

Test questions for Bi7291en Human Genetics

- 1) Middle age theories of genetics
- 2) Mendel´s CV
- 3) Monohybrid crossing
- 4) Reciprocal crossing
- 5) Dihybrid crossing
- 6) Molecular basis of Mendel´s laws
- 7) Heredity of sexuality and sex-linked traits
- 8) Rules of sex-linked inheritance
- 9) Human sex linked traits and heredity
- 10) Exceptions from Mendel´s ratios
- 11) Central dogma of molecular biology
- 12) Proof of DNA as a carrier of genetic information
- 13) Watson-Crick model of DNA
- 14) *In situ* hybridization
- 15) DNA sequencing principles
- 16) PCR method
- 17) Massive parallel sequencing
- 18) Human genome project
- 19) 1000Genomes project
- 20) Development of human cytogenetics
- 21) Chromosomes and DNA
- 22) Human karyotype
- 23) Stratification of human chromosomes
- 24) Microscopic and submicroscopic structure of chromosomes
- 25) Morphology of chromosomes
- 26) Chromosome staining
- 27) Metaphase detection techniques
- 28) Techniques of molecular cytogenetics
- 29) Structural, numerical aberrations
- 30) Translocations
- 31) Origin of aneuploidies
- 32) Trisomic syndromes

- 33) Gonosome syndromes
- 34) Structural variability of chromosomes
- 35) Molecular mechanisms of creation of structural chromosomal aberrations
- 36) Reparation mechanisms of DNA breaks
- 37) Microdeletion syndromes
- 38) Genomic imprinting
- 39) Duplication and their pathological effects
- 40) Marker chromosomes
- 41) Genetic significance of inversions
- 42) Congenital reciprocal translocations in humans
- 43) Problems of genetic studies in humans
- 44) Variability of human genome
- 45) Genetically determined pathologies in humans
- 46) Genetic counselling
- 47) Monogenic diseases
- 48) Autosomal dominant inheritance and human diseases
- 49) Autosomal recessive inheritance and human diseases
- 50) Genetic basis of cystic fibrosis
- 51) Examples of X-linked recessive inheritance diseases in humans
- 52) Development of human oncogenetics
- 53) The origin of the Philadelphia chromosome
- 54) The role of cytogenetic testing in oncology
- 55) Clonal CHAs in tumors
- 56) Genome destabilization and the multistep process of tumor formation
- 57) Protooncogens vs tumor suppressor genes
- 58) Chromosomal aberrations in tumors - basic classification
- 59) Numerical CHAs in tumor diseases
- 60) Translocations and tumors
- 61) Translations associated with creation of chimeric proteins
- 62) Translocations associated with protooncogen activation
- 63) Involvement of *IgH* locus 14q32 in cancer diseases
- 64) Chromosomal deletions in the process of carcinogenesis
- 65) Knuston hypothesis
- 66) Aberrations with gain of genetic material
- 67) Double minute chromosomes

- 68) Chromothripsis and tumor diseases
- 69) Prognostic changes of chromosomal aberrations associated with negative prognosis in hematologic malignancies
- 70) Prognostic changes of chromosomal aberrations associated with negative prognosis of solid tumors
- 71) Personalized medicine and genetics
- 72) Assisted human reproduction (AHR)
- 73) Genetic causes of male and female infertility
- 74) Methods of AHR
- 75) IVF cycle, Chromosomal aberrations in embryos
- 76) Preimplantation genetic analyses
- 77) Embryo biopsy difference
- 78) Vitrification
- 79) Problems of PGA
- 80) Karyomapping
- 81) Massive parallel sequencing technology (MPS) in IVF