NATIONAL SENIOR CERTIFICATE EXAMINATION
NOVEMBER 2018

LIFE SCIENCES: PAPER II

SOURCE MATERIAL BOOKLET FOR QUESTIONS 1, 2 AND 3

SECTION A

The Source Material on pages ii–iv relate to Question 1.

QUESTION 1

Read the information and answer the questions which appear on page 2 in the question paper.

The blue people of Troublesome Creek

Six generations after Martin Fugate settled on the banks of eastern Kentucky's Troublesome Creek (river) with his redheaded American bride, his great-great-great great grandson Benjy Stacy was born in a modern hospital not far from where the river still runs. What was unusual about Benjy was that he was born with a dark blue skin.

Doctors were so astonished by the colour of Benjy's skin that they raced him by ambulance from the maternity ward to a medical clinic in Lexington. Two days of tests produced no explanation for the condition.

A blood transfusion was being prepared when Benjy's grandmother spoke up. "Have you ever heard of the blue Fugates of Troublesome Creek?" she asked the doctors. "My grandmother Luna on my dad's side was a blue Fugate. It was real bad in her," the boy's father explained.

After ruling out heart and lung diseases, the doctors suspected **methaemoglobinemia** (MHG), a rare hereditary blood disorder that results from excess levels of methaemoglobin (met-Hb) in the blood.

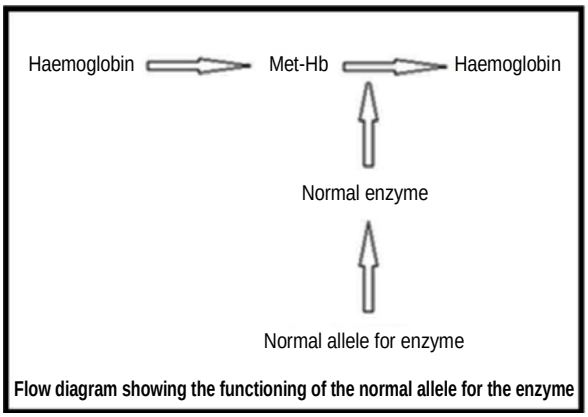
Haemoglobin is the protein which carries oxygen in red blood cells (haemoglobin is responsible for giving the red colour to red blood cells).

Met-Hb is a non-functional form of haemoglobin – it does not carry oxygen. The more met-Hb in the blood, the less oxygen the blood can carry. Met-Hb is a bluish-coloured molecule and instead of being red in colour, the arterial blood of MHG patients is blue.

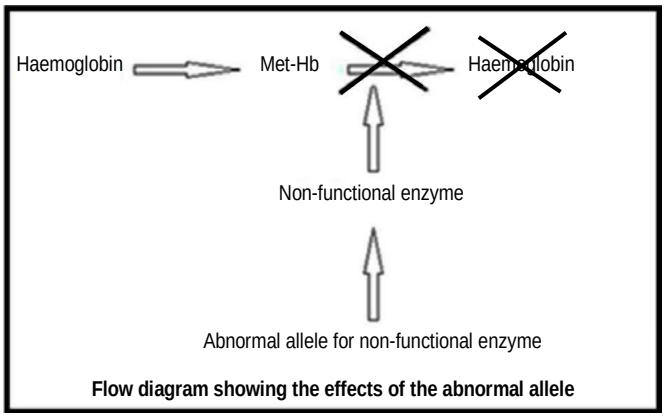
In normal people haemoglobin is naturally converted to met-Hb at a very slow rate. If this conversion continued, all the body's haemoglobin would eventually become useless. An enzyme is responsible for continually changing met-Hb back to normal haemoglobin.

The enzyme is coded for by a gene on the short arm of human chromosome 22. The expression of this gene follows simple Mendelian genetics laws. The normal allele of this gene codes for the functional enzyme.

However, there is another allele for this gene which codes for a different non-functional version of the enzyme. This enzyme will not work to turn met-Hb back to haemoglobin. The condition MHG results only when a person has inherited two of these abnormal alleles.



Flow diagram showing the functioning of the normal allele for the enzyme



Flow diagram showing the effects of the abnormal allele

The mutation that gives rise to the abnormal allele is at nucleotide number 875 of the gene, where cytosine has been replaced by thymine. This results in the amino acid aspartic acid being coded for, instead of glycine, during RNA translation of this allele.

