



# **Genetics & Behaviour**

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Genes & Behaviour: One Gene & its Effect on Behaviour

# Genes & Behaviour: One gene & its Effect on Behaviour

## What are genes?

- A gene is the basic unit of heredity in a living organism, e.g. humans, animals, plants
- All living organisms depend on genes as they provide the basic building blocks which determine the nature and function of that organism
- Genes hold the information to build and maintain their cells and pass genetic traits to offspring
- The genotype of any organism refers to all the genetic information held within the organism's DNA
- The **human genome** consists of around 25,000 genes
- A gene represents a segment of DNA and is located at a specific point on an organism's chromosome
- Human beings have 23 pairs of chromosomes (except for conditions such as **Down's Syndrome** which results in a person being born with an extra chromosome)
- Alleles are different version of the same gene e.g. there is an allele for blue eyes and an allele for brown
  eyes
- Each allele in a pair is inherited directly from each parent
- The **phenotype** (which is the expression of the genotype) of an organism refers to the observable physical and behavioural traits e.g. hair colour, physique, abilities e.g. in music or sport, hormone levels etc.
- A phenotype is also influenced by environmental factors such as geographical location, upbringing, education etc. which explains why identical twins (monozygotic) do not have identical personalities or abilities
- To date there is no evidence to show that particular abilities or traits are governed by one single gene, rather it seems that people inherit specific vulnerabilities or sensitivities which predispose them to certain conditions, e.g. depression



## What is the MAOA gene?

- Although no one gene could be said to code for one behaviour it has been established that specific genes may contribute to specific behaviours or conditions
- The MAOA gene regulates the levels and activity of MAOA in humans
- MAOA, or monoamine oxidase A to give it the full title, is an enzyme that has been linked to specific neurotransmitters such as serotonin and dopamine
- MAOA breaks down these neurotransmitters which can result in irregular, disrupted or low levels of dopamine or serotonin which in turn may lead to disorders such as depression or behaviours such as aggression
- The MAOA gene varies across humans which means that some people will experience low levels of the enzyme and others will have high levels of it
- There is research evidence to support the idea that there is correlation between low levels of MAOA and antisocial, aggressive behaviour due to a dysfunctional MAOA gene

## What is the link between MAOA and serotonin?

- When levels of MAOA increase, levels of serotonin in the brain decrease, which in turn has been linked to depression
- Disorders related to depression are also affected by this drop in serotonin e.g. night terrors, increased aggression, mood imbalance
- A dysfunctional MAOA gene can be inherited which means that the affected person will have a tendency towards depression and other negative behaviours throughout their life

# Which studies investigate the MAOA gene?

■ **Brunner et al. (1993)** – a dysfunctional MAOA gene was linked to anti-social behaviour seen in the affected males of a large family in the Netherlands

# What is the link between the 5-HTT gene and serotonin?

- Although no one gene could be said to code for one behaviour, it has been established that specific genes may contribute to specific behaviours or conditions
- The MAOA gene has been linked to the **5-HTT** gene which transports serotonin around the brain
- Dysfunction and mutations of the 5-HTT gene (which results in changes in the transport of serotonin)
   has been linked to depression and anxiety
- The 5-HTT gene consists of long and short allele variants which may contribute to disorders such as depression

# Which studies investigate the 5-HTT gene?

 Caspi et al. (2003) – length of alleles on the 5-HTT gene (linked to MAOA) may be related to stress and depression

The studies by Brunner et al. (1993) and Caspi et al. (2003) can be found in 'Two Key Studies of Genes & Behaviour' on this site: just navigate the Genetics & Behaviour topic to find it.





## **Worked Example**

## SAQ (Short Answer Question) - 9 marks

'Outline how one gene may affect behaviour, using research to support your answer'. [9]

The following two paragraphs outline the gene and link it to related research:

The serotonin transporter gene (5-HTT gene) has been identified as a genetic contributor to anxiety and depression because it is linked to the reuptake of serotonin in the synapse. It has been suggested that adaptations in this gene affect the incidence of depression in an individual. Caspi et al. (2003) investigated whether a functional change in the 5-HTT gene is linked to a higher or lower risk of depression in an individual. The researchers used a sample of 847 participants aged 26 who were split into three groups, depending on the length of the alleles on their 5-HTT transporter gene.

The results showed that the participants with two short alleles on the 5-HTT gene reported more symptoms of depression in response to stressful life events than either of the other two groups. Those participants with two long alleles reported less depressive symptoms than either of the other two groups.



# Two Key Studies of Genes & Behaviour: Caspi et al. (2003) & Brunner et al. (1993)

# Key Study: Brunner et al. (1993)

**Aim**: To investigate the violent, anti-social behaviour of specific male members of a large family in the Netherlands. The behaviour exhibited by the males in the family was borderline **mental retardation** (their average **IQ** was around 85), and violent behaviour.

