

EDMGG Examination: Sample MCQs

1.	A 10-year-old child has a bilateral radial ray deficiency. The child has normal neurodevelopment and growth, with head circumference on the 50 th centile. Physical examination shows absent thumbs and normal ears.	
	Cardiac ultrasound: <i>ostium secundum</i> atrial septal defect	
	What is the most likely diagnosis ?	
	a.	22q11 microdeletion syndrome
	b.	CHARGE syndrome
	c.	Fanconi anaemia
	d.	Holt-Oram syndrome
	e.	TAR syndrome
Answer	D	

2.	A 27 year-old man has proportionate short stature. He has a newborn son.	
	Genetic testing: Pathogenic mutation identified in the <i>SHOX</i> gene (located in a pseudoautosomal region)	
	What is the risk that the son carries the mutation ?	
	a.	<1%
	b.	10%
	c.	25%
	d.	50%
	e.	100%
Answer	D	

3.	A 26-year old woman is known to have a premutation in the fragile X gene.	
	Which clinical complication will she be at risk of ?	
	a.	Fragile X tremor ataxia syndrome
	b.	Joint hyperlaxity
	c.	Premature ovarian failure
	d.	Seizures
	e.	Severe Intellectual disability
Answer	C	

4.	A 10-year-old girl has autism spectrum disorder. There is no family history of intellectual disability. Trio-based exome sequencing is performed.	
	Results: <i>de novo</i> point mutation in <i>CHD8</i> : c.[3519-2A>G];[=]	
	Which is the most likely effect of this mutation ?	
	a.	Missense mutation
	b.	No effect
	c.	Nonsense mutation
	d.	Synonymous variant
	e.	Splicing mutation
Answer	E	

5.	A 65-year-old male presents with fine tremor in his hands, progressive gait instability and poor balance and coordination. He has two healthy daughters one of whom has an 8-year-old son with significant intellectual disability.	
	Which one of the following genes is most likely to show a causative mutation?	
	a.	Androgen receptor
	b.	<i>FMR1</i>
	c.	Dystrophi
	d.	Huntingtin
	e.	<i>SMN</i>
Answer	B	