary

- Literature searching is less frustrating when:
  - You select a single, high quality resource such as PubMed to search
  - You break up your query into useful keywords
  - You make use of advanced search pages to build up a query
  - You look up MeSH terms that might make your search more specific