NATIONAL SENIOR CERTIFICATE EXAMINATION
NOVEMBER 2022

LIFE SCIENCES: PAPER II

SOURCE MATERIAL BOOKLET FOR QUESTIONS 1, 2 AND 3

The numbers in bold on the right after each piece of information refer to the references that are in endnotes starting on page xvii.

SECTION A

QUESTION 1

Read the information below. Refer to this information, as well as your own knowledge, to answer Question 1 in the question paper.

Vampires may be real people with a blood disorder

1. Porphyria

Porphyria is a group of genetic diseases that affect the body's ability to make a substance called **haem**. Haem has many functions in the body – the most important of which is to join to iron to form a molecule called **haemoglobin** in red blood cells. Haemoglobin carries oxygen and gives the blood its characteristic red colour.

The process of **haem** production occurs via a **metabolic pathway**. A metabolic pathway is a series of chemical reactions with each step being controlled by an enzyme – enzymes are proteins made during protein synthesis. There are eight of these steps in the pathway to make **haem**, starting with the amino acid 'glycine' and an enzyme. The in-between products formed in the pathway are called **porphyrins** (Figure 1.1).

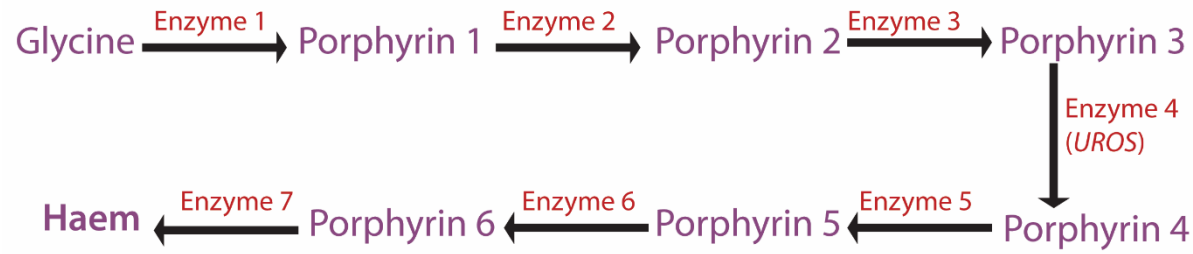

Figure 1.1: Metabolic pathway leading to the production of **haem**

If an enzyme controlling any step is mutated, or not produced, the whole process of **haem** production stops, leading to a build-up of certain types of **porphyrin**. Porphyrins are toxic in large quantities. Porphyrins are a brown-red colour. They build up in the skin and other organs before being excreted in faeces and urine (which may turn a blood-red or purple colour)*.

*Porphyria comes from the ancient Greek '*porphura*', meaning purple – referring to the colour of the urine.

2. Congenital Erythropoietic Porphyria (CEP)

Congenital erythropoietic porphyria (CEP) is a very rare form of porphyria resulting from incorrect functioning of an enzyme known as the *UROS* enzyme:

- This is the fourth enzyme in the ***haem*** synthesis pathway (see Figure 1.1).
- It is coded for by the *U1* gene.
- The *U1* gene is located on chromosome 10.
- The *UROS* enzyme consists of 265 amino acids.
- The faulty version of the *U1* gene has a mistake in the DNA sequence where one nucleotide has been substituted by another. This results in the amino acid **arginine** being substituted for **cysteine** after transcription and translation have occurred to make the *UROS* enzyme.

- The resulting defective *UROS* enzyme cannot turn porphyrin 3 into porphyrin 4. This leads to abnormally high levels of porphyrin 3 in the erythrocytes.
- This mutation is recessive.
- It is estimated that the disorder occurs in about **1 in 74 300 individuals** in South Africa.

