

100 YEARS OF PROGRESS:

II International research and practice online conference, Mongolia, May 5, 2022

TEACHING MEDICAL GENETICS USING MODERN DIGITAL TOOLS ONLINE

*Professor Leila Akhmadeeva
Bashkir State Medical University, Ufa, Russia*





- *I live and work in Ufa, Russia*
- I started teaching genetics to medical students in 1994
- I'm sure that midwives are, probably, the most important health care professionals



Family History Screening: Use of the Three Generation Pedigree in Clinical Practice

Jo-Ann K. Brock, MD, PhD, FRCSC,¹ Victoria M. Allen, MD, MSc, FRCSC,¹
Katharina Kieser, MD, MSc, FRCSC,¹ Sylvie Langlois, MD, FRCPC²

¹Department of Obstetrics and Gynaecology, Dalhousie University, Halifax NS

²Department of Medical Genetics, University of British Columbia, Vancouver BC

Abstract

With a growing understanding of genetic disorders in the scientific and lay literature, it is becoming increasingly important to consider risk factors based on family history and ethnicity for identifying individuals for whom genetic testing is indicated and will be most beneficial. A

You are seeing Mary Smith, a 31-year-old patient, for a routine well woman exam. She tells you that she is considering pregnancy, but is concerned because her brother was affected with autism.

J Obstet Gynaecol Can 2010;32(7):663–672





Mary herself is healthy, and she has two brothers. One of her brothers was diagnosed as a young child as having autism, and he died at age eight of an accidental drowning. Her younger brother is aged 25 and has no health concerns. Both of her parents are living and in general good health. Her mother, age 56, has recent onset of hypothyroidism; her father, age 59, takes medication for high blood pressure.

Mary's mother has one sister and a brother who is deceased. She reports that her maternal aunt is healthy, and has three healthy teenage children. Mary has little information regarding her mother's brother, other than that he was institutionalized for most of his life with mental retardation and died at age 32. Her maternal grandparents both died in their 80s. They were relatively healthy for most of their lives.

Mary's father has one sister and three brothers. Her paternal aunt died of colon cancer at age 48; she had no children. One of Mary's paternal uncles also died of colon cancer at the age of 45; he had three boys, who are currently alive and healthy. One of these cousins has a three-year-old daughter with no health concerns. Mary has two other paternal uncles who are alive and healthy; one of these uncles has two children, a daughter age 28 and son age 25, who are both in good health.

Mary's paternal grandmother is alive at age 87 and in general good health. Her paternal grandfather died at age 62 of complications related to ischemic heart disease.



The first thing to learn/teach in medical genetics is **FAMILY TREE**

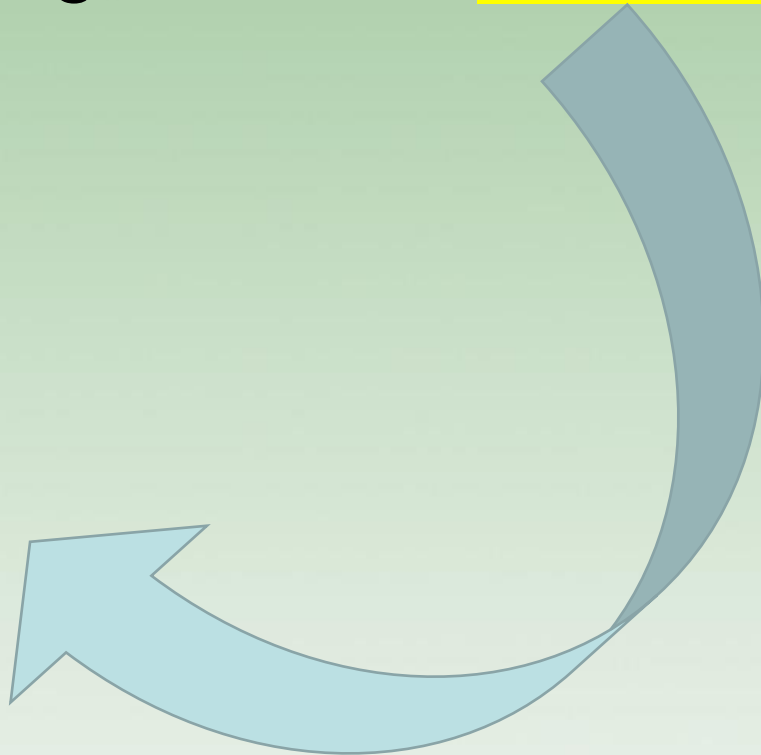




Figure 3. Family history questionnaire

(Please complete to the best of your ability)

Contact/registration information (please complete all areas)

