

Essential Clinical Genetics for LEND and UCEDD Programs

Readiness Assurance Test

1. Which of the following tests has the highest diagnostic yield in a child with intellectual disability, autism, or multiple congenital anomalies?
 - A. Brain MRI
 - B. Chromosomal microarray analysis
 - C. FISH analysis
 - D. Karyotype
2. A mutation is any change in the DNA sequence.
 - A. True
 - B. False
3. A patient in Genetics Clinic has hearing loss, Hirschsprung disease, and a white forelock. Which of the following databases can help determine which genetic conditions are associated with these clinical features?
 - A. American College of Medical Genetics (ACMG) ACTION sheets
 - B. GeneTests.org
 - C. Online Mendelian Inheritance in Man (OMIM)
 - D. National Coalition for Health Professional Education in Genetics
4. Etiologies for sensorineural hearing loss include which of the following?
 - A. Single gene mutations
 - B. Exposure to teratogenic drugs prenatally (like aminoglycosides)
 - C. Intrauterine infections
 - D. All of the above
5. Which of the following statements is TRUE about genetic testing?
 - A. Genetic testing should only be performed by a geneticist or genetic counselor
 - B. Genetic testing should only be performed once the patient or surrogate has received pre-test counseling.
 - C. A child with intellectual disability should only receive genetic testing if he has dysmorphic features.
 - D. Genetic testing should only be recommended if there is a family history of a genetic condition.