



# H3ABioNet

Pan African Bioinformatics Network for H3Africa

## Introduction to Bioinformatics Online Course : IBT

# Introduction to Databases and Resources Advanced Literature Searching



**H3ABioNet**

Pan African Bioinformatics Network for H3Africa



Introduction to Bioinformatics Online Course:IBT  
Introduction to Databases and Resources | Shaun Aron

# 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



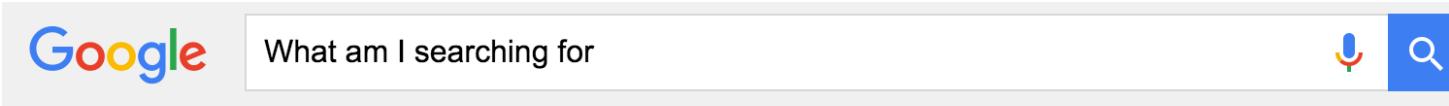
- Essential aspect of all research
- Most resources are now electronically available
- Not necessarily easier to retrieve
- Searching the literature can be a challenging task

# Google

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

– Neil Gaiman

# Useless Results



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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

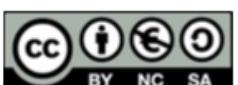
[easysearch.lib.cuhk.edu.hk/](http://easysearch.lib.cuhk.edu.hk/) ▾

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 ▾](#)

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/ ▾](#)