The Fugate clan kept multiplying over the years since Martin Fugate settled in Kentucky. Fugates married other Fugates. Sometimes they married first cousins. And they married the people who lived closest to them, the Combses, Smiths, Ritchies, and Stacys. All lived in isolation from the world, bunched in log cabins up and down the valleys, and so it was only natural that a boy married the girl next door, even if she had the same last name. By the time a young *haematologist from the University of Kentucky came down to Troublesome Creek in the 1960s to cure the blue people, Martin Fugate's descendants had multiplied their alleles for the characteristic all over the area.

Two families outside their log cabins in Troublesome Creek, 1920

[Adapted: <https://encrypted-tbn0.gstatic.com/images>; <https://i.pinimg.com>]

Most people with this blood disorder lived to their 80s and 90s without serious illness associated with the condition. For some, though, there was a pain not seen in lab tests. That was the pain of being blue in a world that is mostly shades of white to black.

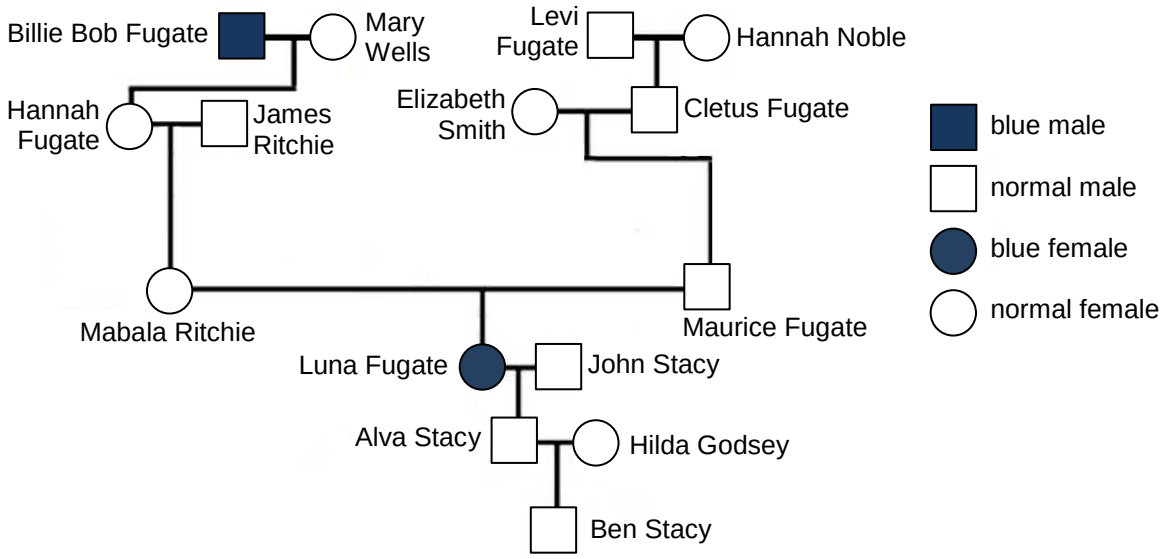
As coal mining and the railroads brought progress to Kentucky, the Fugates started moving out of their communities and marrying other people. The strain of inherited blue began to disappear as the allele spread to families where it was unlikely to be paired with the same allele.

Benjy Stacy's blue colouration eventually disappeared. Doctors predicted that even though he only had one allele for the condition, he was blue as a baby, as babies tend to produce less enzyme than adults.

* haematologist: a person who treats blood disorders

Pedigree chart of the Fugate family

(Billie Bob Fugate and Levi Fugate are cousins)



[Adapted from: *The Fugate Family Newsletter*]

[Adapted: <https://ghr.nlm.nih.gov; www.indiana.edu/; NCBI 1000 Genomes Browser>]

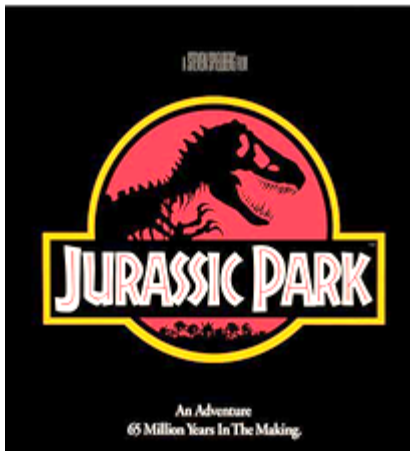
The Source Material on pages iv–vii relate to Question 2.

QUESTION 2

Read the information below and answer the questions which appear on pages 3–4 in the question paper.

