Biology 2019 v1.2

IA3 high-level annotated sample response

July 2018

Research investigation (20%)

This sample has been compiled by the QCAA to assist and support teachers to match evidence in student responses to the characteristics described in the instrument-specific marking guide (ISMG).

Assessment objectives

This assessment instrument is used to determine student achievement in the following objectives:

- 2. apply understanding of DNA, genes and the continuity of life or the continuity of life on Earth to develop research questions
- 3. analyse research evidence about DNA, genes and the continuity of life or the continuity of life on Earth
- 4. interpret research evidence about DNA, genes and the continuity of life or the continuity of life on Earth
- 5. investigate phenomena associated with DNA, genes and the continuity of life or the continuity of life on Earth through research
- 6. evaluate research processes, claims and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth
- 7. communicate understandings and research findings, arguments and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth.

Note: Objective 1 is not assessed in this instrument.





Instrument-specific marking guide (ISMG)

Criterion: Research and planning

Assessment objectives

- 2. apply understanding of DNA, genes and the continuity of life or the continuity of life on Earth to develop research questions
- 5. investigate phenomena associated with DNA, genes and the continuity of life or the continuity of life on Earth through research

The student work has the following characteristics:	Marks
 informed application of understanding of DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by a considered rationale identifying clear development of the research question from the claim effective and efficient investigation of phenomena associated with DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by a specific and relevant research question selection of sufficient and relevant sources. 	5–6
 adequate application of understanding of DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by a reasonable rationale that links the research question and the claim effective investigation of phenomena associated with DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by a relevant research question selection of relevant sources. 	3–4
 rudimentary application of understanding of DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by a vague or irrelevant rationale for the investigation ineffective investigation of phenomena associated with DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by an inappropriate research question selection of insufficient and irrelevant sources. 	1–2
does not satisfy any of the descriptors above.	0

Criterion: Analysis and interpretation

Assessment objectives

- 3. analyse research evidence about DNA, genes and the continuity of life or the continuity of life on Earth
- 4. interpret research evidence about DNA, genes and the continuity of life or the continuity of life on Earth

The student work has the following characteristics:	Marks
 systematic and effective analysis of qualitative data and/or quantitative data within the sources about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by the identification of sufficient and relevant evidence thorough identification of relevant trends, patterns or relationships thorough and appropriate identification of limitations of evidence insightful interpretation of research evidence about DNA, genes and the continuity of life or the continuity of life or the continuity of life on Earth demonstrated by justified scientific argument/s. 	<mark>5</mark> –6
 effective analysis of qualitative data and/or quantitative data within the sources about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by the identification of relevant evidence identification of obvious trends, patterns or relationships <u>basic identification of limitations of evidence</u> adequate interpretation of research evidence about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by reasonable scientific argument/s. 	3–4
 rudimentary analysis of qualitative data and/or quantitative data within the sources about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by the identification of insufficient and irrelevant evidence identification of incorrect or irrelevant trends, patterns or relationships incorrect or insufficient identification of limitations of evidence invalid interpretation of research evidence about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by inappropriate or irrelevant argument/s. 	1–2
does not satisfy any of the descriptors above.	0

Criterion: Conclusion and evaluation

Assessment objectives

- 4. interpret research evidence about DNA, genes and the continuity of life or the continuity of life on Earth
- 6. evaluate research processes, claims and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth

The student work has the following characteristics:	Marks
 insightful interpretation of research evidence about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by justified conclusion/s linked to the research guestion critical evaluation of the research processes, claims and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by insightful discussion of the quality of evidence extrapolation of credible findings of the research to the claim suggested improvements and extensions to the investigation that are considered and relevant to the claim. 	5–6
 adequate interpretation of research evidence about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by reasonable conclusion/s relevant to the research question basic evaluation of the research processes, claims and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by reasonable description of the quality of evidence application of relevant findings of the research to the claim suggested improvements and extensions to the investigation that are relevant to the claim. 	3– <mark>4</mark>
 invalid interpretation of research evidence about DNA, genes and the continuity of life and or continuity of life on Earth demonstrated by inappropriate or irrelevant conclusion/s superficial evaluation of the research processes, claims and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by cursory or simplistic statements about the quality of evidence application of insufficient or inappropriate findings of the research to the claim ineffective or irrelevant suggestions. 	1–2
does not satisfy any of the descriptors above.	0