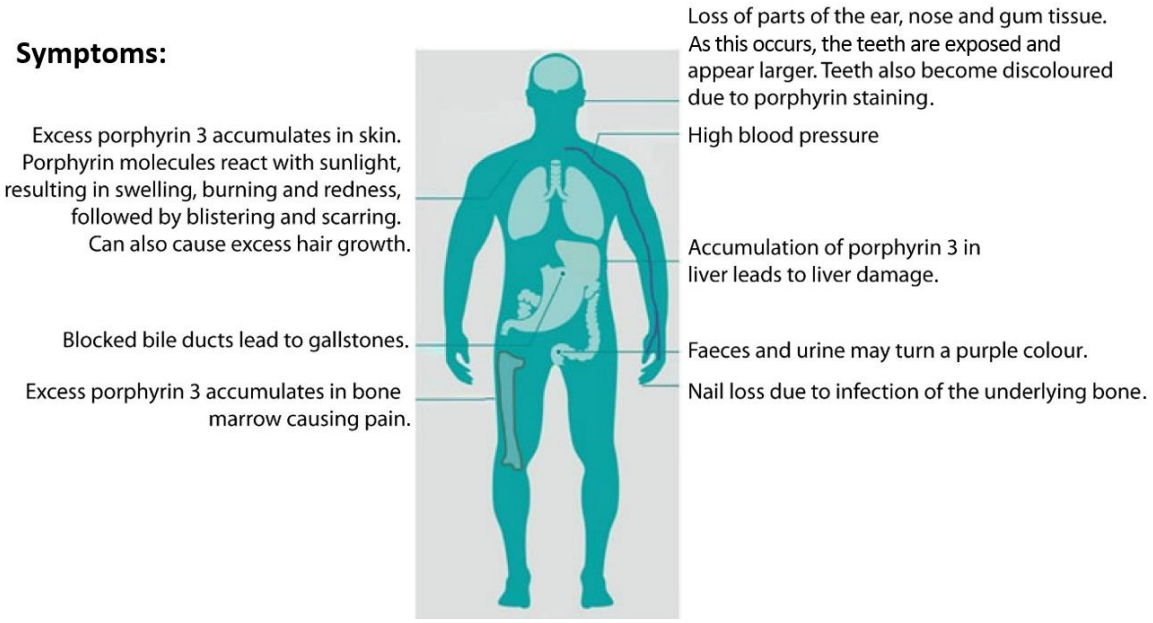

Figure 1.2: Symptoms of CEP **1**

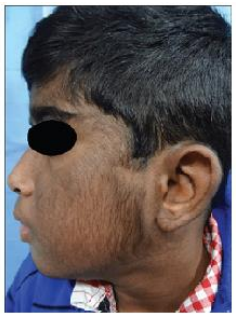

Figure 1.3: (a) Excess hair growth in a person with CEP

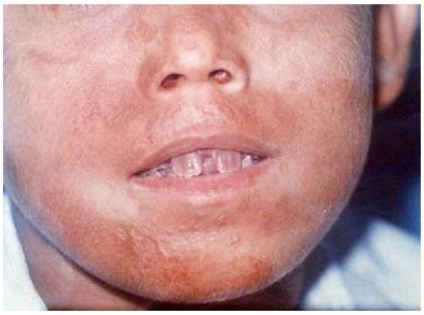

Figure 1.3: (b) Skin and teeth of a person suffering from CEP

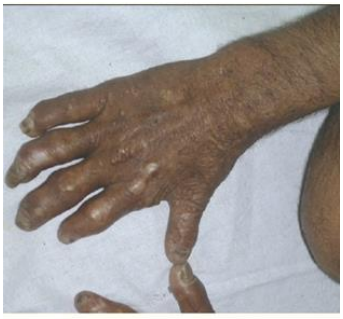

Figure 1.3: (c) Mutilated hands resulting from CEP

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3. What is a vampire?

Some of the physical and behavioural features of people with CEP show striking similarities to fictional creatures known as vampires. Vampires are evil imaginary beings who are supposed to wander around at night searching for people upon whose blood they feed.

Vampires have captured people's imagination for hundreds of years. They have been portrayed in various ways, such as in television series like *Vampire Diaries* and *True Blood* and in books such as Bram Stoker's *Dracula*. Nowadays most people accept that vampires are fictional.

It is now thought that many people who were accused of being vampires in the past could actually have been suffering from CEP.

Figure 1.4: Vampire characteristics

3

4. Pedigree

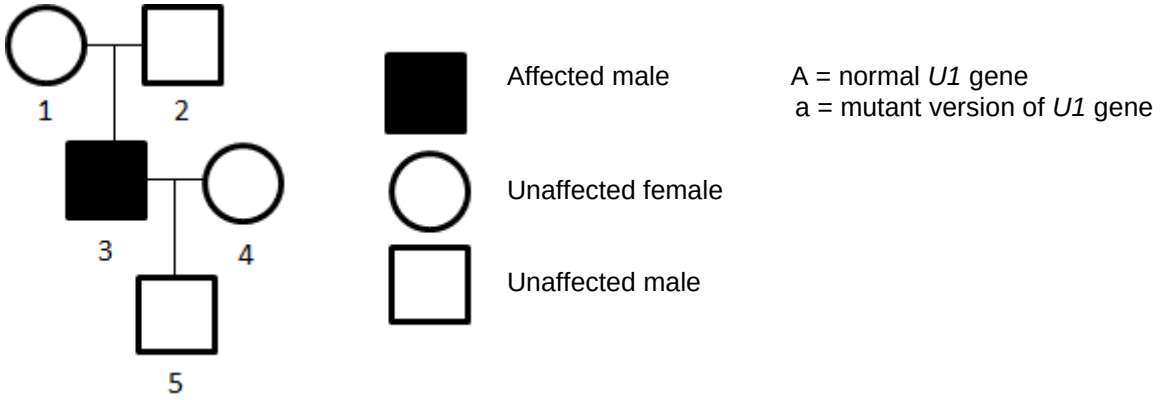

Figure 1.5: Pedigree of a family with CEP

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5. Treatments for CEP

1. Change in behaviour

- Avoid sunlight and apply sunscreen when outside.
- Wear clothing that covers as much of the body as possible, including hats and sunglasses.
- Avoid foods high in sulphur (e.g., garlic) as they worsen symptoms.

2. Medications and therapies

- Doctors used to advise patients to eat meat or even drink animal blood to provide **haem**.
- Genetic counselling is recommended for affected individuals and their families.

3. Genetic techniques

- Gene therapy.
- CRISPR.

The CRISPR Cas-9 gene-editing procedure was pioneered by researchers Jennifer Doudna and Emmanuele Charpentier and they received the 2021 Nobel Prize for Chemistry for its discovery – the first time a team of only women had received the prize in this field.

Figure 1.6: Professors Jennifer Doudna (left) and Emmanuele Charpentier (right)

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Experiments are currently underway to determine whether the method of CRISPR gene editing can be conducted to 'fix' the mutation in the *U1* gene causing CEP. The process is shown in Figure 1.7 below.