Full name of referred individual	Previous name	Date of birth (D/M/Y)	Provincial health card #	Health card expiry
Home phone	Work/other phone	Complete mailing address		

Family information: Please fill in for family of person being referred

Father (full name)	Date of birth (D/M/Y), if known	Ethnic background
Mother (full name)	Date of birth (D/M/Y), if known	
Sister/brother (full name)	Date of birth (D/M/Y), if known	
Sister/brother (full name)	Date of birth (D/M/Y), if known	

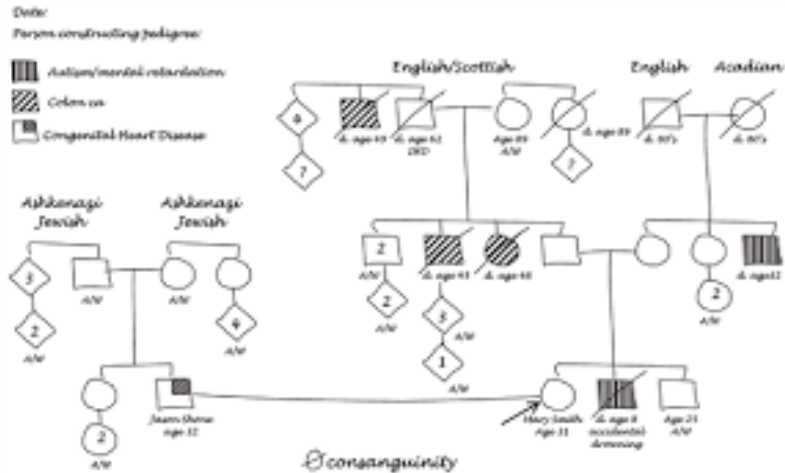
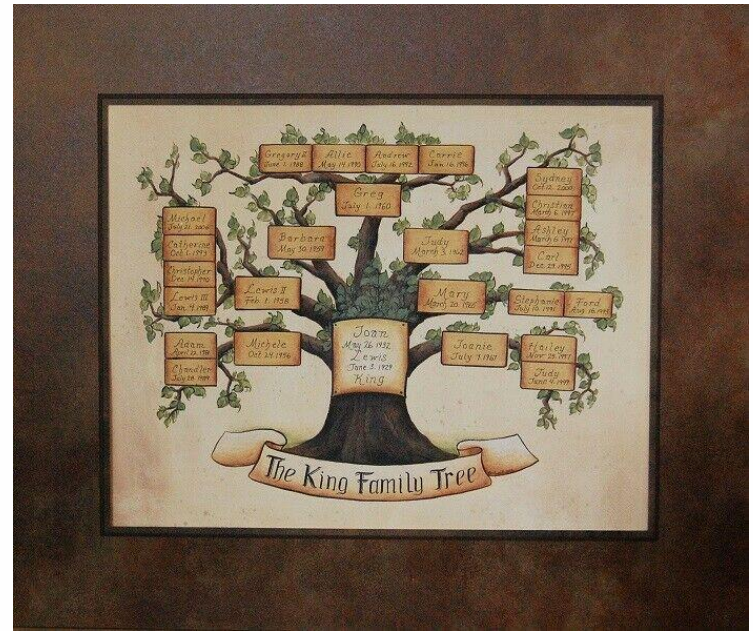
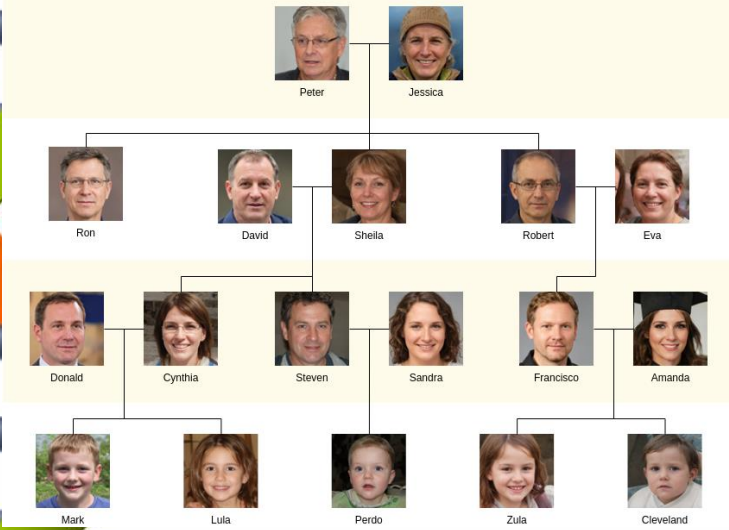
Do all these brothers/sisters share both the same parents? Yes No

If no, please explain _____

Are there any husband and wife couples in your family who are related by blood? Yes No

Please list any personal health problems (past or present) for the person being referred.

