PSY.OPT10 Biological Explanations for Schizophrenia: Genetics and Neural Correlates

Total Marks

AQA 7182 · Calculators not allowed · about 90 minutes

Name: _____ Date: ____ / ____ / _____

Answer all questions. For questions worth 4 marks or more write in full sentences. Total time guidance 90 minutes.

1 Outline one piece of genetic evidence from family studies that suggests schizophrenia has a hereditary component. Refer to concordance or relative risk in your answer and name the context.

(Total for Question 1 is 3 marks)

2 Outline the evidence from twin and adoption studies that is used to support genetic explanations for schizophrenia. Mention how each design helps separate genetic and environmental influences.

(Total for Question 2 is 3 marks)

3 Explain the concept of polygenic inheritance in schizophrenia and define what is meant by candidate genes in this context, giving an example of a gene often investigated.

(Total for Question 3 is 4 marks)

4 Outline the original dopamine hypothesis for schizophrenia and name one observation about antipsychotic drugs that supported this original view, in the context of positive symptoms.

(Total for Question 4 is 3 marks)

5 Explain the revised dopamine hypothesis, distinguishing hyperdopaminergia and hypodopaminergia and stating which brain regions are implicated for positive and negative symptoms respectively.

(Total for Question 5 is 4 marks)

6 Outline one neural correlate that has been associated with positive symptoms of schizophrenia, for example auditory verbal hallucinations. Name the brain area and the typical finding.

(Total for Question 6 is 3 marks)

7 Outline one neural correlate that has been associated with negative symptoms of schizophrenia, for example avolition. Name the brain area and the typical finding.

(Total for Question 7 is 3 marks)

8 Case application, Dopamine hypothesis. Noah reports recent onset paranoid delusions and frequent auditory hallucinations. A clinician considers the dopamine explanation. Explain, using the revised dopamine hypothesis, how dopamine abnormalities could account for Noah's positive symptoms. Refer to brain regions and mechanisms in your answer.

(Total for Question 8 is 4 marks)

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9 State the concordance rates for schizophrenia reported by Gottesman for monozygotic and dizygotic twins, giving the approximate figures and naming the study author and year, in the context of genetic evidence.

(Total for Question 9 is 2 marks)

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10 Define what is meant by a neural correlate in the study of schizophrenia and give one concise example in your answer, naming the symptom and brain region.

(Total for Question 10 is 2 marks)

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11 Explain how hypodopaminergia in the prefrontal cortex might lead to negative symptoms such as avolition or reduced emotional expression. Use neurocognitive reasoning in your answer.

(Total for Question 11 is 4 marks)

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12 Explain two methodological or interpretative limitations of using genetic and neural correlate evidence to claim a purely biological cause of schizophrenia. Give one point about genetics and one about neural correlates, and explain why each weakens a simple biological causal claim.

(Total for Question 12 is 4 marks)

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13 Discuss biological explanations for schizophrenia, including genetic explanations, the dopamine hypothesis and neural correlates. Refer to supporting evidence and to evidence or arguments that challenge biological accounts. Use the material above where relevant, and consider methodological and theoretical strengths and limitations.

(Total for Question 13 is 16 marks)

Mark scheme · PSY.OPT10 Biological Explanations for Schizophrenia: Genetics and Neural Correlates

Question 1

- **M1** states a relevant finding from family studies, for example higher concordance or increased relative risk for first degree relatives compared with the general population oe
- **M1** gives a comparative figure or clear example, e.g. siblings or children of a person with schizophrenia have a greater risk than population baseline oe
- **A1** explains why this supports a hereditary component, linking increased concordance or risk in relatives to shared genes oe
- **Answer: Family studies show higher concordance or relative risk among first degree relatives than in the general population, for example siblings or children of someone with schizophrenia have a substantially increased risk; this supports a hereditary component because close relatives share more genes.**