Participants: 5 males from a family in the Netherlands, all of whom had the same **genetic** condition, transmitted via the **X chromosome** on the **MAOA gene**. The family lived in a remote rural region of the Netherlands. Two **carrier** females and one **non-carrier** female were used as a control and compared with 3 clinically affected males. (Carrier means that some of the females carried the faulty gene in their **genotype** but it was not expressed in the **phenotype** i.e. their behaviour).

All of the affected males acted aggressively when angry, fearful, or frustrated. Examples of their violent, anti-social behaviour included attempted rape of one of the female members of the family, arson, attacking a mental institute warden with a pitchfork, voyeurism (spying on the females in the family at night), exhibitionism (appearing naked in public). Only one of the males in the family with the faulty gene finished primary education.

**Procedure**: A **case study** (close study of a small group of individuals from one family) and **quasi-experiment**. A quasi experiment is one in which the **IV** is naturally occurring i.e. it can't be manipulated by the researcher – in this case the individuals involved either had the faulty gene or they didn't have the faulty gene. Brunner conducted **DNA** analysis, obtained via urine samples. **Observations** of the males and **interviews** with the family provided **qualitative** data.

**Results**: None of the affected males had **dysmorphic** signs of the genetic mutation i.e. they didn't 'look abnormal' or different physically to the unaffected males. Unaffected males in this family attended normal schools, and most had steady jobs. All the females (including several carriers) also functioned normally.

A base change in the DNA structure was identified in all 5 affected males. This in turn resulted in flawed monoamine metabolism, which is linked with a deficit of the enzyme monoamine oxidase A (MAOA) – an enzyme which (among other functions) regulates the supply of serotonin levels to the brain. The reason only males are affected is because it is specifically the single X chromosome which is responsible for the production of MAOA.

**Conclusion**: The dysfunctional MAOA gene may be linked to irregular serotonin **metabolism** which could in turn be responsible for the mental retardation and aggressive behaviour of the affected males. MAOA deficiency may account for an individual's inability to regulate their aggression. This MAOA deficiency is now known as 'Brunner syndrome'.



# Evaluation of Brunner et al. (1993)

## Strengths

- This is a case study of one family so the researchers were able to amass a good amount of data on which to build their theory
- By using one extended family the researchers were able to directly test their theory by using family
  members as control samples rather than an unrelated general population, thus validating the idea that
  the males' behaviour was genetic rather than as a result of their environment

#### Limitations

- The use of only one family, however, does limit the **generalisability** of the findings
- The affected males may have encountered more adverse reactions from others e.g. hostility, aggression, confrontations due to their reduced IQ and lack of impulse-control which could have exacerbated their anti-social tendencies i.e. nurture may have influenced their behaviour as well as nature

#### Key terms:

- X Chromosome
- MAOA gene
- Serotonin

# Key Study: Caspi et al. (2003)

Aim: To investigate the link between the alleles of the 5-HTT gene and depression.

**Participants**: An **opportunity sample** of 847 participants aged 26 years. The sample was split into three groups, depending on the length of the alleles on their 5-HTT transporter gene:

- Group 1 two short alleles
- Group 2 one short and one long allele
- Group 3 two long alleles



**Procedure**: The participants were asked to report any **stressful** life events that had taken place between their 21st birthday up to their 26th birthday. The **Diagnostic Interview Schedule** was used to assess incidences of depression over the past year.

The researchers carried out correlational analyses between the following co-variables:

- each participant's stressful life events and incidences of depression;
- the length of each participant's alleles and incidences of depression;
- perceived stress and length of each participant's alleles.

**Results**: More depression in response to stressful life events was reported from the participants who had two short 5-HTT alleles compared to the other two groups. The participants with two long alleles reported fewer depression symptoms overall.

**Conclusion**: There may be a relationship between short 5-HTT alleles and depression - i.e. stressful life events are more likely to trigger depression in people with this genetic make-up. Long 5-HTT alleles may provide protection against stress-induced depression. The onset of depression appears to be an interaction between environment (stressful events) and **genetic make-up**.

## Evaluation of Caspi et al. (2003)

### Strengths

- The researchers exerted control by restricting the measurement of stressful life events in their sample as falling between the ages of 21 and 26 which should ensure a degree of **consistency** across the measurement thus increasing **reliability**
- Conducting three separate correlational analyses means that each measure is checked by the findings
  of the other measures which should ensure internal validity.

#### Limitations

- The experience and aetiology of depression is complex and may be due to a number of factors, both biological and non-biological which makes this study somewhat reductionist as it provides an overly simplistic explanation
- Using a self-report to collect data is prone to bias (e.g. social desirability bias, response bias) which
  reduces the validity of the findings

#### Key terms:

- 5-HTT
- Allele
- Correlation