ECMGG 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 50th centile. Physical examination shows absent thumbs and normal ears.		
	Cardiac ultrasound: *ostium secundum* 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 *SHOX* 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: *de novo* point mutation in *CHD8*: 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.	*FMR1*	
	c.	Dystrophin	
	d.	Huntingtin	
	e.	*SMN*	
Answer	B		