Criterion: Communication

Assessment objective

7. communicate understandings and research findings, arguments and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth

The student work has the following characteristics:	Marks
• effective communication of understandings and research findings, arguments and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by	
 fluent and concise use of scientific language and representations 	<u>2</u>
 appropriate use of genre conventions 	
 acknowledgment of sources of information through appropriate use of referencing conventions. 	
 adequate communication of understandings and research findings, arguments and conclusions about DNA, genes and the continuity of life or the continuity of life on Earth demonstrated by competent use of scientific language and representations 	1
 use of basic genre conventions 	
- use of basic referencing conventions.	
 does not satisfy any of the descriptors above. 	0

Task

Context

Investigate one of the following claims:

- Gene therapy can reverse ageing.
- DNA testing can prove genealogical ancestry.
- Transgenic organisms offer a viable and effective future for human health.
- Sustainable reserve size should be based on the data from gene pool variability.

You may identify an alternative claim in consultation with your teacher. This claim must be related to Unit 4 subject matter.

Task

Gather secondary evidence related to a research question in order to evaluate the claim. Develop your research question based on a number of possible claims provided by your teacher.

Obtain evidence by researching scientifically credible sources, such as scientific journals, books by wellcredentialed scientists, and websites of governments, universities, independent research bodies or science and technology manufacturers. You must adhere to research conventions.

Sample response

Criterion	Allocated marks	Marks awarded
Research and planning Assessment objectives 2, 5	6	6
Analysis and interpretation Assessment objectives 3, 4	6	5
Conclusion and evaluation Assessment objectives 4, 6	6	4
Communication Assessment objective 7	2	2
Total	20	17

The annotations show the match to the instrument-specific marking guide (ISMG) performancelevel descriptors.

Key: Research and planning

Analysis and interpretation Conclusion and evaluation

Communication

Note: Colour shadings show the characteristics evident in the response for each criterion.

Research and planning [5-6]

a considered rationale identifying clear development of the research question from the claim

The rationale shows the process by which the research question has been developed from the claim.

Research and planning [5-6]

selection of sufficient and relevant sources

Sources are related to the topics covered in the unit and are adequate for the development of a scientific argument that responds to the research question.

Rationale

Brown (2010) in the X-Men Mail recently claimed that 'human evolution is still occurring'. The article discussed broadly the link between cultural practice and humans but did not detail a specific example of a scientific case study where cultural practice was driving this supposed evolution.

Consequently, with initial research, a broad research question 'does cultural practice drive human evolution' was developed based on the initial claim. This was further refined to specifically consider human geneculture coevolution. A summary of this refinement and the specific research question is detailed below.

The Homo sapien population has grown from an estimated 5 million people in the pre-Neolithic era to an estimated 7.4 billion in 2016 (Biraben 2003) (Population Reference Bureau 2016). The exponential growth observed in the last 50 000 years can be linked to a variety of cultural factors that have enabled a greater portion of humans to reach reproductive age and live longer lives (Cochran and Harpending, 2009). Consequently, in this larger population, higher levels of gene flow and variation occur between individuals (microevolution) and the likelihood of macro evolutionary change is increased. This can be attributed to an increased range of favourable, advantageous genetic combinations arising in a population whilst smaller populations may be more likely to experience a high degree of genetic drift. However large-scale scans using patterns of linkage disequilibrium to detect recent selection (i.e. in the last 10, 000 years) suggest that many genes may have evolved in response to agriculture (Richerson et al 2010).

Research and planning [5–6]

a specific and relevant research question

The response clearly defines the research question so sufficient and relevant data can be collected. The research question is connected to the rationale and the topics covered in the unit.

Analysis and interpretation [5–6]

justified scientific argument/s

Scientific arguments are evident throughout the response. The background shows development of the argument by explaining lactose, lactase persistence as a phenotype, genetic markers for LP alleles and links these to environmental changes.

Communication [2]

appropriate use of genre conventions

The use of headings and paragraphs fits the purpose of a scientific essay.

