

Genes or lifestyle? exam style question

Introduction

This question looks at the evidence for genetic risk factors for a major cause of blindness and then at the influence of two lifestyle factors and how these might interact with the genetic risk factors. Finally students have to translate the information into a health education message.

Suggested answers

(a)(i)	<ul style="list-style-type: none"> one allele increases risk two increases risk (by approximately same amount) again gene 2 has greater effect than gene 1 very strong effect/ each allele doubles risk 	<i>any 3 for 1 mark each</i>	3
(ii)	<ul style="list-style-type: none"> 2 CF alleles leads to certain illness AMD alleles increase risk /not certain CF/ sickle cell are recessive one allele has no effect 	<i>any 2 for 1 mark each</i>	2
(iii)	<ul style="list-style-type: none"> group studied prospectively monitored for risk factors outcomes compared for groups with and without risk factors 	<i>any 2 for 1 mark each</i>	2
(b)(i)	<ul style="list-style-type: none"> $5.66 \times 6 = 34$ risk is 34 per 1000 give 2 marks for correct answer	<i>for 1 mark each</i>	2
(ii)	<ul style="list-style-type: none"> true value not known true value lies somewhere between 3.69-8.76 this is known with 95% probability wide confidence interval means a lot of uncertainty over true value 	<i>any 2 for 1 mark each</i>	2
(c)(i)	<ul style="list-style-type: none"> obesity and smoking increase risk of AMD increases risk for those with normal and high risk alleles an example of risk increase e.g. from 1.00 to 1.98 for obesity in YY 	<i>any 2 for 1 mark each</i>	2
(ii)	<ul style="list-style-type: none"> both increase risk neither causes directly effect is additive /lifestyle factors have greater effect if risk alleles also present 	<i>any 2 for 1 mark each</i>	2
(d)	use level guidance below to put answer into one of 3 levels Points that might be included, credit any other valid ones <ul style="list-style-type: none"> what is AMD AMD families <ul style="list-style-type: none"> at greater risk/ inherited risk not certain to develop AMD risk can be reduced by lifestyle choices specific advice on lifestyle general public <ul style="list-style-type: none"> risk figure lifestyle choices increase/double risk specific advice on lifestyle 		

Level	Descriptor	Mark range	Level guidance for this question
3	<p>Good</p> <p>Claims supported by an appropriate range of evidence</p> <p>Good use of information or ideas about science going beyond those given in the question</p> <p>Argument well structured with minimal repetition or irrelevant points</p> <p>Accurate and clear expression of ideas with only minor errors of grammar, punctuation and spelling</p>	5-6	<p>two points from each section well done</p> <p>Use structure/ appropriate language for 5/6</p> <p>Must be aimed at public for 6</p>
2	<p>Modest</p> <p>Claims partially supported by evidence</p> <p>Good use of information or ideas about science given in the question but limited beyond this</p> <p>The argument shows some attempt at structure</p> <p>The ideas are expressed with reasonable clarity but with a few errors of grammar, punctuation or spelling</p>	3-4	<p>any 3 points</p> <p>Use quality of explanation structure/ language for 3/4</p>
1	<p>Limited</p> <p>Valid points but not clearly linked to an argument structure</p> <p>Limited use of new information or ideas about science</p> <p>Unstructured</p> <p>Errors in grammar, punctuation and spelling or lack of fluency</p>	1-2	<p>only 1 or 2 points</p> <p>Quality of explanation and language for 1/2</p>
0	Incorrect or no response	0	

Data taken from D.A.Schaumberg et al (2007) *A prospective Study of 2 Major Age-Related Macular Degeneration Susceptibility Alleles and Interactions With Modifiable Risk Factors* Arch Ophthalmology. 2007;125:55-62

October 2008

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Age-related macular degeneration, AMD, is one of the most common causes of blindness or poor vision in older people. There is some evidence that the causes are genetic and two genes have been identified as increasing the risk of AMD. The data in this question are all taken from an ongoing *cohort study* of a large number of health workers in the USA.

risk gene	allele	Incidence rate of AMD in those with risk allele as a ratio of rate in those with normal gene (IRR) (95% confidence interval)
1	YY (normal)	1.00
	YH	1.98 (1.64 – 2.40)
	HH	3.92 (2.69 – 5.76)
2	AA (normal)	1.00
	AS	2.38 (1.92 – 2.96)
	SS	5.66 (3.69-8.76)

Figure 1 Risk of developing AMD associated with two particular genes

(a) (i) What does the information in figure 1 tell you about the effect of these alleles on the risk of developing AMD?

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(3 marks)

(ii) How is this different from the effect of an allele such that for cystic fibrosis or sickle cell disease?

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(2 marks)

(iii) What is meant by a cohort study?

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(2 marks)

(b) (i) If the incidence of AMD in the normal population of this age group is 6 per 1000 then what is the incidence in those with SS alleles on gene 2?

.....

(2 marks)

(ii) What is the meaning of the '95% confidence interval' shown in brackets for the SS alleles as 5.66 (3.69 –8.76)

.....

(2 marks)

Risk factor		Alleles	
		YY	HH
BMI* <30	IRR	1.00	3.96
BMI >30	IRR	1.98	12.28
non-smoker	IRR	1.00	4.23
smoker	IRR	2.34	8.69

*BMI stands for Body Mass Index. High values are an indication of obesity

Figure 2 Risk of developing AMD according to two genotypes and two lifestyle risk factors

(c) (i) What does the information in Figure 2 tell you about the affect of the lifestyle factors of obesity and smoking on the risk of developing AMD?

.....

(2 marks)

(ii) What would you say if you were asked, "Is AMD caused by genes or by lifestyle?"

.....

(2 marks)

- (d) Write a short information leaflet, based on research reported here, suitable for use in an optician's waiting room entitled;

How to reduce your risk of developing age-related macular degeneration

Include information for those who know AMD runs in their family as well as for the general public.

(6 marks)

Total 21 marks