

FINAL QS

- 1-Mismatch: down syndrome > 47 ch 18
- 2-best method to decrease genetic disorders: preventive sth
- 3-Baldness w moustache male: sex influenced
- 4-Neonatal screening routinely involve: hypothyroidism
- 5-Checkups that should be made for babies in Jordan: hypothyroidism, g6pd and pku
- 6-Wrong statement: can't use bone marrow for stem cells aw eshi haik
- 7-Female has small ear, hypotonia, congenital heart disease and protruded tongue : trisomy 21
- 8-group of diseases related together without a known etiology: association
- 9- Can we have XY but a female? >> all of the above (deletion of sry gene, mutation of sry gene, translocation of sry gene from y to x)
- 10-Not associated with cancer inherited gene > leukemia
- 11-Not associated with metabolic treatment: enzyme replacement
- 12-least invasive and highest invasive: ultrasound // fetoscopy (highest abortion rate)
- 13-Father and mother have recessive albinism gene but their children were normal, this trait is? Mosaicism
- 14-PKU best treatment: food restriction
- 15-Wrong about autosomal recessive indications: treatment should be available
- 16-Translocation between ch 9 and 22? Chronic myelogenous leukemia
- 17-Alkaptonurea? Homogeniatic oxidase
- 18-Aneuploidy most compatible with life? Down syndrome
- 19-Not associated with multifactorial disease? Increased incidence of the siblings having the disease if a person has the disease late in life
- 20-proto oncogene function? Cell growth and development
- 22-Uses of FISH: whole chromosomal painting
- 23-Cant be seen by light microscope: reciprocal translocation
- 24-Chemical not used in metaphase screening: quinacrine
- 25-Angelman syndrome? Unilateral from parent
- 26-Causes of hypercholestermia? LDL receptor

27-study of chromosomes and cell division : cytogenetics
28-Not from hardy weinberg principles? Small population
29-Most stable chromosome >> 45 xy 14-21, t(14,21) or eshi haik 30-
Klinefelter syndrome: male with 47 XXY
31- expressivity definition: degree to which a gene is expressed
depending on other genes or environmental factors
32- Pedigree of ? > autosomal dominant
33- : angelman - micro deletion incorrect
34-Define hereditary
35-Which can't be used for neural tube open : CVS
36-True about mitochondrial inheritance
37-Not a storage disease: galactosemia
38-Not true: maple syndrome - autosomal dominant
39-wrong about prenatal diagnosis: parents have bipolar maniac
depression
41-wrong about inborn errors: appear in adolescence
44- monozygotic twin study 40% diseased, while normal siblings 10%
disease shared, best statement describing this gene abnormality? gene
contributes but doesn't determine the disease
45-karyotyping not used in : insertion/deletion
46- all of the following require a parent chromosomal analysis except: a
short daughter