FAMILY TREE



During the era of digital technologies and Internet



Google × 🔍 🗨️ 🔊 🔍
ОСТАВИТЬ ОТЗЫВ

<https://www.holonic-systems.com> > ... ▼ [Перевести эту страницу](#)
Genetic pedigree chart creation software f-tree
f-tree is a **software** application that automatically creates **medical pedigree** chart of a family by simply using the family's information, which is provided ...
18 февр. 2016 г. · Добавлено пользователем TogoTV

<https://www.trakgene.com> ▼ [Перевести эту страницу](#)
Pedigree Drawing Tool | Clinical Genetics Database Software
Our integrated **pedigree** drawing tool and clinical genetics database **software** is used by clinical genetics services around the world to manage patient data.

<https://www.researchgate.net> > ... ▼ [Перевести эту страницу](#)
Anyone know of a useful pedigree drawing program?
20 ответов
PhenoTips (<https://phenotips.org/>) **software** has a built-in **pedigree** drawing tool that is very easy to use. Moreover, it is free to use for research purposes and ...

<https://bmcmmedgenet.biomedcentral.com> > ... ▼ [Перевести эту страницу](#)
f-treeGC: a questionnaire-based family tree-creation software ...
автор: T Tokutomi · 2017 · Цитируется: 4 — The Tohoku **Medical** Megabank project aims to create a next-generation personalized healthcare system by conducting large-scale genome-...

<https://bmcmmedgenet.biomedcentral.com> > ... ▼ [Перевести эту страницу](#)
PediDraw: A web-based tool for drawing a pedigree in genetic ...
автор: M He · 2007 · Цитируется: 17 — In **medical** records, a **pedigree** is useful to document the family history ... Currently, most **pedigrees** are drawn by hand or by drawing **software**.

<https://www.jax.org> > pedigree... ▼ [Перевести эту страницу](#)
Pedigree Tool - The Jackson Laboratory
Pedigree Tool. Collecting and recording family health history in a **pedigree** format can help identify patterns suggestive of genetic risk. Use this **pedigree** ...

<https://gitmind.com> > pedigree... ▼ [Перевести эту страницу](#)
7 Best Free Online Pedigree Chart Maker Websites in 2022

**S
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BUT

- Most of them are **expensive**
- Most of them **do not** give you a chance to make it look **professional**
- Most of them are **difficult** to learn and manage

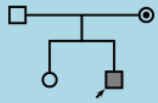




We use

<https://pedigree.varphi.com/>





A complete pedigree is often a work of great labour, and its finished form is frequently a work of art. Karl Pearson, 1912.

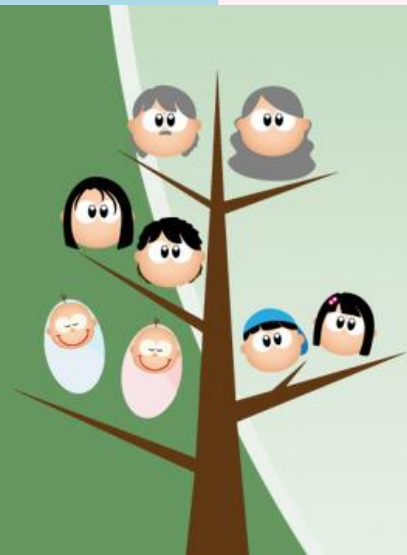
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- Retrieve a Saved Pedigree
- Screenshots
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Thanks for visiting the site about medical pedigrees—an important tool for researchers, geneticists, students and educators. You can find more about pedigrees in the papers in our [bibliography section](#). We provide a set of free programs for creating high quality pedigrees based on TeX and PostScript. Check out [the download section](#). You may also want to browse [the documentation](#). You can try to make a pedigree without downloading or installing anything with the [online version](#) of our programs. Please read the [Terms of Service](#) and the [Privacy Policy](#) before uploading anything to our site.

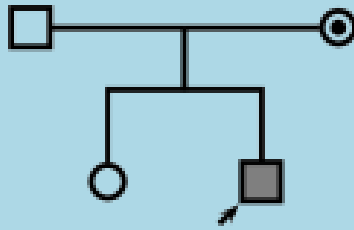
News

- 2018/05/19: Updated [screenshots](#).
- 2010/05/04: Now you can retrieve a previously saved pedigree.
- 2010/05/03: Updated [screenshots](#).
- 2010/01/04: New help function in the [online tool](#). Just move your mouse over the column headers in the table.
- 2010/01/04: Support for arbitrary family size in [online tool](#).
- 2009/12/23: New [screenshots](#).
- 2009/12/18: Now you can [create a pedigree chart](#) online.
- 2009/12/11: The site is launched!