Question 2

- **M1** identifies twin study evidence, for example higher concordance in monozygotic MZ twins than dizygotic DZ twins oe
- **M1** identifies adoption study evidence, for example adopted-away children of parents with schizophrenia show elevated risk even when raised apart oe
- **A1** briefly explains why these findings support genetic influence, because MZ twins share 100% of genes while DZ twins share about 50%, and adoption separates genetic from family environment oe
- **Answer: Twin studies find higher concordance for schizophrenia in monozygotic than dizygotic twins, suggesting genetic influence, and adoption studies show adopted-away children of parents with schizophrenia still have an elevated risk, which supports a genetic contribution because these designs help separate genes from shared environment.**

Question 3

- **M1** defines polygenic inheritance, for example schizophrenia is influenced by many genes each contributing a small effect oe
- **M1** explains implication, for example there is no single schizophrenia gene but many loci increase risk, accounting for variability in presentation oe
- **M1** defines candidate genes, for example specific genes investigated for association with schizophrenia such as COMT or DRD2 oe
- **A1** explains how candidate genes fit into the polygenic model, for example particular candidate genes may increase risk modestly as part of a larger set of risk alleles oe
- **Answer: Polygenic inheritance means schizophrenia is influenced by many genes, each contributing a small increase in risk rather than one single gene causing the disorder. Candidate genes are particular genes studied for an association with schizophrenia, for example COMT or DRD2; these candidate genes are thought to contribute modestly to risk as part of a wider polygenic architecture.**

Question 4

- **M1** states the core claim, for example schizophrenia symptoms result from excess dopamine activity, particularly in subcortical regions, linked to positive symptoms oe
- **M1** gives a supporting observation, for example typical antipsychotic drugs block dopamine D2 receptors and reduce positive symptoms oe
- **A1** explains why this observation supports the original hypothesis, linking dopamine blockade to symptom reduction oe
- **Answer: The original dopamine hypothesis proposed that schizophrenia, especially positive symptoms like hallucinations and delusions, results from excess dopamine activity, particularly in subcortical regions. This was supported by the observation that typical antipsychotic drugs block dopamine D2 receptors and reduce positive symptoms, suggesting a link between dopamine and those symptoms.**

Question 5

- **M1** states the revision, for example the dopamine story was refined to say there may be too much dopamine in some regions and too little in others oe
- **M1** defines hyperdopaminergia in subcortical regions, such as the mesolimbic pathway, and links it to positive symptoms oe
- **M1** defines hypodopaminergia in the prefrontal cortex, such as the mesocortical pathway, and links it to negative and cognitive symptoms oe
- **A1** explains the functional implication, for example excess subcortical dopamine may cause sensory salience and hallucinations while low prefrontal dopamine may impair motivation and executive function oe
- **Answer: The revised dopamine hypothesis recognises that dopamine abnormalities are region specific: hyperdopaminergia, too much dopamine activity, in subcortical regions such as the mesolimbic pathway is linked to positive symptoms, while hypodopaminergia, too little dopamine activity, in the prefrontal cortex, is linked to negative and cognitive symptoms. Excess subcortical dopamine may cause abnormal signalling of importance leading to hallucinations, while low prefrontal dopamine may impair motivation and planning.**

Question 6

- **M1** identifies a relevant neural correlate, for example reduced activity or altered activation in Broca's area or superior temporal gyrus during auditory verbal hallucinations oe
- **M1** describes the typical finding, for example increased spontaneous activity or decreased task-related activation in language production areas linked to hearing voices oe
- **A1** briefly explains why this is relevant to positive symptoms, for example disruption in language regions may lead to misattribution of inner speech as external voices oe
- **Answer: One neural correlate of positive symptoms, such as auditory verbal hallucinations, is altered activity in language-related areas like Broca's area or the superior temporal gyrus. Studies report increased spontaneous activation or atypical task-related activity in these regions, which could lead to misattribution of inner speech as external voices.**