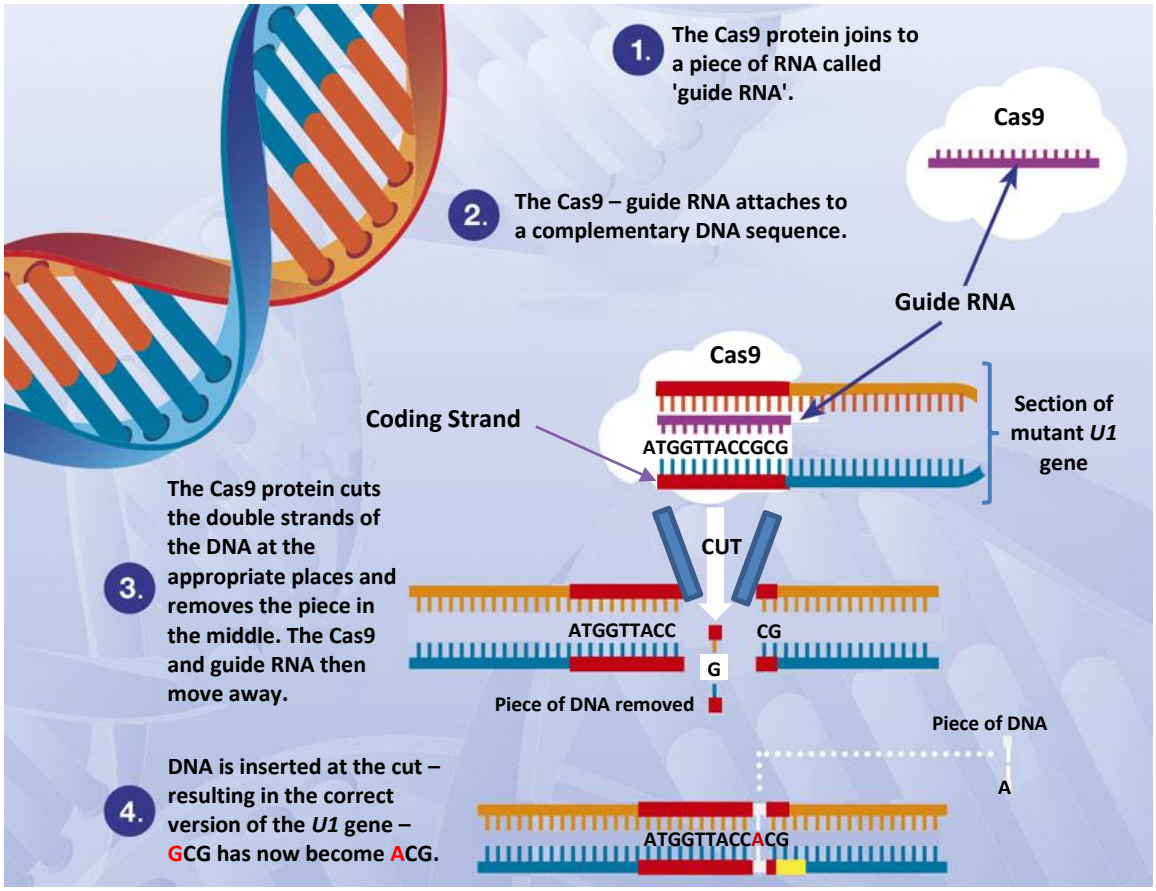

Figure 1.7: CRISPR editing to correct mutation-causing CEP

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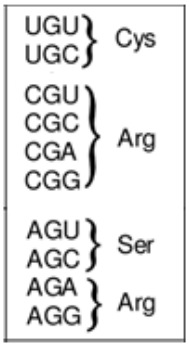

Cys = cysteine
Arg = arginine
Ser = serine

Figure 1.8: Section of an amino acid codon table for mRNA

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QUESTION 2

Read the information below. Refer to this information, as well as your own knowledge, to answer Question 2 in the question paper.

Mitochondrial DNA, genetics and ancestry

1. South Africa population genetics – the Karretjie people

Himla Soodyall is a South African geneticist involved in the study of population genetics. She has done extensive work on the genetic ancestry of different population groups in South Africa and is one of the leading population geneticists in the world.

Director of the Human Genomic Diversity and Disease Research Laboratory, National Health Laboratory Service, Wits.
Awarded Bronze Order of Mapungubwe in 2005 for 'Outstanding contributions in the fields of science'. Author and co-author of more than 90 academic publications.

Figure 2.1: Prof Himla Soodyall

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The term 'Khoisan' refers to the indigenous peoples of southern Africa who have languages that are characterised by the use of click sounds.

They were largely isolated from other human populations up until the arrival of black populations in South Africa from the north during the last 2000 years. Later, European farmers settled in southern Africa.

*'Black' in this context refers to dark-skinned people of sub-Saharan Africa who moved south to southern Africa during the last two thousand years.

The Karretjie people of the South African Great Karoo are nomads* of Khoisan ancestry who live around the town of Colesberg (Figure 2.3). The Karretjie people have an interesting history regarding their origins.

*nomad = a person with no fixed home and who moves from place to place.

Figure 2.2: Khoisan people **10**

Figure 2.3: Map showing position of Colesberg and pictures of the Karretjie people

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Proposed history of the ancestry of the Karretjie people

By the eighteenth century, white Caucasian (European) settlers had started farming in the Great Karoo of South Africa. Many of the Khoisan settled on the farms (especially women and children), whereas many of the men remained nomads. By the late 1700s, there were many descendants of these Khoisan women working on the farms. (The women often had children with the Caucasian farmers.)

Many of the descendants of these 'farm' Khoisan moved from farm to farm by means of donkey-drawn carts and later, cars, looking for work. Thus came into use the term Karretjie people – from the Afrikaans word 'Karretjie', meaning small car or cart. Many of them still move from farm to farm looking for work.

It seems therefore that their paternal ancestry is mostly Caucasian while their maternal ancestry is Khoisan.

2. mtDNA and Y chromosomes

Although most DNA is packaged in chromosomes within the nucleus, mitochondria also have a small amount of their own DNA, known as mitochondrial DNA (mtDNA). In humans, mtDNA consists of about 16 500 nucleotide base pairs, making up 37 genes and non-coding regions.

mtDNA tends to remain unchanged over many generations. This is because; unlike in nuclear DNA, **crossing over in Prophase 1** does not occur– in fact meiosis does not occur at all.