\$Revision: 1.10 \$ \$Date: 2018/05/20 02:05:16 \$



- *Free*
- *Easy to operate*
- *Creates very professional pedigree charts that one can use for publishing papers*



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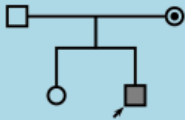
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- Home
- Download
- Documentation
- Create a Pedigree
- Retrieve a Saved Pedigree
- Screenshots
- Bibliography
- Terms of Service
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Create a pedigree

This tool allows you to create a pedigree without installing anything on your computer. It has some limitations compared to our [full suite](#): there is no support for drawing symbols for twins, infertility symbols. Use the full suite if you need these features. Of course, the full suite is free.

Please fill the table below. If you need more table rows, just press "Add 5 rows" as many times as you wish. Do not forget to anonymize the patients!

Move the mouse over a column header in the table to get help for the column. See [screenshots](#) for an example of input and output.

Id	Name	Sex	Date of Birth	Date of Death	Mother	Father	Condition	Proband	Comment	Sort Order
A0	<input type="text"/>	unknown ▼	<input type="text"/>	alive	▼	▼	normal ▼	<input checked="" type="radio"/>	<input type="text"/>	<input type="text"/>
A1	<input type="text"/>	unknown ▼	<input type="text"/>	alive	▼	▼	normal ▼	<input type="radio"/>	<input type="text"/>	<input type="text"/>
A2	<input type="text"/>	unknown ▼	<input type="text"/>	alive	▼	▼	normal ▼	<input type="radio"/>	<input type="text"/>	<input type="text"/>
A3	<input type="text"/>	unknown ▼	<input type="text"/>	alive	▼	▼	normal ▼	<input type="radio"/>	<input type="text"/>	<input type="text"/>
A4	<input type="text"/>	unknown ▼	<input type="text"/>	alive	▼	▼	normal ▼	<input type="radio"/>	<input type="text"/>	<input type="text"/>

Check the fields you want to be shown on the *chart* (you probably do not want to clutter the chart with too many fields; besides, they all are always present on the *legend*):

- Id
- Name
- Date of Birth
- Date of Death
- Age at Death
- Comment

I agree to the [Terms of Service](#) and certify that my input does not contain individually Identifiable Health Information as defined by [HIPAA Privacy Laws in the USA](#) and similar laws of my jurisdiction.

Mary herself is healthy, and she has two brothers. One of her brothers was diagnosed as a young child as having autism, and he died at age eight of an accidental drowning. Her younger brother is aged 25 and has no health concerns. Both of her parents are living and in general good health. Her mother, age 56, has recent onset of hypothyroidism; her father, age 59, takes medication for high blood pressure.

Id	Name	Sex	Date of Birth	Date of Death	Mother	Father	Condition	Proband
A0	Mary	female	unknown	alive	A3	A4	normal	<input checked="" type="radio"/>
A1	Brother1	male	unknown	2020	A3	A4	affected	<input type="radio"/>
A2	Brother2	male	unknown	alive	A3	A4	normal	<input type="radio"/>
A3	Mother	unknown	unknown	alive			normal	<input type="radio"/>
A4	Father	unknown	unknown	alive			normal	<input type="radio"/>

I agree to the [Terms of Service](#) and certify that my input does not contain individually Identifiable Health Information as defined by [HIPAA Privacy Laws in the USA](#) and similar laws of my jurisdiction.

Create Pedigree

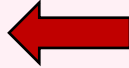


I agree to the [Terms of Service](#) and certify that my input does not contain individually Identifiable Health Information as defined by [HIPAA Privacy Laws in the USA](#) and similar laws of my jurisdiction.


Create Pedigree

Here is your chart:


 [PNG File](#)



 [GIF File](#)

 [PDF File](#)

 [PostScript File](#)

 [DVI File](#)

 [TeX File](#)

- I:1 Father; born: unknown.
- I:2 Mother; born: unknown.
- II:1 Brother2; born: unknown.
- II:2 Mary; born: unknown.
- II:3 Brother1; born: unknown; age at death: unknown.