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**

< Gooooooooooooogle >

Previous

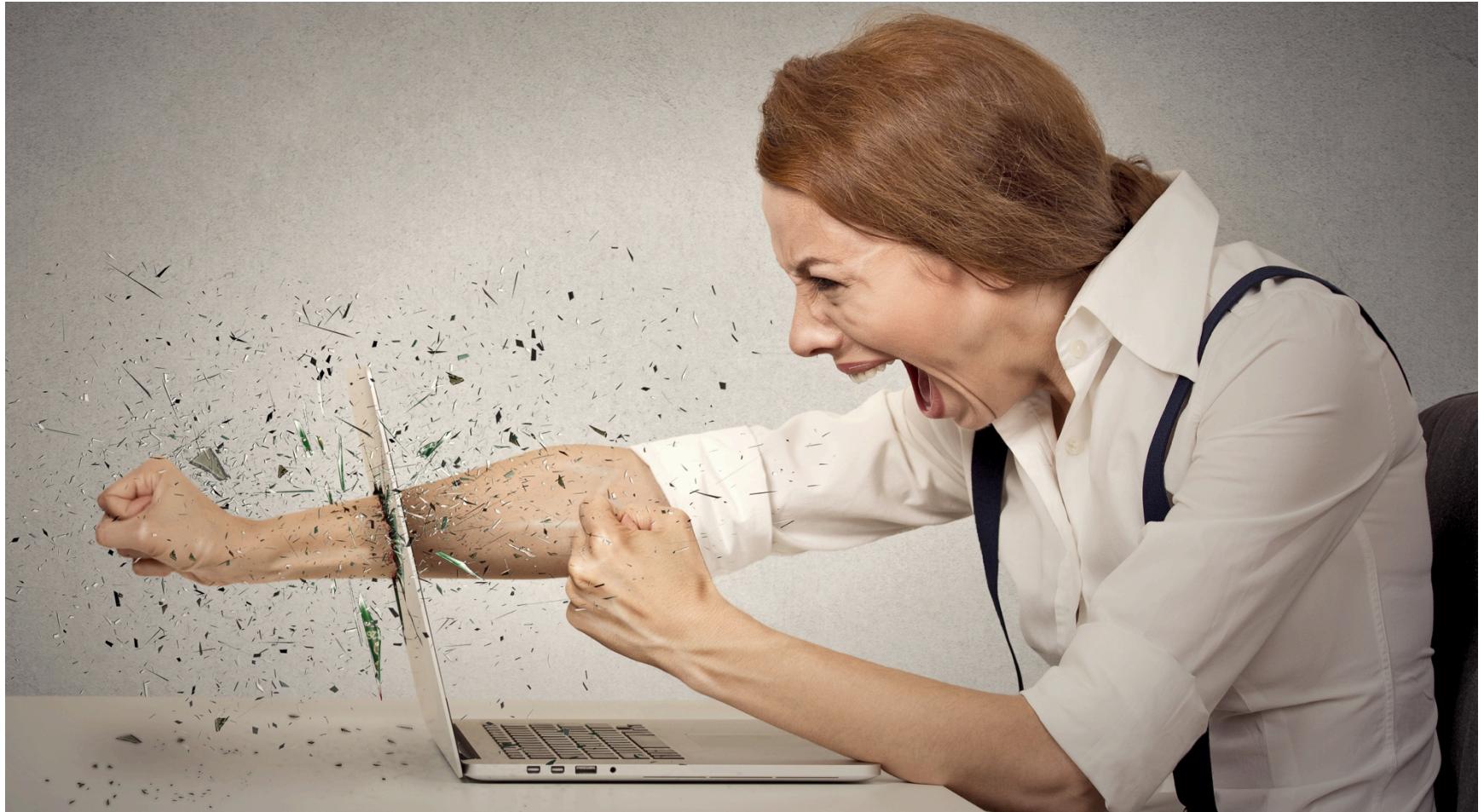
5 6 7 8 9 10 11 12 13 14

Next



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# This lecture should save you from this!



# 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



NLM, the World's  
Largest Medical  
Library produces  
MEDLINE



## MEDLINE

MEDLINE is a  
bibliographic  
database with over  
24 million citations



Free!



PubMED (**Public**  
**MEDLINE**) is NLM's  
search engine interface  
for MEDLINE

These vendors *lease*  
MEDLINE from NLM so it  
can be searched through  
their own interfaces



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



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# 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



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# 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

NCBI Resources How To

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PubMed

US National Library of Medicine  
National Institutes of Health



PubMed

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literature from  
Citations may  
and publisher

## Using PubMed

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Foxp2

[Search](#)

Help

- foxp2
- foxp2 language
- foxp2 gene
- foxp2 cancer
- foxp2 review
- foxp1 foxp2
- foxp2 mouse
- foxp2 brain
- foxp2 expression
- enard foxp2
- foxp2 and the neuroanatomy of speech and language
- foxp2 autism
- foxp2 evolution
- foxp2 human
- foxp2 and cancer
- foxp2 deletion
- foxp2 neanderthal
- foxp2 speech
- foxp2 knockout
- foxp2 schizophrenia

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# Results

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Help

Article types Clinical Trial Review Customize ...

Text availability Abstract Free full text Full text

PubMed Commons Reader comments Trending articles

Publication dates 5 years 10 years Custom range...

Species Humans Other Animals

[Clear all](#)

www.nlm.nih.gov

Format: Summary Sort by: Most Recent Send to Filters: Manage Filters

See 122 articles about **FOXP2** gene function

See also: [FOXP2 forkhead box P2](#) in the Gene database

**foxp2** in [Homo sapiens](#) [Mus musculus](#) [Rattus norvegicus](#) All 183 Gene records

See also: [22 tests](#) for **FOXP2** in the Genetic Testing Registry

Related searches

- [foxp2 gene](#)
- [foxp2 review](#)
- [foxp2 cancer](#)
- [foxp1 foxp2](#)
- [foxp2 mouse](#)

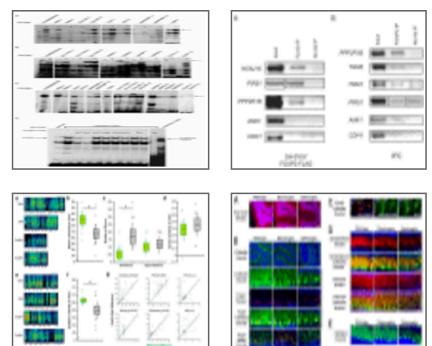
Search results

Items: 1 to 20 of 427 << First < Prev Page 1 of 22 Next > Last >>

[Foxp2 Regulates Identities and Projection Patterns of Thalamic Nuclei During Development.](#)  
 Ebisu H, Iwai-Takekoshi L, Fujita-Jimbo E, Momoi T, Kawasaki H.  
*Cereb Cortex.* 2016 Jul 6. [Epub ahead of print]  
 PMID: 27384060

[MicroRNA-190 regulates FOXP2 genes in human gastric cancer.](#)  
 2. Jia WZ, Yu T, An Q, Yang H, Zhang Z, Liu X, Xiao G.  
*Onco Targets Ther.* 2016 Jun 20;9:3643-51. doi: 10.2147/OTT.S103682. eCollection 2016.  
 PMID: 27382302 [Free PMC Article](#)