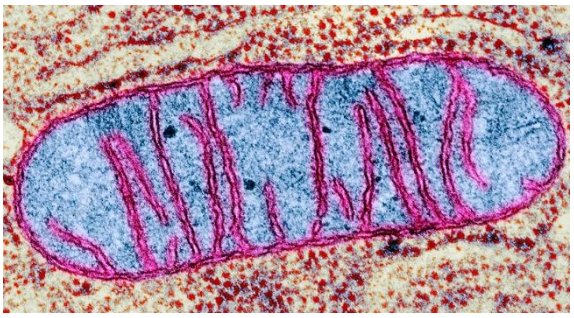

Figure 2.4: A mitochondrion in the cytoplasm

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Figure 2.5: Human X and Y chromosome

The Y chromosome is part of the nuclear DNA in a cell. Even though the Y chromosome is considered to be a partner to the X chromosome, the X and Y are not actually a homologous set. This is because there are no genes on the X that have a partner on the Y chromosome. The X and the Y also do not undergo crossing over during gamete formation. Therefore, the Y chromosome is also passed on unchanged from one generation to the next.

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3. Haplogroups

Researchers have sampled both the mtDNA and Y chromosomes from a huge number of people around the world in order to determine all of the places where their DNA differs. They have recorded the identity of different groups of alleles in each of the mtDNA and Y chromosome samples. Related groups of people tend to share the same groups of alleles. A **haplogroup** is a combination of alleles that are shared by a group of people.

By taking DNA samples, researchers can identify the mtDNA and Y chromosome haplotypes to which a person belongs. Identifying their mtDNA haplotype will give an indication of their maternal ancestry (as mtDNA is passed along the female line). Their Y chromosome haplotype (if they are male) will give an indication of their paternal ancestry (as Y chromosomes are passed from father to son).

4. Investigation

Prof Soodyall and her colleagues wanted to determine whether the proposed history of the origin of the Karretjie people could be confirmed by identifying the particular mtDNA and Y chromosome haplogroups in individuals of Karretjie people.

DNA samples were taken from 70 male Karretjie people and 70 male Khoisan individuals. The mtDNA and Y chromosome DNA were sequenced, and the researchers then identified the mtDNA and Y chromosome haplogroups to which each person belonged.

A. The following **mtDNA haplogroups** were recorded in the people studied:

Haplogroup L0d – most common among Khoisan people.
Haplogroup L1 – most common in black populations in southern Africa.
Haplogroup U – common in Europe (Caucasian populations).

Table 2.1: Table showing the percentage of the 70 people of each population group **possessing mtDNA haplogroups L0d, L1 or U.**

Population group	**Percentage of people of each population group possessing either mtDNA haplogroup L0d, L1 or U (%)**		
	Haplogroup L0d	Haplogroup L1	Haplogroup U
Khoisan	76	23,3	1,7
Karretjie	99	0,5	0,5

B. The following **Y chromosome haplogroups** were recorded in the people studied:

Haplogroup A – most commonly found in Khoisan populations.
Haplogroup E – common in black African populations.
Haplogroup I – found commonly in Caucasian populations.

Table 2.2: Table showing the percentage of 70 people of different population groups possessing Y chromosome haplogroups A, E or I.

Population group	**Percentage of people of each different population groups possessing either Y chromosome haplogroup A, E or I (%)**		
	Haplogroup A	Haplogroup E	Haplogroup I
Khoisan	85,3	14,0	0,7
Karretjie	14,5	9,2	76,3

5. Baby mix-up

In 2002, young parents from the Karretjie community, Katjie Geduld and Hendrik Veroor, gave birth to a baby at the local Colesberg hospital. Plaatjie and Meitjies Januarie also had a baby at the same hospital. However, it appears that the two babies of the two couples – both boys – may have been mixed up at the hospital.

A sample of each parent was taken, along with samples from the babies. PCR was conducted and DNA fingerprinting carried out in order to produce genetic profiles for each person. These are shown in Figure 2.6.

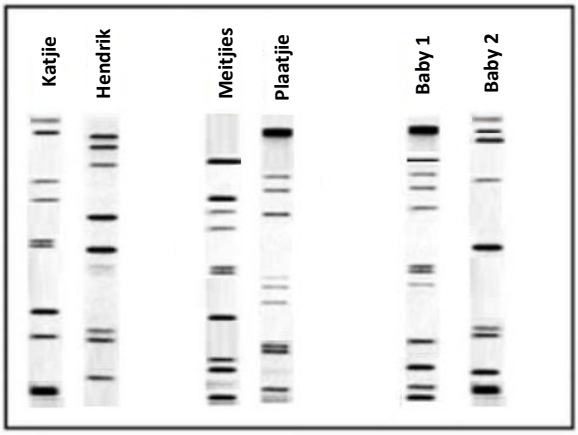

Figure 2.6: Six gels showing the DNA fingerprinting results on two sets of parents and two babies

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SECTION B

QUESTION 3

Read the information below. Use this information, as well as your own knowledge, to answer Question 3 in the question paper.

SOURCE A The changing role of women

Can you name a female geneticist?

Who are the greatest geneticists of all time? Chances are Gregor Mendel, Maurice Wilkins, James Watson, Francis Crick or other big names come to mind – and for good reason. Those scientists made remarkable discoveries and changed how we understand the world.

Genetics is often considered a male-dominated field. However, female geneticists have made significant contributions and discoveries. In the past, women have consistently been underrepresented in the field of genetics and in many cases their important discoveries were minimised or neglected.