Analysis and interpretation [5–6]

identification of sufficient and relevant evidence

The evidence in the response draws upon the available qualitative and quantitative data to respond to the research question. It links directly to the research question. Coevolution is the process used to describe cases where species influence each other during their evolution. Human gene-culture coevolution can be observed in technological advancements such as agriculture. With the concept being that if 'the cultural inheritance of an environment-modifying human activity persists for long enough to generate a stable selection pressure; it will be able to co-direct human evolution' (Gerbault 2011). The cultural process of managing wild cattle into herded livestock, and consequently domestication, has its oldest evidence dated at 10 500 BP (Helmer 2005). Thus, this *environmentmodifying activity could have co-directed the evolution of lactase persistence in humans.* Therefore, this essay proposes the following research question:

'Is the prevalence of hypolactasia (lactose intolerance) in Finland linked to the practice of domesticating cattle for dairy purposes?'

Background

Lactose is the main carbohydrate found in milk. Typically, human infants can readily digest milk in order to grow and develop due to the presence of the enzyme lactase in their intestine. The production of this enzyme typically declines in adulthood removing a significant source of available dietary carbohydrate. Some humans continue to express lactase throughout adult life, and are thus able to digest the lactose found in fresh milk. This trait is called lactase persistence (LP) and the frequency of this phenotype is found in around 35 per cent of adults living in the world today (Gerbault 2011).

According to Ward (2009) through tracing the genetic markers of previous generations, that early Europeans, particularly from Sweden and Denmark, had the capacity to digest milk, while others from China and Africa from around a similar time period appear to have had a milk intolerance. Gerbault (2011) supports this finding with evidence showing that that a single mutation (-13910*T) can be used to explain the distribution of lactase persistence across Europe, whereas several mutations are associated with it in Africa and the Middle East. Additionally, Pagel (1999) also noted that there were three, distinctly different groups of humans that appeared to independently demonstrate, through the selection of rare alleles, lactose tolerance into adulthood, suggesting that this mutation was brought about by a change in the environment of these early Europeans. Current estimates for the age of lactase persistence-associated alleles appear to align with the origins of the culturally transmitted practice of dairying (Gerbault 2011).

Evidence

In Finland there are a variety of genealogies forming the genetic makeup of the country. The population is mainly comprised of a variety of groups of people who identify as speaking Finnish, Swedish and Norwegian. There are also the minority (60 000 – 110 000 individuals) indigenous people of *Sápmi*, a territory comprising parts of Arctic Norway, Sweden, Finland, and Russia's Kola Peninsula, called the Sami (Bjerregaard and Young 2008 as cited in Melbøe et al 2016). According to census data (which requires people to self-identify as Sami) the Finnish population of Sami is approximately 4 500 compared to 5.496 million Finns (Official

statistics of Finland 2016). This minority group show differences in cultural practices and genealogy when compared to the majority Finnish demographic.

In terms of the practice of ancient dairying current evidence suggests domestication of livestock in Finland and large-scale animal husbandry was occurring during the Bronze Age (3500-2500 years ago) (Præbel 2016). To further support this, analysis of core ware pottery may have 'revealed Neolithic settlers in Finland may have been consuming dairy foods as early as 2500 BC (Cramp et al 2014)'. However, data from Hansen et al (2015) states that the prevalence of lactose intolerance in Finnish-speaking Finns has been documented at 17% whilst in the Finnish Sami population the prevalence ranges from 25 to 60%. This is a noticeable variance which could be explained by the access to cow's milk between the different populations.

On one hand the Sami have traditionally participated in farming, fishing, trapping and reindeer husbandry (breeding and herding). Their ancient diet may have consisted of a 'high intake of fatty fish, red meat (primarily reindeer), fat, blood and organs, wild berries and boiled, unfiltered coffee, and low intakes of cultivated vegetables and fruit, bread and fibre (Nilsson 2012)'. Sahi 1994 indicates that reindeer dairy farming however only developed late in the 18th century. This combined evidence would suggest that cow's milk was not a part of the Sami diet and thus would not have influenced the lactase persistence frequency in this gene pool.

A further comparison in Table 1: Comparison of average cow and reindeer milk illustrates that the lactose percentages and production rates are very different in each of the dairy sources. Thus, it could be assumed that lactase persistence would not be required to supplement nutrition in a population consuming small amounts of low lactose milk.