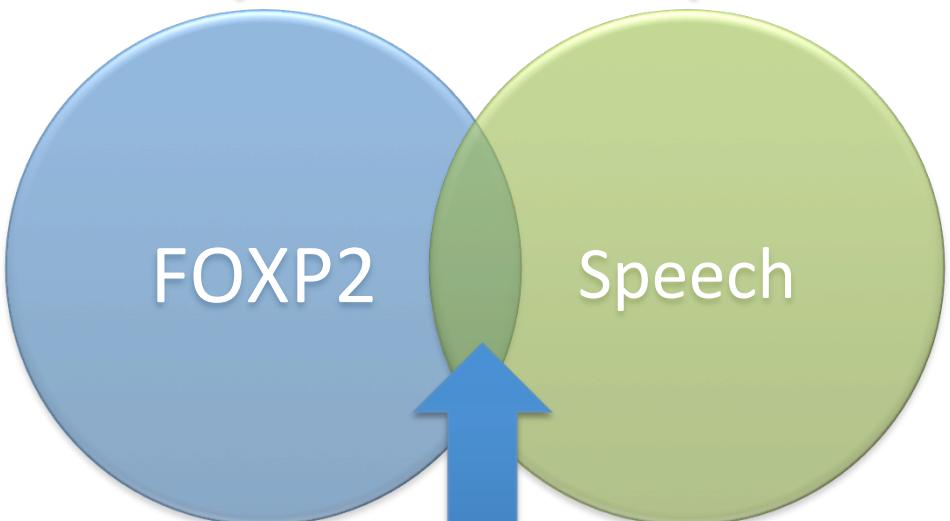
PMC Images search for *Foxp2*



# Combining Search Terms – Boolean Operators

FOXP2 – 427

OR  
100 756



NOT  
202

AND 225



# Other search operators

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

# PubMed Advanced Search Builder

NCBI Resources How To

PubMed Home More Resources Help

## PubMed Advanced Search Builder

Use the builder below to create your search.

[Edit](#)

### Builder

All Fields

AND All Fields

[Search](#) or [Add to history](#)

You are here: NCBI > Literature

### GETTING STARTED

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[NCBI Help Manual](#)

[NCBI Handbook](#)

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### Builder

AND

[Search](#)

### History

Search

#

- Affiliation
- All Fields
- Author
- Author - Corporate
- Author - First
- Author - Full
- Author - Identifier
- Author - Last
- Book
- Date - Completion
- Date - Create
- Date - Entrez
- Date - MeSH
- Date - Modification**
- Date - Publication
- EC/RN Number
- Editor
- Filter
- Grant Number
- ISBN
- Investigator
- Investigator - Full
- 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

Search

Sign in to NCBI

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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

## PubMed Advanced Search Builder

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### Builder

All Fields



[Show index list](#)

AND  All Fields



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**Search** or [Add to history](#)



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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

PubMed Advanced Search Builder

 YouTube Tutorial

FOXP2

[Edit](#)

[Clear](#)

## Builder

All Fields

FOXP2



[Show index list](#)

AND

All Fields



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[Search](#)

or [Add to history](#)

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## History

| Search | Add to builder      | Query                     | Items found            | Time     |
|--------|---------------------|---------------------------|------------------------|----------|
| #14    | <a href="#">Add</a> | Search mutation           | <a href="#">762205</a> | 09:32:25 |
| #13    | <a href="#">Add</a> | Search speech development | <a href="#">15901</a>  | 09:32:15 |
| #12    | <a href="#">Add</a> | Search FOXP2              | <a href="#">427</a>    | 09:31:52 |

# FOXP2 Query Building

PubMed Advanced Search Builder

 Tutorial

((FOXP2) AND speech development) AND mutation

[Edit](#)

[Clear](#)

## Builder

|            |                    |                                 |
|------------|--------------------|---------------------------------|
| All Fields | FOXP2              | <a href="#">Show index list</a> |
| AND        | speech development | <a href="#">Show index list</a> |
| AND        | mutation           | <a href="#">Show index list</a> |
| AND        |                    | <a href="#">Show index list</a> |

## History

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| Search | Add to builder      | Query  | Items found | Time     |
|--------|---------------------|--|-------------|----------|
| #15    | <a href="#">Add</a> | Search ((FOXP2) AND speech development) AND mutation | 49          | 09:34:49 |
| #14    | <a href="#">Add</a> | Search mutation                                      | 762205      | 09:32:25 |
| #13    | <a href="#">Add</a> | Search speech development                            | 15901       | 09:32:15 |
| #12    | <a href="#">Add</a> | Search FOXP2   | 427         | 09:31:52 |