It was only recently that female geneticists have come out of the shadows of history. Since the mid-twentieth century, new generations of women geneticists have contributed to research, but do these women receive appropriate recognition for their research? **16**

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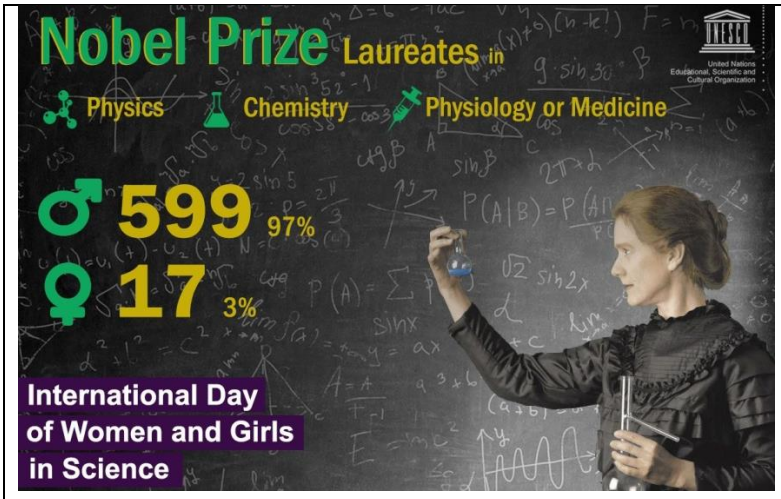

The United Nations General Assembly declared 11 February as the International Day of Women and Girls in Science in 2015

The Israeli Government proclaimed 2002–2003 the year of advancing women in science and Technology.

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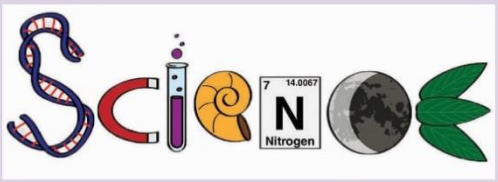
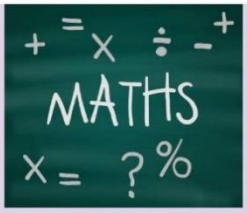
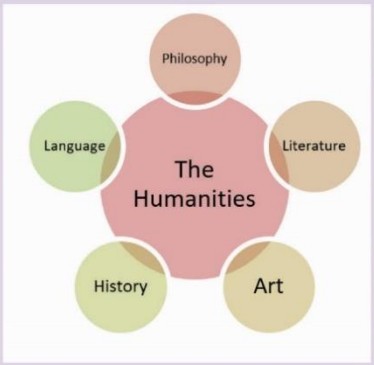
Historical reasons for fewer women in genetics …





The myth of the maths brain is one of the most self-destructive ideas in education – there are no differences between men and women in maths scores, yet people still talk about it.

Teachers and parents often underestimate girls' maths abilities.
Teachers often have maths anxiety they pass onto girls, and they are often stricter with girls when marking them, and assume girls need to work harder to achieve the same level as boys.



Social attitudes encourage males to go into difficult fields … females are encouraged to study the humanities rather than the sciences.

Girls have seen fewer role models to inspire their interest in science.

Political and religious beliefs have sometimes resulted in gender inequalities.

Women often battle to balance work and family commitments.

In 2015, the Nobel Prize winner and biochemist Tim Hunt stated that laboratories should be single sex, because 'girls … fall in love with you, and when you criticise them, they cry'.

… have any of these facts changed over time?

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SOURCE B Statistics

Student enrolment by gender in the faculty of genetics at the University of the Witwatersrand in 2020.

Gender	Number enrolled
Female	2 569
Gender neutral	2
Male	2 793

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'I think having all these women around makes it more fun for the men but they're probably less effective.'

James Watson, 2001

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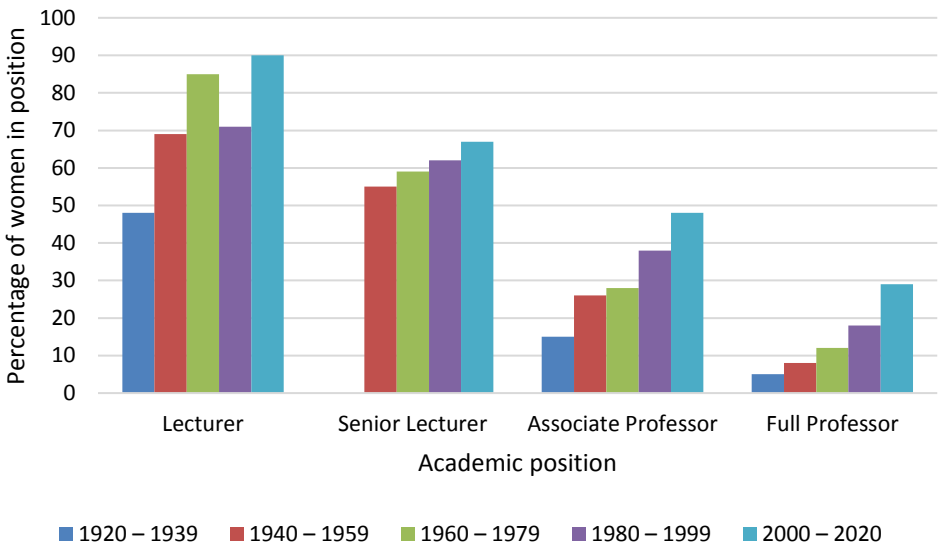

Percentage of women in various academic positions (from lecturer to full professor) in the faculties of genetics at top academic institutions in the 1920s to 2020.

Explanation of different academic positions in order of status:

- **Lecturer**

This is the lowest level required for a permanent academic position. Lecturers are responsible both for teaching and conducting research.

- **Senior Lecturer**

This is a teaching position. They are appointed for a longer term than lecturers.

- **Associate Professor**

A professor without a permanent position.

- **Full Professor**

The most senior academic position – they are expected to conduct research and to guide students in their research.