Table 1: Comparison of average cow and reindeer milk

Ruminant breed	Milk description and production
Average dairy cow	 5.0% lactose Produce approximately 30.0L per day 3.8 - 3.9% fat 3.0% protein
Reindeer	 2.4 % lactose Produce approximately 0.4L per day 22.5% butterfat 10.3% protein

Source: (Haglin 1991)

Genetic information further supports this assumption. <u>Table 2: Lactase</u> persistence-associated alleles frequencies in populations of Finland shows the prevalence of the -13910T allele is higher in the population of Finns compared to the Sami. It should also be noted that none of the other multiple alleles associated with lactase persistence are shown in either population.

Analysis and interpretation [5–6]

thorough identification of relevant trends, patterns or relationships

The identified relationships are adequate for the purpose of responding to the research question and can support a valid conclusion. They have direct bearing upon and are applicable to the formation of the scientific argument.

Population Long Lat Ν -14010 -13915 -13907 -13910Sum of all LP Predicted G>C T>G C>G C>T associated lactase alleles persistence

0.00

0.00

-

0.58

0.17

0.58

0.17

Table 2: Lactase persistence-associated alleles frequencies in populations of Finland

Source: Enattah et al 2008 Am J Hum Genet. (via Global Lactase persistence association database).

-

Evaluation

28.00

29.00

65.00

69.00

1876

60

0.00

_

Finns

Saami

Analysis and interpretation [3-4]

basic identification of limitations of evidence

The response gives fundamental limitations of the evidence. However, this is not thorough, as the response only shows partial identification of limitations of the evidence.

There are issues associated with the evidence presented however. Haglin was the only source for information about the lactose percentages in reindeer's milk. It could be assumed that this is because it is very cold in Finland and it takes two people to milk a reindeer to get 2 cups of milk. Maybe scientists are not interested in these conditions and don't really want to find out more about reindeer. Consequently, only one source of evidence limits the reliability of this information.

Also the sample size (N) in Table 2 is not indicative of the Finnish population. The genetic survey of 1876 Finns is only about 3.5% of the population and 60 of the Sami is 1.3%. This is not the entire population for either group so therefore cannot really be used to make significant conclusions. Also, only one source of evidence was found which again limits the reliability of this data.

The data in Table 2 is also limited due to the testing process used to obtain the data. Itan 2010 indicates that the BG (blood glucose) and BH (breath hydrogen) tests have error rates associated with them. These are type I and II errors. They also state that loss of lactase can be associated with gut trauma not just the LP phenotype. In addition to this the technique used to obtain the data in Table 2 can only test for the alleles in the table. How do we know that more allele types don't exist?

frequency

0.82

0.31

Conclusion and evaluation [3–4]

reasonable description of the guality of evidence

The response gives a sound account of the standard of evidence. However, the response does not describe type I and II errors and sampling techniques for data.

Conclusion and evaluation [5–6]

justified conclusion/s linked to the research guestion

The conclusion provides sound reasons that draw upon the scientific arguments.

Conclusion and evaluation [3–4]

application of relevant findings of the research to the claim

The response uses some pertinent outcomes of the research to address the claim. However, the response does not identify the plausible implications of other conclusions.

Conclusion and evaluation [3-4]

suggested improvements and extensions to the investigation that are relevant to the claim

The improvements and extensions to the investigation are applicable to the claim but do not show evidence of careful or deliberate thought.

Communication [2]

fluent and concise use of scientific language and representations

The response is easily understood, avoids unnecessary repetition and meets the required length. This data also doesn't take into account that the change in alleles between populations isn't just because of strong genetic drift. Without further research to support how the Finns and Sami differ it is difficult to comment on this. Therefore, this would be a plausible area of further research to find out more on.

It should also be noted that just because dairying and the LP alleles appear at similar times it doesn't mean this is a causal relationship. It could simply be a strong correlation. This difference means that whilst the two variables correlate it does not mean one thing caused the other.

Conclusion

In conclusion, the claim that human evolution is still occurring is not fully supported. Because the question is broad it requires more experimentation than is possible. If the specific research question is considered this also cannot be fully supported. This is because the evidence supplied for the research question is about historical data. A longitudinal study could be done to track the Finn and Sami populations over time. Also, further research could be done to see if there are more unknown alleles in the population.

Word count: <u>1591</u>

Research and planning [5–6]

selection of sufficient and relevant sources

Sources are scientific and provide enough evidence for the development of a scientific argument that responds to the research question.

Communication [2]

acknowledgment of sources of information through appropriate use of referencing conventions

The use of a referencing system fits the purpose of an essay.