Format: Summary ▾ Sort by: Most Recent ▾

Send to ▾ Filters: [Manage Filters](#)

## Search results

Items: 1 to 20 of 49

<< First < Prev Page 1 of 3 Next > Last >>

### Find related data

Database: Select

[Find items](#)

- [The language of speech development in FOXP2 mutants is similar to that of human monogenic language disorders](#)
1. **Query Translation:**
- ```
(FOXP2[All Fields] AND (( "speech"[MeSH Terms] OR "speech"[All Fields]) AND ("growth and development"[Subheading] OR ("growth"[All Fields] AND "development"[All Fields]) OR "growth and development"[All Fields] OR "development"[All Fields]))) AND ("mutation"[MeSH Terms] OR "mutation"[All Fields])
```
- [The language of speech development in FOXP2 mutants is similar to that of human monogenic language disorders](#)
2. **Mere J Cell PMID: Similar articles**
- [Monogenic language disorders](#)
3. **Adegbola R, Prockop A, et al. (2014) Language delay in a child with a de novo mutation in the FOXP2 gene. *J Speech Lang Hear Res* 57(1):1–10.**
- PMID: 25422445 [Free PMC Article](#)  
[Similar articles](#)
- [Drosophila FoxP mutants are deficient in operant self-learning.](#)
4. **Mendoza E, Colomb J, Rybak J, Pflüger HJ, Zars T, Scharff C, Brembs B. (2014) Drosophila FoxP mutants are deficient in operant self-learning. *PLoS One*. 2014 Jun 25;9(6):e100648. doi: 10.1371/journal.pone.0100648. eCollection 2014.**
- PMID: 24964149 [Free PMC Article](#)  
[Similar articles](#)
- eech"

See more...

Off Clear

ent) AND PubMed

guage

PubMed

aisoraer[Title/Abstract] AND Language Development Disorders MeSH

speech development disorder (1) MeSH

((FOXP2[Title/Abstract]) AND Language developmental disorder[Title/Abstract] (2) PubMed

See more...



# Narrower search

PubMed Advanced Search Builder

 Tutorial

Use the builder below to create your search

[Edit](#)

[Clear](#)

## Builder

|                                                          |   |                      |                                                                                                                                                                                                           |
|----------------------------------------------------------|---|----------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| All Fields                                               | ▼ | <input type="text"/> |  <a href="#">Show index list</a>                                                                                       |
| AND                                                      | ▼ | All Fields           | ▼   <a href="#">Show index list</a> |
| <a href="#">Search</a> or <a href="#">Add to history</a> |   |                      |                                                                                                                                                                                                           |

## History

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| Search | Add to builder      | Query                                       | Items found | Time     |
|--------|---------------------|---------------------------------------------|-------------|----------|
| #14    | <a href="#">Add</a> | Search <b>mutation</b> [Title/Abstract]     | 288772      | 09:42:12 |
| #13    | <a href="#">Add</a> | Search "speech development"[Title/Abstract] | 897         | 09:41:57 |
| #12    | <a href="#">Add</a> | Search <b>FOXP2</b> [Title/Abstract]        | 387         | 09:41:34 |



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# Narrower search

## History

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| Search | Add to builder      | Query                                                                                                  | Items found            | Time     |
|--------|---------------------|--------------------------------------------------------------------------------------------------------|------------------------|----------|
| #15    | <a href="#">Add</a> | Search ((FOXP2[Title/Abstract]) AND "speech development"[Title/Abstract]) AND mutation[Title/Abstract] | <a href="#">3</a>      | 09:43:29 |
| #14    | <a href="#">Add</a> | Search mutation[Title/Abstract]                                                                        | <a href="#">288772</a> | 09:42:12 |
| #13    | <a href="#">Add</a> | Search "speech development"[Title/Abstract]                                                            | <a href="#">897</a>    | 09:41:57 |
| #12    | <a href="#">Add</a> | Search FOXP2[Title/Abstract]                                                                           | <a href="#">387</a>    | 09:41:34 |

## Search results

Items: 3

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

1. [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

[Similar articles](#)

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

2. 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](#)

[Similar articles](#)

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

3. [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](#)

[Similar articles](#)