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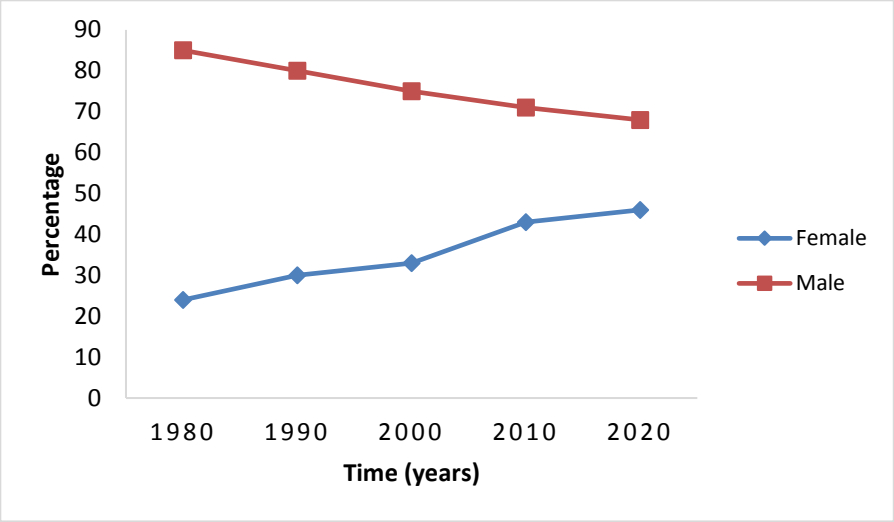

Percentage of men and women involved in genetics research work between 1980 and 2020

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SOURCE C Society and the Gender Wage War

Researchers at Yale University published a study showing that genetics professors view a male scientist more favourably than a woman with the same qualifications. Presented with identical summaries of the accomplishments of two imaginary applicants, professors at six universities were significantly more willing to offer the man a job. If they did hire the woman, they set her salary nearly R60 000 lower than the man's. Surprisingly, female professors were as biased as their male counterparts.

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People protest difference in pay between men and women at a 2018 rally for International Women's Day in New York City.

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The gender gap in salary disappeared among researchers who have reached very high positions – male and female researchers both reported an average salary of R500 000 in Canada.

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SOURCE D FAMOUS MEN AND WOMEN IN SCIENCE

Nettie Stevens – discovery of sex chromosomes. Stevens was not recognised immediately after her discovery.

Marie Maynard Daly – biochemist. Studied chemistry of histones and protein synthesis. First black American woman to earn a PhD in chemistry in the USA.

Esther Lederberg – studied gene regulation and genetic recombination.

Barbara McClintock – **Nobel Prize** in Physiology or Medicine. Identified transposable elements also known as 'jumping' genes.

Margaret Oakley Dayhoof – application of mathematics and computational techniques to sequenceing of proteins and nucleic acids.

Nadia Sakati – established first genetics department in Saudi Arabia. Discovered rare genetic syndrome that was named after her.

Yue Wan – RNA functional structures and roles in the regulation of cellular processes.

Chanchao Lorthongpanich – research on reverting differentiated adult cells from patients to stem cells.

Narry Kim – molecular cell biologist, study of RNA biology.

Professor Sarah Gilbert – woman behind the Oxford/Astra-Zeneca vaccine.

1900 1925 1950 1975 2000 2020

Charlotte Auerbach – German Jewish geneticist-demonstrated that mustard gas could induce mutations.

Martha Chase – American gen-eticist, helped confirm that DNA, rather than proteins, is the genetic material of life. **Husband awar-ded Nobel Prize in Physiology or Medicine yet Chase excluded.**

Dorothy Crowfoot Hodgkin – English chemist whose determination led to the discovery of the structure of penicillin and vitamin B12.

Mary Frances Lyon – discovery of chromosome inactivation. Not awarded the Nobel Prize, which many scientists claim she deserved.

Elizabeth Blackburn – best known for her work on telomeres and co-discovery of telomerase.

Samia Aly Temtamy – one of the first Arab females to earn a doctorate in genetics, found first human genetics department in the National Research Centre.

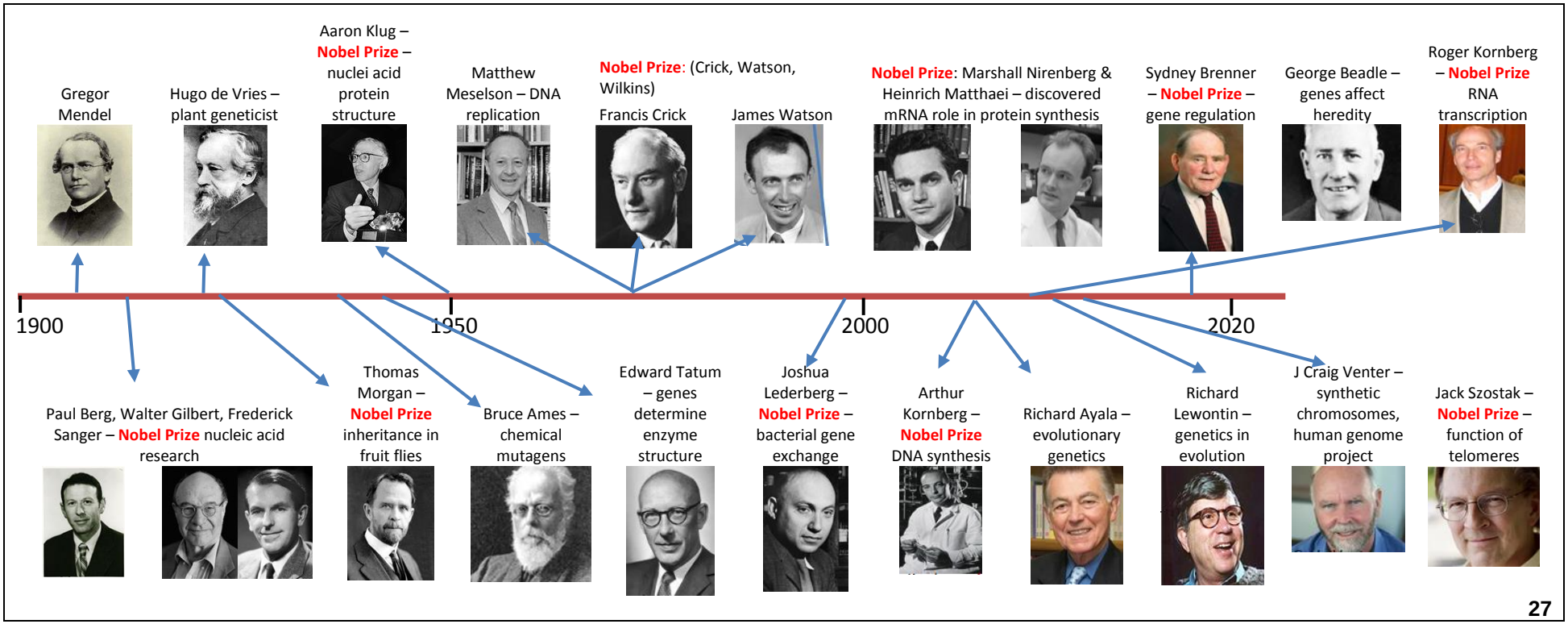
Rana Dajani – stem cell research ethics laws, expert on the genetics of Circassia and Chechen population in Jordan.