Question 7

- **M1** identifies a relevant neural correlate, for example reduced activity in the ventral striatum or prefrontal cortex associated with avolition or reduced motivation oe
- **M1** describes the typical finding, for example lower activation during reward anticipation or reduced metabolic activity in these regions oe
- **A1** briefly explains why this is relevant to negative symptoms, for example reduced ventral striatum response may underlie lack of motivation and diminished reward sensitivity oe
- **Answer: A neural correlate linked to negative symptoms like avolition is reduced activity in the ventral striatum or in parts of the prefrontal cortex. Imaging studies find lower activation during reward anticipation and lower metabolic activity in these regions, which could underlie reduced motivation and diminished response to rewards.**

Question 8

- **M1** applies hyperdopaminergia to Noah's positive symptoms, stating excess dopamine in subcortical regions such as the mesolimbic pathway oe
- **M1** links excess subcortical dopamine to abnormal salience or misattribution processes, explaining how this could produce delusions or hallucinations oe
- **M1** refers to specific regions relevant to hallucinations, for example increased activity in auditory/language regions combined with dopamine dysregulation oe
- **A1** integrates these points to explain plausibly how Noah's paranoid delusions and hearing voices could result from regional dopamine excess producing aberrant signalling and altered perception of internal thoughts as external oe
- **Answer: The revised dopamine hypothesis would explain Noah's delusions and hallucinations by hyperdopaminergia in subcortical regions, such as the mesolimbic pathway, causing abnormal assignment of salience to internal thoughts and perceptions. This excess dopamine can lead to aberrant signalling that helps form delusional beliefs, while altered activity in auditory and language regions may combine with dopamine dysregulation to make inner speech seem like external voices.**

Question 9

- **B1** states an approximate concordance for monozygotic twins, for example around 48% attributed to Gottesman 1991 oe
- **B1** states an approximate concordance for dizygotic twins, for example around 17% attributed to Gottesman 1991 oe
- **Answer: Gottesman reported approximate concordance rates of about 48% for monozygotic twins and about 17% for dizygotic twins, reported in work summarised around 1991.**

Question 10

- **B1** defines neural correlate, for example a brain structure or activity pattern reliably associated with a particular symptom or behaviour oe
- **B1** gives a concise example, for example reduced ventral striatum activity linked with avolition, or altered Broca's area activity linked with auditory hallucinations oe
- **Answer: A neural correlate is a brain structure or activity pattern reliably associated with a particular symptom or behaviour; for example, reduced ventral striatum activity has been linked with avolition, and altered Broca's area activity has been linked with auditory hallucinations.**

Question 11

- **M1** states that hypodopaminergia in the prefrontal cortex means reduced dopamine activity in prefrontal regions oe
- **M1** links reduced prefrontal dopamine to impaired executive functions such as planning, motivation and working memory oe
- **M1** connects these cognitive impairments to observed negative symptoms, for example reduced motivation leading to avolition oe
- **A1** explains why this matters behaviourally, for example impaired prefrontal dopamine undermines goal-directed behaviour and emotional modulation, producing flattened affect and reduced initiation oe
- **Answer: Hypodopaminergia in the prefrontal cortex means reduced dopamine activity in frontally mediated circuits, which impairs executive functions such as planning, motivation and working memory. These cognitive impairments can produce negative symptoms: reduced motivation and difficulty initiating actions can be experienced as avolition, while disrupted emotional regulation can lead to reduced emotional expression.**

Question 12

- **M1** identifies a limitation of genetic evidence, for example high heritability estimates do not prove determinism because shared environment or gene-environment interaction may be important oe
- **A1** explains why this weakens a simple biological cause claim, for example adoption and twin studies show association but cannot fully exclude environmental confounds or epigenetic mechanisms oe
- **M1** identifies a limitation of neural correlate evidence, for example correlation does not imply causation, brain differences may be a consequence of illness, medication or chronic stress rather than the cause oe
- **A1** explains why this weakens a simple biological cause claim, for example altered brain activity may reflect symptom expression or effects of antipsychotic treatment rather than being the primary cause of schizophrenia oe
- **Answer: Genetic evidence, while showing increased risk, does not prove determinism because gene-environment interactions and shared environmental factors can contribute; adoption and twin studies reduce but do not eliminate these confounds. Neural correlate evidence shows associations between brain activity and symptoms, but correlation does not imply causation, and brain differences could be consequences of the disorder, of medication, or of prolonged stress, so they do not by themselves establish a purely biological cause.**