# 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



Search

Create alert   Limits   Advanced

Help

## All MeSH Categories

### Diseases Category

#### Pathological Conditions, Signs and Symptoms

##### Signs and Symptoms

##### Neurologic Manifestations

##### Neurobehavioral Manifestations

##### Communication Disorders

##### Language Disorders

##### **Language Development Disorders**

# FOXP2 + MeSH

PubMed Advanced Search Builder

[YouTube](#) Tutorial

((FOXP2[Title/Abstract]) AND mutation[Title/Abstract]) AND language disorders[Title/Abstract]

[Edit](#)

[Clear](#)

## Builder

|            |   |                       |                   |                                    |                   |                                 |                                 |
|------------|---|-----------------------|-------------------|------------------------------------|-------------------|---------------------------------|---------------------------------|
| All Fields | ↔ | FOXP2[Title/Abstract] | <a href="#">-</a> | <a href="#">Show index list</a>    |                   |                                 |                                 |
| AND        | ↔ | All Fields            | ↔                 | mutation[Title/Abstract]           | <a href="#">-</a> | <a href="#">Show index list</a> |                                 |
| AND        | ↔ | All Fields            | ↔                 | language disorders[Title/Abstract] | <a href="#">-</a> | <a href="#">Show index list</a> |                                 |
| AND        | ↔ | All Fields            | ↔                 |                                    | <a href="#">-</a> | <a href="#">+</a>               | <a href="#">Show index list</a> |

[Search](#) or [Add to history](#)

## History

[Download history](#) [Clear history](#)

| Search | Add to builder      | Query                                                                                                  | Items found            | Time     |
|--------|---------------------|--------------------------------------------------------------------------------------------------------|------------------------|----------|
| #17    | <a href="#">Add</a> | Search ((FOXP2[Title/Abstract]) AND mutation[Title/Abstract]) AND language disorders[Title/Abstract]   | <a href="#">12</a>     | 09:47:57 |
| #16    | <a href="#">Add</a> | Search language disorders[Title/Abstract]                                                              | <a href="#">1398</a>   | 09:47:39 |
| #15    | <a href="#">Add</a> | Search ((FOXP2[Title/Abstract]) AND "speech development"[Title/Abstract]) AND mutation[Title/Abstract] | <a href="#">3</a>      | 09:45:02 |
| #14    | <a href="#">Add</a> | Search mutation[Title/Abstract]                                                                        | <a href="#">288772</a> | 09:42:12 |
| #13    | <a href="#">Add</a> | Search "speech development"[Title/Abstract]                                                            | <a href="#">897</a>    | 09:41:57 |
| #12    | <a href="#">Add</a> | Search FOXP2[Title/Abstract]                                                                           | <a href="#">387</a>    | 09:41:34 |

# FOXP2 + MeSH

## Search results

Items: 12

- [Ultrasonic vocalizations of adult male \*\*Foxp2\*\*-mutant mice: behavioral contexts of arousal and emotion.](#)  
1. [Gaub S, Fisher SE, Ehret G.](#)  
Genes Brain Behav. 2016 Feb;15(2):243-59. doi: 10.1111/gbb.12274. Epub 2016 Jan 13.  
PMID: 26566793  
[Similar articles](#)
  
- [Assessing the effects of common variation in the \*\*FOXP2\*\* gene on human brain structure.](#)  
2. [Hoogman M, Guadalupe T, Zwiers MP, Klarenbeek P, Francks C, Fisher SE.](#)  
Front Hum Neurosci. 2014 Jul 1;8:473. doi: 10.3389/fnhum.2014.00473. eCollection 2014.  
PMID: 25013396    [\*\*Free PMC Article\*\*](#)  
[Similar articles](#)
  
- [Recent Advances in the Genetics of Vocal Learning.](#)  
3. [Condro MC, White SA.](#)  
Comp Cogn Behav Rev. 2014;9:75-98.  
PMID: 26052371    [\*\*Free PMC Article\*\*](#)  
[Similar articles](#)
  
- [\*\*Foxp2\*\* mutations impair auditory-motor association learning.](#)  
4. [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\*\*](#)  
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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

((Lai[Author]) AND FOXP2[Title/Abstract]) AND ("2001"[Date - Publication] : "2001"[Date - Publication])

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## Builder

Author  [Show index list](#)

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# FOXP2 Query Building

Format: Abstract ▾

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Nature. 2001 Oct 4;413(6855):519-23.

## A forkhead-domain gene is mutated in a severe speech and language disorder.

Lai CS<sup>1</sup>, Fisher SE, Hurst JA, Vargha-Khadem F, Monaco AP.