Here is the number of your pedigree:

Pedigree number: 7153803221352

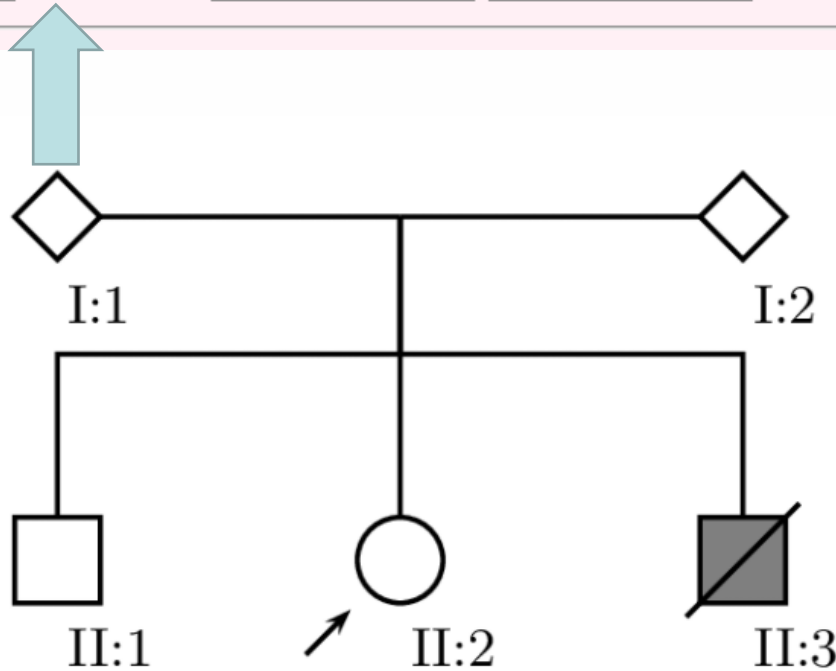
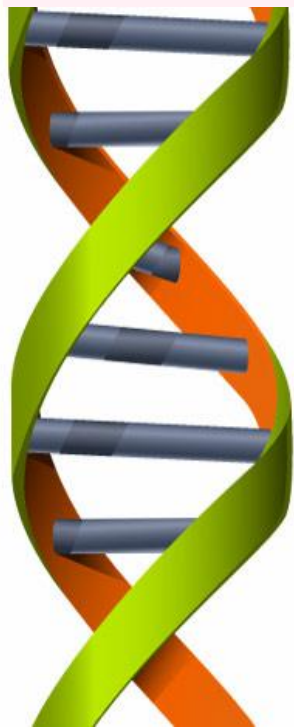
Write down this number to [retrieve](#) your pedigree later.

Any format



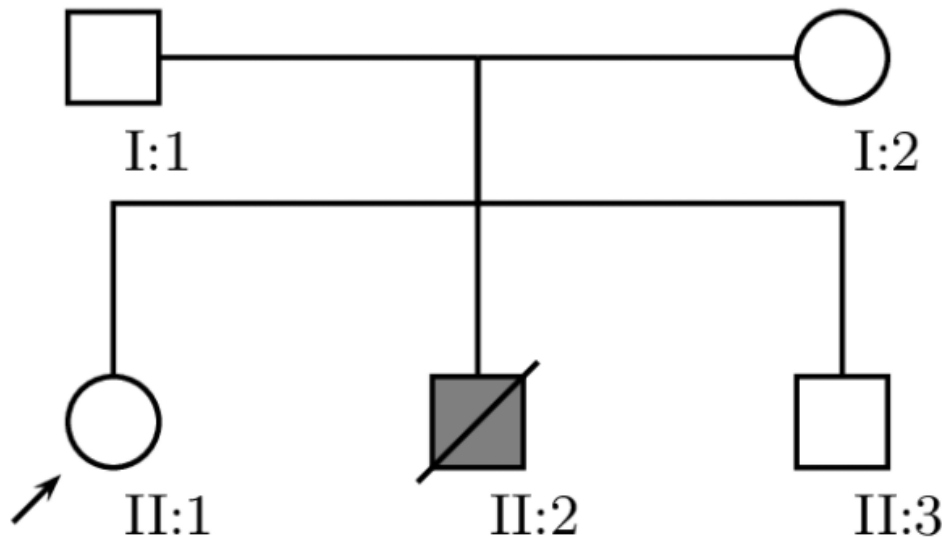
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A1	Brother1	male	unknown	2020	A3	A4	affected	<input type="radio"/>
A2	Brother2	male	unknown	alive	A3	A4	normal	<input type="radio"/>
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A3	Mother	female	unknown	alive			normal	<input type="radio"/>
A4	Father	male	unknown	alive			normal	<input type="radio"/>