Jurassic Park (1995) is a film directed by Steven Spielberg based on a book by Michael Crichton. The plot centres on a disastrous attempt to create a theme park of cloned dinosaurs that escape confinement and proceed to terrorise and kill the human characters.

In the film, a biotechnology company called *InGen* discovered a method of cloning dinosaurs. They found mosquitoes that had been trapped in amber (fossilised tree resin) 66 million years ago. These mosquitoes still had blood in their guts that had been sucked from dinosaurs. Geneticists extracted the DNA from the blood cells and then used the DNA to clone dinosaurs.



[Adapted: <www.movieposter.com/posters/>]

Mosquito trapped in amber

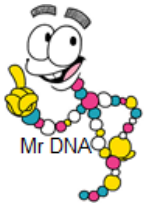
[Adapted: <i.pinimg.com>]

In the movie, the scientists used a variety of cloning and recombinant DNA technology techniques. The DNA that was extracted from the mosquitoes was obviously partially degraded (it had missing sections) as it was so old. However, the scientists of "Jurassic Park" solved the problem of the missing DNA by filling in the missing pieces with frog DNA.

[Adapted: <https://science.howstuffworks.com>]

Extract from the Jurassic Park film script

Visitors to Jurassic Park are taken on a tour and are watching an animation showing how the dinosaurs were cloned:



[Adapted: <iho.redbubble.net>]

Mr DNA is a cartoon character, a double-helix strand of recombinant DNA. Mr DNA jumps down onto the screen.

HAMMOND (the CEO of Jurassic Park): Well! Mr DNA! Where'd you come from?

Mr DNA: From your blood! Just one drop of your blood contains billions of **strands of DNA**, the **building blocks of life**!

A DNA strand like me is a blueprint for building a living thing! And sometimes animals that went extinct millions of years ago, like dinosaurs, left their blueprints behind for us to find! We just had to know where to look!

….

One SCIENTIST moves a complicated drill apparatus next to the amber with a fossilised mosquito inside and bores into the side of it. Mr DNA escapes through the drill hole as the scientist moves the amber onto a microscope and peers through the eyepiece.

Mr DNA: A full DNA strand contains three billion genetic codes! If we looked at screens like these once a second for eight hours a day, it'd take two years to look at the entire strand! It's that long! And since it's so old, it's full of holes! That's where our geneticists take over! … We use the DNA of a frog to fill in the missing pieces in the dinosaur DNA.

*We see an actual DNA strand, except it has a big hole in the centre, where the vital information is missing. Mr DNA appears, carrying a **bunch of DNA letters** in one hand. He puts it in the gap.*

[Adapted: <dailyscript.com/scripts/jurassicpark>]

Should we bring extinct species back from the dead?

Earth is in the middle of its sixth mass extinction: somewhere between 30 and 159 species disappear every day, thanks largely to humans.

For decades the notion of "de-extinction" ("bringing back" species from extinction) was not possible, but new advances in genetic engineering, especially the CRISPR-Cas9 revolution, have researchers believing that it's time to start thinking seriously about which animals we might be able to bring back.

The Pyrenean ibex or bucardo (*Capra pyrenaica pyrenaica*) was a large, wild goat, with long, gently curved horns. For thousands of years it lived high in the Pyrenees, the mountain range that divides France from Spain, where it clambered along cliffs, nibbling on leaves and stems.

They were once common but extensive hunting reduced their numbers to 100 individuals in 1899. They were eventually declared protected in 1973, but by 1981 just 30 remained. In 1989 Spanish scientists did a survey and concluded that there were only 12 individuals left. The last bucardo, a 13-year-old female known as Celia, was found dead in January 2000 by park rangers.

Dr Jose Folch, from the Centre of Agro-Nutrition and Research in Aragon, Spain, had however captured Celia the previous year and had taken a tissue sample from her ear.

