

Syllabus	PR_IK_03
Topic	Malignant hyperthermia

Adam is a 10-year old boy who presents to the Emergency Department with abdominal pain. He is subsequently listed by the general surgeons for an emergency appendicectomy. His father has malignant hyperthermia (MH), and Adam has never been tested. This father is concerned Adam may have inherited the disease

a)
Define MH? (2 mark)

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b)
What is the most common genetic abnormality and what effect does this abnormality have? (2 marks)

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c)
List four clinical features of MH (4 marks)

1.
2.
3.
4.

d)
How does MH effect muscle contraction? (7 marks)

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e)

Over what time period could MH recur? (1 mark)

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f)

Where is the MH testing centre in the UK? (1 mark)

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g)

What is in-vitro contracture testing? (3 marks)

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Q	Answer	Mark	Guidance
a)	<ul style="list-style-type: none"> Progressive Life-threatening Hyperthermic reaction Occurring during general anaesthesia 	2	<p><u>Must</u> include during anaesthesia for full marks</p>
b)	<ul style="list-style-type: none"> RYR1 gene mutation Sustained release of calcium into cell 	2	
c)	<ul style="list-style-type: none"> Rise in EtCO₂ Tachypnoea <u>if breathing spontaneously</u> <u>Unexpected</u> increase in HR Rise in core body temperature Muscle rigidity 	4	<p>Must specify:</p> <ul style="list-style-type: none"> If breathing <u>spontaneously</u> <u>Unexpected</u> increase in HR
d)	<ul style="list-style-type: none"> Excitation-contraction coupling is the transduction of electrical energy to mechanical work of muscle contraction Exposure to trigger agents leads to dysregulation of ECC and sustained release of calcium into the cytosol Initially this causes increased metabolic demand for ATP to sequester calcium causing increase in CO₂ production of O₂ consumption Rise in CO₂ stimulates SNS causing increase in HR As the process continues calcium release into cell exceeds calcium sequestration causing muscle rigidity and heat production The initial acid-base disturbance is a respiratory acidosis Sustained muscle contraction affects sarcolemmal integrity leading to release of K ions, CK and myoglobin This leads to hyperkalaemia, AKI, hyperthermia, rhabdomyolysis and DIC 	7	
e)	<ul style="list-style-type: none"> 14 hours 	1	<ul style="list-style-type: none"> Accept monitor for 24 hrs
f)	<ul style="list-style-type: none"> Leeds 	1	
g)	<ul style="list-style-type: none"> Fresh vastus muscle biopsy Taken under LA 	3	

	<ul style="list-style-type: none">• Specimens exposed to increasing concentrations of halothane or caffeine to assess rate of contracture		
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References:

- 1) Gupta PK, Hopkins PM. Diagnosis and management of malignant hyperthermia. (2017) BJA Education 17(7)249-254 <https://academic.oup.com/bjaed/article/17/7/249/3752451>