Id	Name	Sex	Date of Birth	Date of Death	Mother	Father	Condition	Proband
A0	Mary	female ▾	unknown	alive	A3 ▾	A4 ▾	normal ▾	<input checked="" type="radio"/>
A1	Brother1	male ▾	unknown	2020	A3 ▾	A4 ▾	affected ▾	<input type="radio"/>
A2	Brother2	male ▾	unknown	alive	A3 ▾	A4 ▾	normal ▾	<input type="radio"/>
A3	Mother	female ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A4	Father	male ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A5		unknown ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A6		unknown ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A7		unknown ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A8		unknown ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>
A9		unknown ▾	unknown	alive	▾	▾	normal ▾	<input type="radio"/>

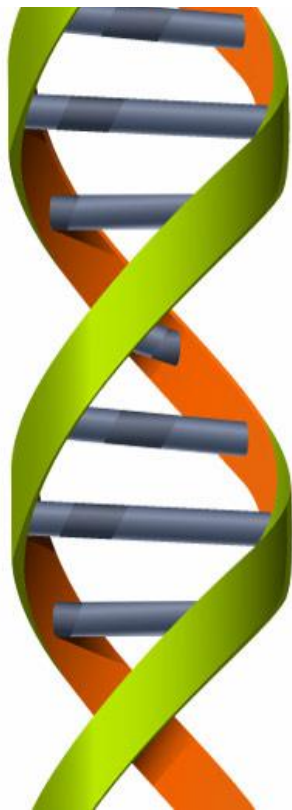
Add 5 rows

Delete 5 rows



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Mary's mother has one sister and a brother who is deceased. She reports that her maternal aunt is healthy, and has three healthy teenage children. Mary has little information regarding her mother's brother, other than that he was institutionalized for most of his life with mental retardation and died at age 32. Her maternal grandparents both died in their 80s. They were relatively healthy for most of their lives.

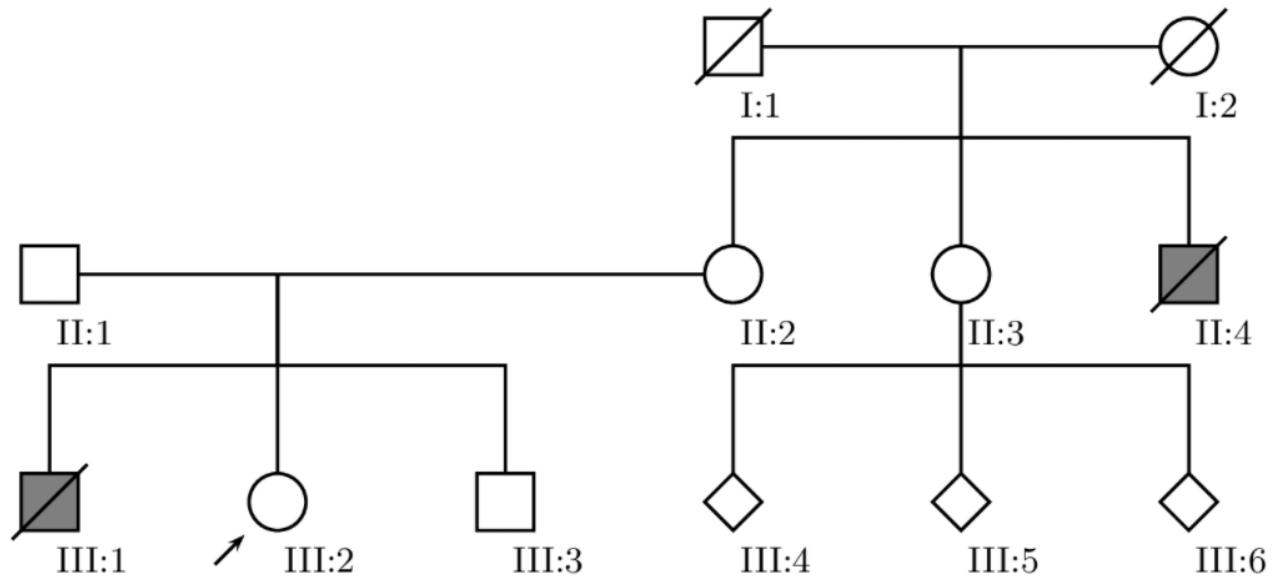


Id	Name	Sex	Date of Birth	Date of Death	Mother	Father	Condition	Proband
A0	Mary	female	unknown	alive	A3	A4	normal	<input checked="" type="radio"/>
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A2	Brother2	male	unknown	alive	A3	A4	normal	<input type="radio"/>
A3	Mother	female	unknown	alive	A5	A6	normal	<input type="radio"/>
A4	Father	male	unknown	alive			normal	<input type="radio"/>
A5	Grandmother M	female	unknown	2022			normal	<input type="radio"/>
A6	Grandfather M	male	unknown	2022			normal	<input type="radio"/>
A7	Mother's brother	male	unknown	2021	A5	A6	affected	<input type="radio"/>
A8	Mother's sister	female	unknown	alive	A5	A6	normal	<input type="radio"/>
A9	Child1 of the aunt	unknown	unknown	alive	A8		normal	<input type="radio"/>
A10	Child2 of the aunt	unknown	unknown	alive	A8		normal	<input type="radio"/>
A11	Child3 of the aunt	unknown	unknown	alive	A8		normal	<input type="radio"/>
A12		unknown	unknown	alive			normal	<input type="radio"/>
A13		unknown	unknown	alive			normal	<input type="radio"/>
A14		unknown	unknown	alive			normal	<input type="radio"/>