Adeyinka Falusi – discovered genetic markers for sickle cell anaemia.

Dr Özlem Türeci – the co-founder of BioNTech – company with 54% female work staff.

Dr Kizzmekia Corbertt – role in the development of Moderna's COVID-19 vaccine.

Christiane Nusslein Volhard – looking at genes involved in the fly body plan and segmen-tation, contrib-uted to our understanding of human embryo development.



SOURCE E Awards and Recognition

Scholarships for women in genetics

- **AOE National Foundation Scholarships** – USA.
- **British Council Scholarships for women in Genetics** – British but awarded for study anywhere in the world.
- **L`Oréal India for Young Women in Science Scholarship** – India.
- **Science Ambassador Scholarship** – for study anywhere in the world.
- **Israeli Science Ambassador Scholarship** – any woman from around the world studying in Israel.

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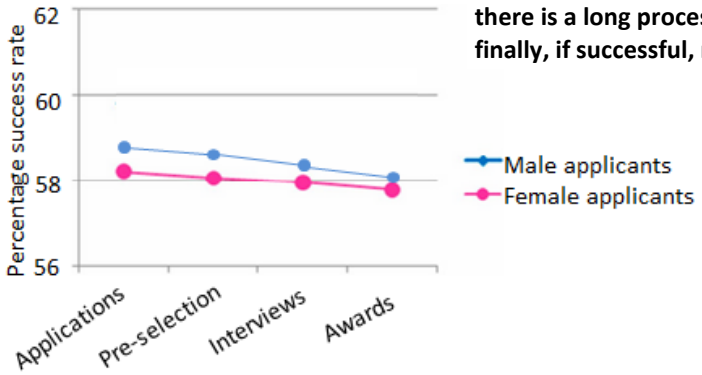
Research grants (money for research) in 2020

When researchers apply for money (grants) for research, there is a long process of pre-selection, interviews and finally, if successful, money is awarded.



Percent success rate for researchers applying for genetics grants (money for genetics experiments)

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Awards

Barbara McClintock – American Academy of Arts and Science (1959); Medal of Science (1970).

YueWan – Genome Web Young Investigator Award (2015).

Narry Kim – Gwanak Grand Prize (2008); Scientist of the Year Award (2013).

Adeyinka Falusi –Woman of Science Award (2009).

Rana Dajani – Fulbright Occasional Lecture Award Spring (2013); Arab Science and Technology Foundation (2019).

Samia Amy Temtamy – Scientific Excellence Award for the National Research Centre (1987)

Mary Lyon – Fellow of the Royal Society (1973); US National Academy of Sciences (1979); Wolf Prize in Medicine (1997).

Charlotte Auerbach – Royal Society of Edinburgh (1947); Fellow of the Royal Society (1957); National Academy of Sciences (1970); Darwin Medal (1976).

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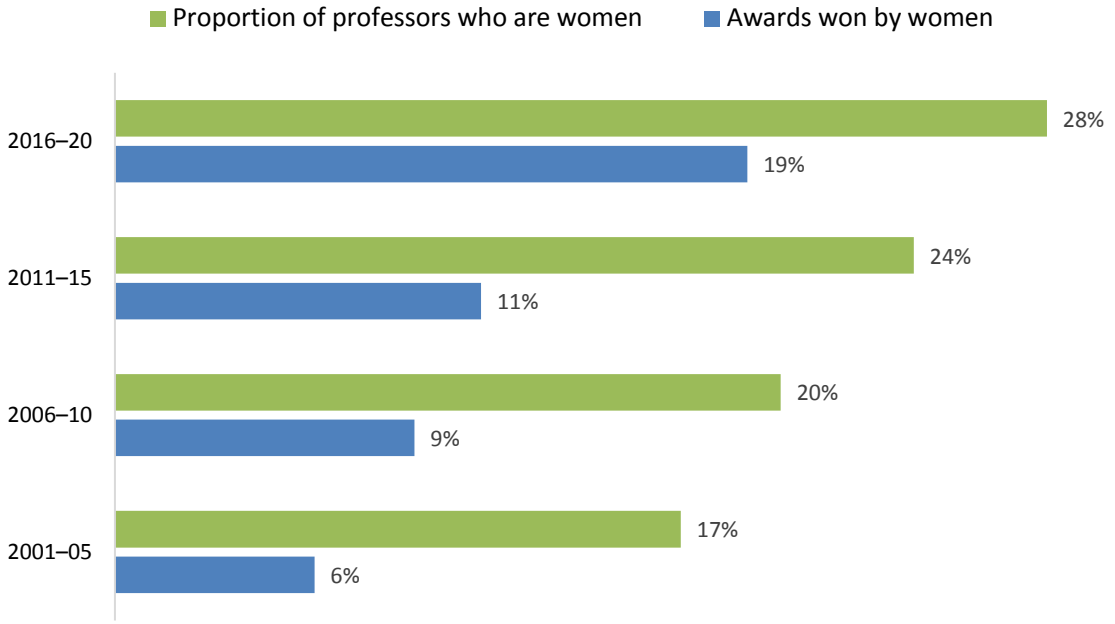
CLOSING THE GAP

Although the proportion of professors who are women has increased in recent decades, the percentage of top research prizes going to women still lags behind.