Question 13

- **Level 4** (13-16): Knowledge of biological explanations is accurate and detailed, showing clear understanding of genetics, dopamine hypotheses and neural correlates. Evaluation is thorough and balanced, using a wide range of relevant evidence and methodological critique, including discussion of gene-environment interaction and issues of causation. Argument is coherent and applied to the topic throughout, with appropriate use of specialist terminology.
- **Level 3** (9-12): Knowledge is clear and generally accurate, covering main biological explanations. Evaluation is present and mostly effective, with some relevant evidence and methodological points. Discussion addresses strengths and limitations, but may not fully integrate evidence or consider alternative explanations in depth. Specialist terminology is used appropriately.
- **Level 2** (5-8): Some relevant knowledge of biological explanations is present but may be partial or simplified. Evaluation is limited or descriptive rather than analytical. References to evidence or methods may be brief or one-sided. Answer may lack clear organisation or full development of points.
- **Level 1** (1-4): Knowledge is minimal or inaccurate. Evaluation is very limited or missing. Answer may be largely a list of points with little coherence or development.
- **Level 0** (0): No creditworthy content.
- **Indicative content:**
  - AO1: Genetic explanations, including family, twin and adoption study findings, for example higher concordance in MZ than DZ twins, and increased risk in first degree relatives; Gottesman is commonly cited for twin concordance figures.
  - AO1: The concept of polygenic inheritance and candidate genes such as COMT and DRD2, and the idea that risk arises from many alleles each with small effect.
  - AO1: The dopamine hypothesis, original and revised. Original claim that excess dopamine activity, particularly via D2 receptors, underlies schizophrenia, supported by observation that dopamine-blocking antipsychotics reduce positive symptoms. Revised account distinguishes hyperdopaminergia in mesolimbic/subcortical regions linked to positive symptoms and hypodopaminergia in prefrontal cortex linked to negative and cognitive symptoms.
  - AO1: Neural correlates associated with positive symptoms, such as altered Broca area and superior temporal gyrus activity linked to auditory hallucinations, and correlates associated with negative symptoms, such as reduced ventral striatum or prefrontal activation linked to avolition and flattened affect.
  - AO3: Strengths, including biological explanations being supported by converging evidence from multiple methods, for example genetic epidemiology, molecular genetics and neuroimaging, and explanatory power for some clinical observations including drug effects.
  - AO3: Methodological limitations, for example twin and family studies may be confounded by shared environment and assortative mating, genome wide association studies show many loci each with tiny effect, and candidate gene findings have often failed to replicate robustly.
  - AO3: Issues of causation and directionality for neural correlates, imaging findings are correlational and may reflect consequences of illness, chronic stress or medication effects rather than primary causes; longitudinal and high risk studies complicate simple causal claims.
  - AO3: The role of gene-environment interaction and epigenetics, showing that genetic vulnerability may require environmental triggers, which weakens strong biological determinism and supports more interactionist models even while biological mechanisms remain important.
  - AO3: Clinical and practical implications, for example biological research has led to drug targets and improved understanding of neural mechanisms, but treatment response heterogeneity and side effects show limits to a purely biological approach.
  - AO3: Alternative or complementary explanations, for example psychological and social factors that interact with biology, and the need for integrative models that account for developmental timing, stress, cannabis use and social adversity.
  - AO3: Evaluation of the evidence quality, including replication issues in genetic associations, sample sizes, population stratification, and differences between groups and individuals, which limit generalisability of some biological findings.