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Mary's father has one sister and three brothers. Her paternal aunt died of colon cancer at age 48; she had no children. One of Mary's paternal uncles also died of colon cancer at the age of 45; he had three boys, who are currently alive and healthy. One of these cousins has a three-year-old daughter with no health concerns. Mary has two other paternal uncles who are alive and healthy; one of these uncles has two children, a daughter age 28 and son age 25, who are both in good health.

Mary's paternal grandmother is alive at age 87 and in general good health. Her paternal grandfather died at age 62 of complications related to ischemic heart disease.



We can add as many rows as needed

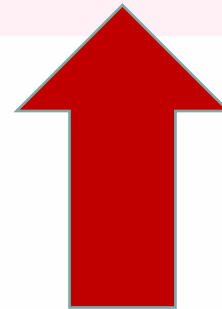




Here is the number of your pedigree:

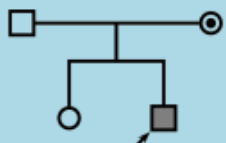
Pedigree number: 7153803221352

Write down this number to [retrieve](#) your pedigree later.



We can add as many rows as needed
WHENEVER we need it!





A complete pedigree is often a work of great labour, and its finished form is frequently a work of art. Karl Pearson, 1912.

- Home
- Download
- Documentation
- Create a Pedigree
- Retrieve a Saved Pedigree
- Screenshots
- Bibliography
- Terms of Service
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Create a pedigree

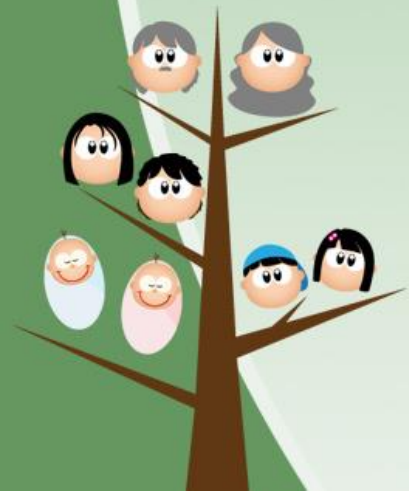
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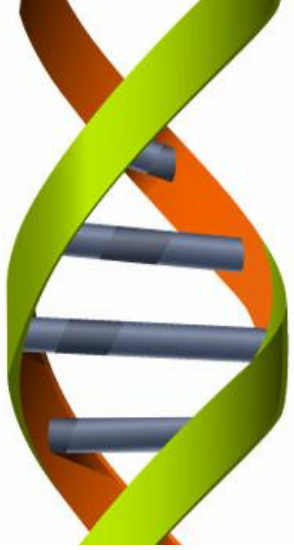
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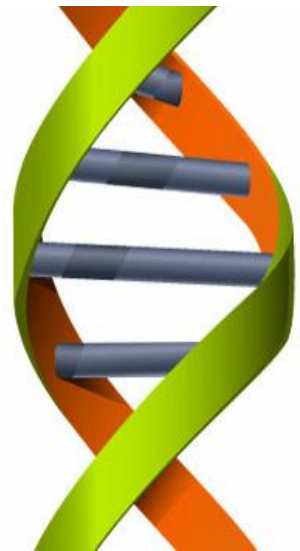
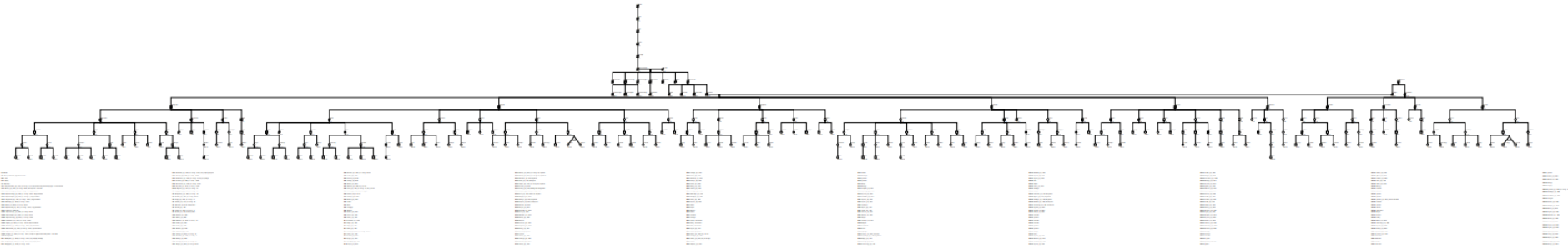
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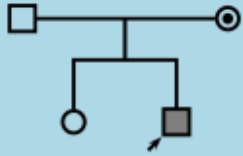
You can make it BIG