Image of a bucardo

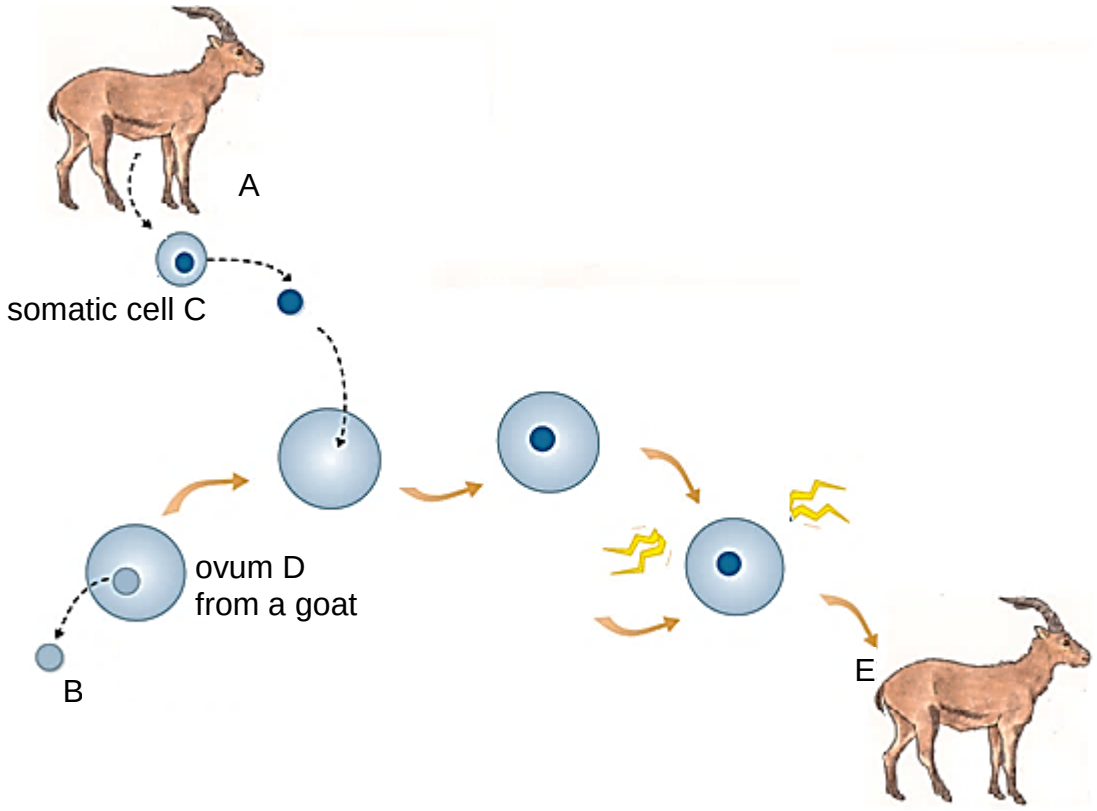
[Adapted: <shared.com/content/images>]

Using techniques similar to those used to clone Dolly the sheep, known as somatic cell nuclear transfer (SCNT), the researchers were able to transplant DNA from the ear tissue into ova taken from domestic goats to create 439 embryos, of which 57 were implanted into surrogate females.

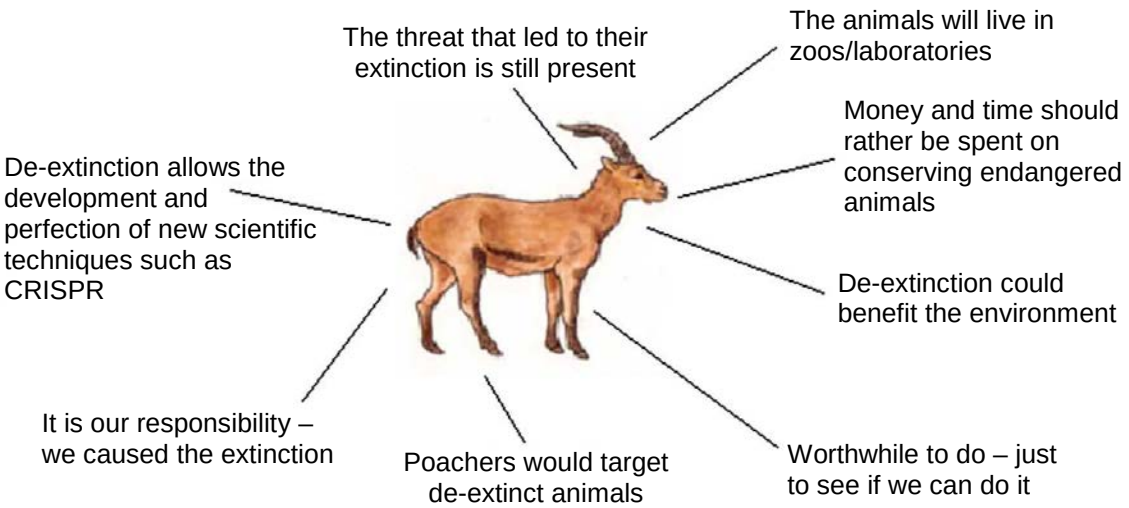
Just seven of the embryos resulted in pregnancies and only one of the goats finally gave birth to a female bucardo. It died seven minutes later due to breathing difficulties, perhaps due to flaws in the DNA used to create the clone. Despite the highly inefficient cloning process and death of the cloned bucardo, many scientists believe similar approaches may be the only way to save critically endangered species from disappearing.