### Author information

#### 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

# Search Results Page

## Article types

Clinical Trial

Review

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## Text availability

Abstract

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Trending articles

## Publication dates

5 years

10 years

Custom range...

## Species

Humans

Other Animals

## Sex

Female

Male

## Journal

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## Search results

Items: 12

- [Ultrasonic vocalizations of adult male \*\*Foxp2\*\*-mutant mice: behavioral contexts of arousal and emotion.](#)

1. Gaub S, Fisher SE, Ehret G.  
Genes Brain Behav. 2016 Feb;15(2):243-59. doi: 10.1111/gbb.12274. Epub 2016 Jan 13.  
PMID: 26566793  
[Similar articles](#)

- [Assessing the effects of common variation in the \*\*FOXP2\*\* gene on human brain structure.](#)

2. Hoogman M, Guadalupe T, Zwiers MP, Klarenbeek P, Francks C, Fisher SE.  
Front Hum Neurosci. 2014 Jul 1;8:473. doi: 10.3389/fnhum.2014.00473. eCollection 2014.  
PMID: 25013396    [Free PMC Article](#)  
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- [Recent Advances in the Genetics of Vocal Learning.](#)

3. Condro MC, White SA.  
Comp Cogn Behav Rev. 2014;9:75-98.  
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[Mutation](#)



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## Article types

- Clinical Trial
- Review
- Customize ...

## Text availability

- Abstract
- Free full text
- Full text

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- Reader comments
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- Custom range...

## Species

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Items: 12

1

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2016 Feb;15(2):243-59. doi:

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# Filtering Search Results



H3ABioNet

**Article types**

Clinical Trial

Review

Customize ...

**Text availability**

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Abstract

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**Items: 7**

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[Assessing the effects of common variation in the FC](#)

1. Hoogman M, Guadalupe T, Zwiers MP, Klarenbeek P, Front Hum Neurosci. 2014 Jul 1;8:473. doi: 10.3389/fnhum.2014.00473. PMID: 25013396 [Free PMC Article](#)  
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2. Condro MC, White SA. Comp Cogn Behav Rev. 2014;9:75-98. PMID: 26052371 [Free PMC Article](#)  
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[Foxp2 mutations impair auditory-motor association](#)

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Pan African Bioinformatics Network for H3Africa

# Free Full Text Articles

Format: Abstract ▾

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

## Foxp2 mutations impair auditory-motor association learning.

Kurt S<sup>1</sup>, Fisher SE, Ehret G.

 Author information

### Abstract

Heterozygous mutations of the human FOXP2 transcription factor gene cause the best-described examples of monogenic speech and language disorders. Acquisition of proficient spoken language involves auditory-guided vocal learning, a specialized form of sensory-motor association learning. The impact of etiological Foxp2 mutations on learning of auditory-motor associations in mammals has not been determined yet. Here, we directly assess this type of learning using a newly developed conditioned avoidance paradigm in a shuttle-box for mice. We show striking deficits in mice heterozygous for either of two different Foxp2 mutations previously implicated in human speech disorders. Both mutations cause delays in acquiring new motor skills. 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. These results are consistent with expression of Foxp2 in distributed circuits of the cortex, striatum and cerebellum that are known to play key roles in acquisition of motor skills and sensory-motor association learning, and suggest differing *in vivo* effects for distinct variants of the Foxp2 protein. Given the importance of such networks for

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Review FOXP2 as a molecular window into speech and language. [Trends Genet. 2009]

The structure of innate vocalizations in Foxp2-deficient mouse pup [Genes Brain Behav. 2010]

Foxp2 regulates gene networks implicated in

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- Summary (text)
- Abstract
- Abstract (text)
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[Actions of adult male \*\*Foxp2\*\*-mutant mice: beha](#)

Gaub S, Fisher SE, Ehret G.

Genes Brain Behav. 2016 Feb;15(2):243-59. doi: 10.1111/gbb.12274. Epub 2

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**Items:** 12



Ultrasonic emotion.

1.

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2-mutant mice: behav

Gaub S, Fisher SE, Ehret G.

Genes Brain Behav. 2016 Feb;15(2):243-59. doi: 10.1111/gbb.12274. Epub 20

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[oice: behavioral contexts of arousal and](#)

274. Epub 2016 Jan 13.

## Search details

(FOXP2[Title/Abstract] AND mutation[Title/Abstract]) language disorders[Title/

# 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