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Mattel™ company released six Barbie dolls to honour women in science and their contributions in the fight against COVID-19

Dr Audrey Sue Cruz
United States

Dr Jaqueline Goes de Jesus
Brazil

Dr Kirby White
Australia

Professor Sarah Gilbert
United Kingdom

Dr Chika Stacy Oriuwa
Canada

Amy O'Sullivan, RN
United States

32

Reference List

QUESTION 1

1 [Adapted: <https://www.findatopdoc.com>]

2 [<https://www.cdriadvlkn.org>] [<https://www.d3i71xaburhd42.cloudfront.net>] [<https://www.ncbi.nlm.nih.gov.za>]

3 [<https://www.cdn.thculturetrip.com>]

4 Examiner's data

5 [Credit: Alexander Heinel/Picture Alliance/DPA]

6 [Adapted: <https://www.cambridge.org>]

7 [Adapted: <https://www.researchgate.net>]

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[Adapted: <https://www.rarediseasenetwork.org>]
[Adapted: <https://www.sciencedirect.com>]

QUESTION 2

9 [<https://www.facebook.com>]

10 [<https:www.//qph.fs.quoracdn.net>]

11 [Adapted: <https://www.researchgate.net>] [<https://www.i.pinimg.com>] [<https://www.groundup.org.za>]

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13 [Adapted: <https://scx1.b-cdn.net>]

14 [Adapted: <https://media.cheggcdn.com>]

15 [Adapted: Achilli, A.et al. 2004. *American Journal of Human Genetics* 75(5): 910 – 918]
[Adapted: Barbieri, C. et al. 2013. *American Journal of Human Genetics* 92(2): 285 – 292]
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[Adapted: Pakendorf, B. & Stoneking, M. 2021. The genomic prehistory of peoples speaking Khoisan languages. *Human Molecular Genetics* 30(R1): R49 – R55]
[Adapted: Schlebusch, C. M. et al. 2011. Different contributions of ancient mitochondrial and Y-chromosomal lineages in 'Karretjie people' of the Great Karoo in South Africa. *Journal of Human Genetics* 56: 623 – 630]
[Adapted: Tishkoff, S. A. et al. 2007. History of click-speaking populations of Africa inferred from mtDNA and Y-chromosome genetic variation. *Molecular Biology and Evolution* 24(10): 2180 – 2195]

QUESTION 3

16 [Adapted: <https:///www.discovermagazine.com>]

17 [Adapted: <https://www.i.cbc.ca>]

18 [Adapted: Messer-Yaron, H & Kahanovich, S.2003. Women and Science in Israel: A Status Report. Council for the Advancement of Women in Science and Technology]
[United Nations General Assembly 2015]

19 [Adapted: Why So Few? Women in Science, Technology, Engineering & Mathematics. Demographics and Statistics in the US. AAUW]
[AAUW (American Association of University Women) is a charitable association supporting women in university]

20 [Adapted: <https://www.wits.ac.za>]

21 [<https://www.smithsonianmag.com>]

22 [Adapted: <https://www.academicpositions.com>]
[Adapted: Huang, J., Gates, A. J., Sinatra, R. & Barabási, A. L. 2020. Historical comparison of gender inequality in scientific careers across countries and disciplines. *PNAS* 117(9): 4609-4616]
[PNAS = Proceedings of the National Academy of Sciences – this is a journal publishing information from all spheres of science]

23 [Adapted: US Census Bureau 1970, 1980, 1990 and 2000 Censuses. 2010 and 2019 American Community Surveys]

24 [Boekhout, H., van der Weijden, I., and Waltman, L. (2021). Gender Differences in Scientific Careers: A Large-Scale Bibliometric Analysis. In press]
['In Press' means that the article is waiting to be published in a journal. Peer review not completed]
[Adapted: Woolston C. 2021. Pay gap widens between female and male scientists in North America. *Nature* 590 (677)]
[*Nature* is a top-rated science journal covering all aspects of the sciences]

25 [Credit: Spencer Platt/Getty]

26 [Adapted: Lerchmueller, M. J. & Sorenson, O. 2017. Research: Junior female scientists aren't getting the credit they deserve. *Harvard Business Review*]
[*The Harvard Business Review is a magazine published by Harvard University dealing with business management*]

27 [Adapted: Elbardisy, H. & Abedalthagafi, M. 2021. History and challenges of women in genetics: A focus on non-western women. *Front. Genet.*]
['Frontiers in Genetics' is a journal publishing research into genetics]
[Adapted: <https://www.genetics.org.uk>]
[*The Genetics Society is a British organisation dealing with genetics. It is open to anyone with an interest in genetics*]

28 [Adapted: <https://www.topuniversities.com>]
[This is a website providing information about universities around the world]

29 [Adapted: <https://www.i.cbc.ca>]
[CBC = Canadian Broadcasting Corporation – the Canadian national TV and radio broadcaster]
Van der Lee, R., Ellemers, N. 2015. Gender contributes to personal research funding success in the Netherlands. *Psychological and Cognitive Sciences* 112(40)]
[Psychological and Cognitive Sciences is a journal publishing articles on psychology]

30 [Watson, C. 2021. 'Women less likely to win major research awards'. *Nature*]

31 [Adapted: Meho, M. L. 2021. The gender gap in highly prestigious international research awards, 2001 – 2020. *Quantitative Science Studies* 2(3): 976–989]
[*Quantitative Science Studies is a journal that publishes research into studies that provide insight into science policy*]

32 [Adapted: <https://www.aauw.org>]