Example: **my** family tree





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- Download
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- Create a Pedigree
- Retrieve a Saved Pedigree
- Screenshots** ←
- Bibliography
- Terms of Service
- Privacy Policy
- Contact Us

Some Screenshots

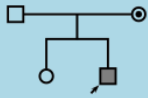
[Pedigree for Akhmadeeva family \(Legend in Russian\)](#)

Online tool: Haemofilia in European royalty (based on http://en.wikipedia.org/wiki/Haemophilia_in_European_royalty)

1. [Input](#)
2. [Output: Legend.](#)
3. [Output: Chart.](#)

\$Revision: 1.3 \$ \$Date: 2010-05-03 20:47:21 \$





A complete pedigree is often a work of great labour, and its finished form is frequently a work of art. Karl Pearson, 1912.

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Selected papers and books about pedigrees

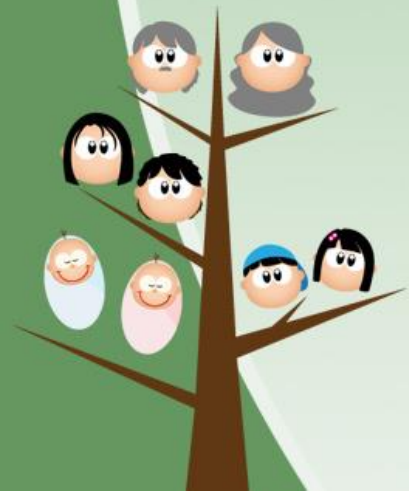
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You can [download](#) the list of references in the BibTeX format.

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Why do midwives may want to use it and learn it?



Genomics in Midwifery

Key notes

Learning

Teaching

Updates

Introduction

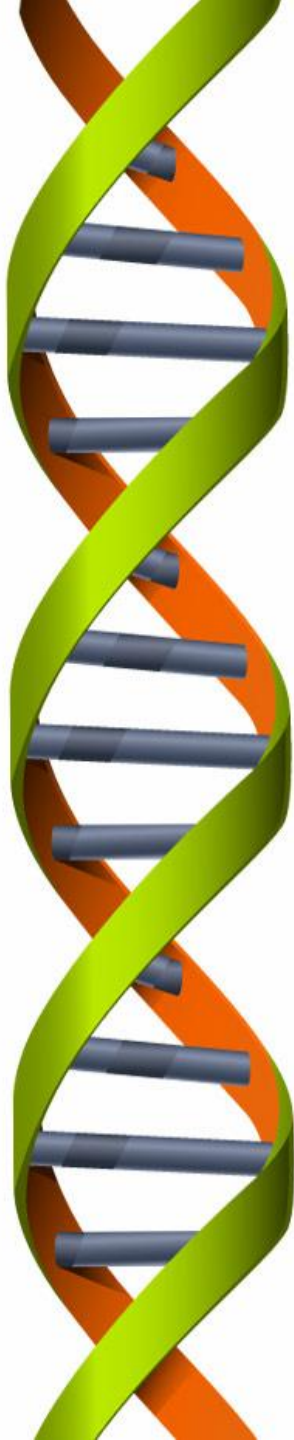
Thank you to members of our nursing and midwifery round table, the education team at the Royal College of Midwives, Donna Kirwan, Joanne Hargrave and Laura-Rose Thorood for their input to this resource.

Genomics is fundamental to your practice as a midwife – it allows you to predict and prepare for situations in pregnancy to better protect and care for the parent and child.

As genomics is now included in the NMC standards of proficiency for midwives



As genomics is now included in the NMC standards of proficiency for midwives and, with rapid advances being made in the field all the time, it's important that *you can talk confidently about it and understand its relevance before, during and after pregnancy.*



100 YEARS OF PROGRESS:

II International research and practice online conference, Mongolia, May 5, 2022

**TEACHING
MEDICAL
GENETICS
USING
MODERN
DIGITAL
TOOLS
ONLINE**



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