[Adapted: <nationalgeographic.com/magazine>]

Somatic cell nuclear transfer to produce a "de-extinct" bucardo



Reasons for and against producing "de-extinct" animals



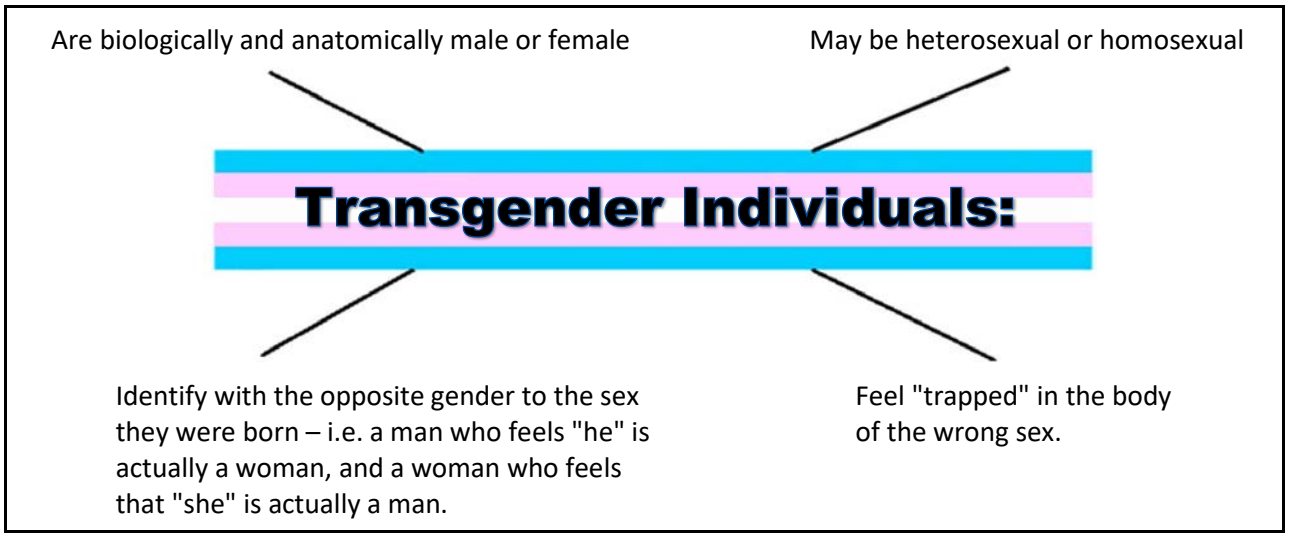
[Adapted: <evolution.berkeley.edu>]

The Source Material on pages viii–xiii relate to the essay on page 5 of the question paper.

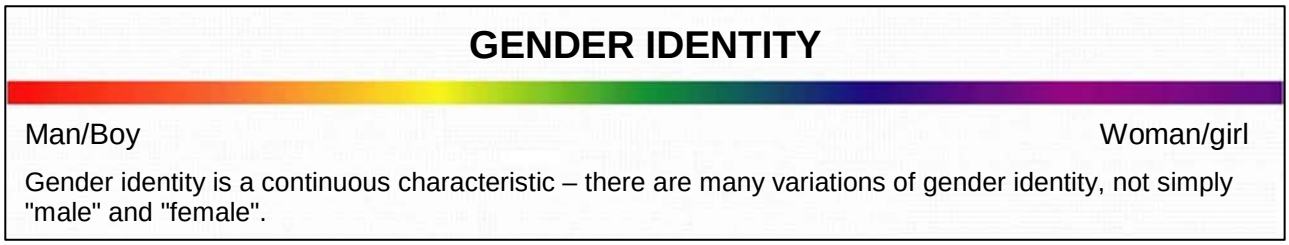
QUESTION 3

SOURCE A

What does "transgender" mean?



[Adapted: Forsyth, C. J. & Copes, H. 2014]



[Adapted: <scalar.usc.edu>]

Possible causes of being transgender

Genetic	Environmental
X or Y chromosome differences	Hormone under- or over-secretion in the womb
Genetic changes	Differences in brain structure
	Influences of society
	Exposure to hormones in the environment
	Psychological – influenced by human bonding and family relationships

[Adapted: <https://www.news-medical.net>]

Two quotes from psychiatrists

Research increasingly points to our brains (our way of thinking) as playing a key role in how we each experience our gender.