Reference List

Gerbault P, Liebert A, Itan Y, Powell A, Currat M, Burger J, Swallow DM, Thomas MG. 2011. Evolution of lactase persistence: an example of human niche construction. Philos Trans R Soc Lond B Biol Sci. 2011 366(1566):863-77.

Biraben J-N. 2003. The rising numbers of humankind. Population & Societies No. 394 Onstitut national d'études démongraphiques

Cochran, G & Harpending, H 2009, The 10,000 Year Explosion, Basic Books, New York.

Cramp L., Evershed R., Lavento. M, Halinen. P, Mannermaa K., Oinonen M., Kettunen J., Perola M., Onkamo P., Heyd V. Neolithic dairy farming at the extreme of agriculture in northern Europe. Proc Biol Sci. 2014 Sep 22; 281(1791): 20140819.

Eliassen BM, Melhus M, Hansen KL, Broderstad AR. Marginalisation and cardiovascular disease among rural Sami in Northern Norway: a population-based cross-sectional study. BMC Public Health. 2013;13:522

Hansen K.L., Brustad. M, Johnsen. K. 2015. Prevalence of self-reported stomach symptoms after consuming milk among indigenous Sami and non-Sami in Northern and Mid Norway – the SAMINOR study

Haglin L. Nutrient intake among Saami people today compared with an old, traditional Saami diet. Arctic Med Res. 1991;(Suppl):741–6. (cross-referenced via Ross A.B. 2006)

Helmer D., Gourichon L., Monchot H., Peters J., Sana Segui M. 2005. Identifying early domestic cattle from pre-pottery Neolithic sites on the Middle Euphrates using sexual dimorphism. In First steps of animal domestication new archaeozoological approach (eds Vigne J. D., Helmer D., Peters J., editors.), pp. 86–95 Oxford, UK: Oxbow

Ingram C.J., Liebert A., Swallow D. 2012. Population Genetics of Lactase Persistence and Lactose Intolerance' University College London, UK

Ingram C. J., Mulcare C. A., Itan Y., Thomas M. G., Swallow D. M. 2009. Lactose digestion and the evolutionary genetics of lactase persistence. Hum. Genet. 124, 579–591

Itan Y., Jones B. L., Ingram C. J., Swallow D. M., Thomas M. G. 2010. A worldwide correlation of lactase persistence phenotype and genotypes. BMC Evol. Biol. 10, 36.

Itan Y., Powell A, Beaumont M., Burger J., Thomas M.G.,2009 The Origins of Lactase Persistence in Europe. PLoS Comput Biol. 2009 Aug; 5(8): e1000491.

Kozlov A, Borinskaya S, Vershubsky G, Vasilyev E, Popov V, Sokolova M, et al. Genes related to the metabolism of nutrients in the Kola Sami population. Int J Circumpolar Health. 2008;67:56–66.

Melboe L., Hansen K.L., Johnsen B-E., Fedreheim G.E., Dinesen T.,

Minde G-T., Rustad M. 2016 Ethical and methodological issues in research with Sami experiencing disability. Int J Circumpolar Health. 2016; 75:
Nilsson LM, Winkvist A, Brustad M, Jansson JH, Johansson I, Lenner P, et al. A traditional Sami diet score as a determinant of mortality in a general northern Swedish population. Int J Circumpolar Health. 2012;71:1–12.
Pagel, M 1999, 'Inferring the historical patterns of biological evolution', Nature, vol 401, no. 6, pp. 877884.
Population Reference Bureau 2016. 2016 World Population Data Sheet retrieved from www.prb.org/Publications/Datasheets/2016/2016-world-population-data-sheet.aspx
Præbel A., Prehistory of farm animals. 2016 NordGen, retrieved from http://www.nordgen.org/index.php/en/content/view/full/1743 on 15/09/16
Richerson, P. Boyd, R, Henrich, J. 2010. Gene-culture coevolution in the age of genomics. PNAS vol. 107 Supplement 2
Ross A.B, Johansson. A, Ingman .M, Gyllensten .U,. 2006. Lifestyle, Genetics and Disease in Sami. Croat Med J. 2006 Aug; 47(4): 553–565.
Sahi T. Genetics and epidemiology of adult-type hypolactasia. Scand J Gastroenterol. 1994;202(Suppl):7–20.