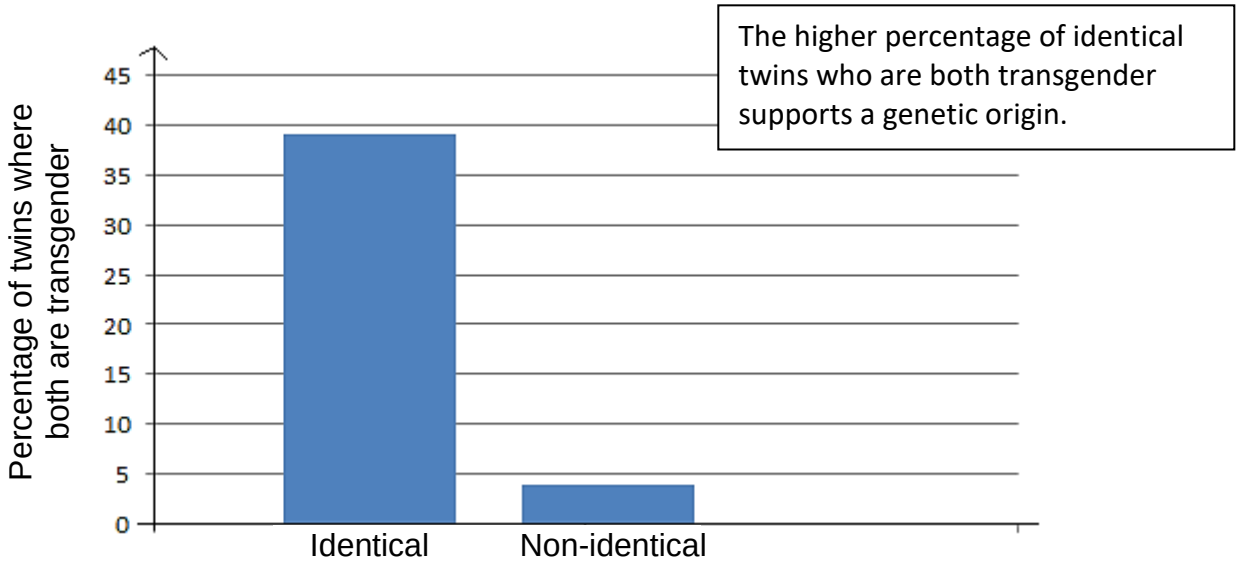
[Adapted: Dr Felix Torres]

By age four, most children have a stable sense of their gender identity. Individuals do not choose their gender, nor can they be made to change it.

[Adapted: American Academy of Paediatrics]

SOURCE B

Graph showing the percentage of identical versus non-identical twins where both are transgender.



[Adapted: Heylens G., De Cuypere G., Zucker K. J., Schelfaut C., Elaut E., Van den Bossche H, De Baere E, & G. T'Sjoen. 2012. Gender identity disorder in twins: A review of the case report literature. *Journal of Sex Medicine* 9: 751–757]

HOWEVER

Identical twins contain 100 percent of the same DNA. So surely we should expect both twins to identify as transgender close to 100 percent of the time.

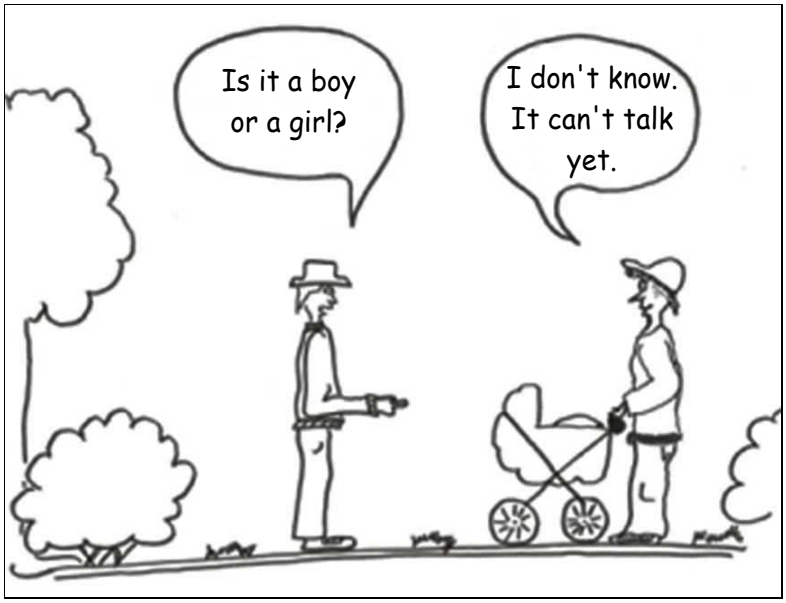
[Adapted: Dr Michelle Cretella America College of Paediatricians]

SOURCE C

Psychiatrist Stephen Stathis, who runs the gender clinic at Brisbane's Lady Cilento Children's Hospital and is responsible for diagnosing transgender individuals, reports that many youth are "trying out being transgender" in order to stand out. Apparently, declaring oneself "transgender" is trendy.

Most patients, however, are simply going through a common phase of adolescent life. By the time boys and girls reach puberty, most identify as their biological sex.

[Adapted: <lifesitenews.com>]



[Adapted: <whatyoucandowithanthropology.wordpress.com>]

[Adapted: Dr Michelle Cretella. America College of Paediatricians]

SOURCE D

Environmental conditions, such as socialisation, seem to contribute to transgenderism. Parents, family and friends, media role models, cultural factors, learning and conditioning determine a child's sense of gender.

[Adapted: <mindconditions.com>]

Famous transgender media personalities

Caitlyn Jenner – olympic gold medal winner and television star.
Carmen Carrera – an actress from "Cake Boss" and "Real Housewives of New York City".
Laverne Cox – an actress and producer, best known from "Orange Is the New Black".
Chaz Bono – advocate, writer, musician – the child of American entertainers Sonny and Cher.
Caroline "Tula" Cossey – an English model and Bond girl.
Larry Wachowski – director.
Alexandra Billings – an actress who appeared on "Grey's Anatomy", "ER", and "Eli Stone".

Carmen Carrera


Laverne Cox


Caroline Cossey


Alexandra Billings

[Adapted: <www.ranker.com>]

Interview with a transgender person

At what age did you realise you were the wrong gender?

"I have never been the wrong gender – I have always been a girl. I was simply in the wrong body. Gender is not the body we are born into. It is at the core of who we are on the inside.

I knew that there was something wrong when I was three or four years old."

How has your life changed since you made the choice to live as a woman?

"Before I decided to live as a woman, I felt as though I was leading a double life.

There was a disconnection between who I felt I was and the person I had this intense internal need to be.

Expecting someone to be something they are not is cruel."

What do you want others to understand about transgender men and women?

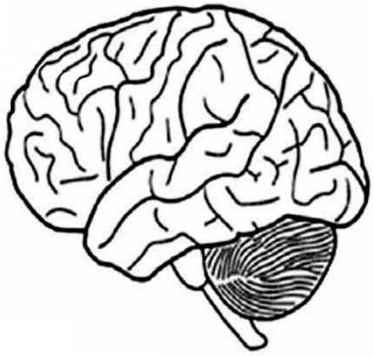
"We're just like everyone else."

[Adapted: <www.chicagonow.com>]

SOURCE E

Summary of several studies revealing brain differences in transgender individuals

Exposure to high levels of testosterone in the womb influences the development of gender identity and sex-related personality characteristics.



Female transgenders have a white matter pattern in the brain that is similar to that of non-transgender males – this is determined by environmental influences.

Studies reveal a correlation between gender identity and the size of a brain region that can be affected by testosterone levels.

Some of these studies are based on just 12 samples.

Factors such as long-term exposure to stress, hormones, drugs and even diet can also alter brain structure.

[Adapted: Erickson-Schroth, L. 2013. Update on the Biology of Transgender Identity. *Journal of Gay & Lesbian Mental Health*. 17: 150–74
Gu J, Kanai R. 2014. What contributes to individual differences in brain structure? *Frontiers in Human Neurosciences* 8: 262.
Hines, M. 2011. Gender Development and the Human Brain. *Annual Review of Neuroscience* 34: 69–88
Swaab, D.F. 2004. Sexual differentiation of the human brain: relevance for gender identity transsexualism and sexual orientation. *Gynaecological Endocrinology*. 19 (6): 301–12
Zhou, J. N., Hofman, M. A, Gooren, L. J. G., & D. F. Swaab. 1995. A sex difference in the human brain and its relation to transsexuality. *Nature*. 378 (6552): 68–70]

SOURCE F

Born this way? Researchers explore the science of gender identity

Five research institutions in Europe and the United States are looking to the genome for clues about whether transgender people are born that way. Two decades of brain research have provided hints of a genetic origin to being transgender.

Now scientists have embarked on what they call the largest-ever study of its kind, searching for a genetic component to explain why people assigned one gender at birth so persistently identify as the other, often from very early childhood, indicating that transgender is determined genetically. Researchers extracted DNA from the blood samples of 10 000 people, 3 000 of them transgender and the rest non-transgender and are testing for genetic differences. A small number of genes have been studied so far which show promising results, including the CYP17A1 and CYP19A1 genes. Unique alleles of the *RYR3* gene have also been found in thirteen transgenic individuals.

[Adapted: <reuters.com>]

Transsexual study reveals genetic link

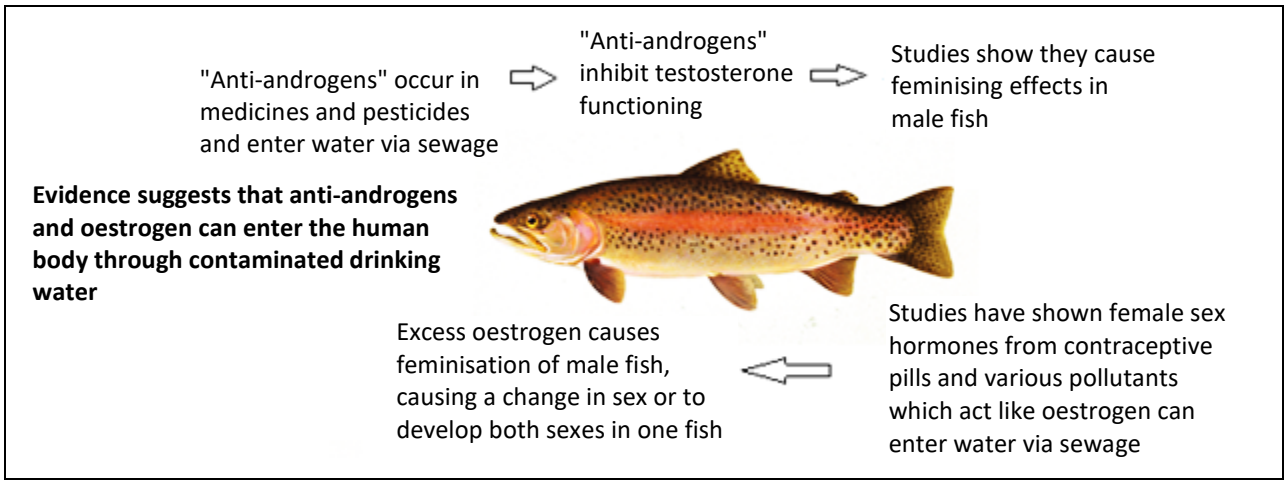
The discovery of a genetic variation in male transgenders adds weight to the view that transgenderism has a biological basis. Male transgenders are more likely than non-transgender males to have different versions of receptor genes for testosterone. This genetic variant might reduce testosterone action and "under-masculinise" or feminise the brain during foetal development. Studies show that this version of the receptor gene works less efficiently at communicating the testosterone message to cells. This contributes to the female gender identity of male to female transgenders.

A variant of the ER beta gene in female transgenders results in faulty receptors for oestrogen.

[Adapted: Hare, L., Bernard, P., Sanchez, F. J. P. Baird, N., Vilain, E., Kennedy, T., V. R. Harley. 2009. Androgen Receptor Repeat Length Polymorphism associated with Male to Female Transsexualism. *Biol. Psych.* 65(1): 93–96]

SOURCE G

Environmental influences



[Adapted: Sohoni, P. & J. P. Sumpter. 1998. Several Environmental Oestrogens are also Anti-androgens. *Journal of Endocrinology* 158(3): 327–3390]

SOURCE H

Medical influences and conditions

Intersex – babies are born with genitalia of both sexes. Operations are performed to remove one set of genitalia to "normalise" the baby. Removing the wrong genitalia at birth could result in transgenderism.	**Congenital Adrenal Hyperplasia (CAH)** – high levels of male hormones produced in a female foetus as a result of certain genes, makes them have genitalia that look like a male. They are then brought up as males, but are actually female.	**Taking medication containing hormones during pregnancy** can cause children to be born with different under-developed genitalia or with a brain that has an opposite gender identity to their biological identity.
	Androgen Insensitivity Syndrome (AIH) – cells have the inability to respond to male hormones – genetically male but develop as females.	

[Adapted: <nhs.uk/conditions/gender-dysphoria>]