**Theme "****Medical genetics. Medical and genetic counseling. Genetic research methods. Pedigree methodology. Hereditary metabolic diseases. Chromosomal diseases. Phacomatosis."**

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| **Вид** | **Код** | **Текст названия трудовой функции/ вопроса задания/ вариантов ответа** |
| Ф |   |   |
|   |   |   |
| В | 001 | **Genes of haploid chromosomes is called:** |
| О | А | Genome |
| О | Б | Genotype |
| О | В | Gene pool |
| О | Г | Phenotype |
|   |   |   |
| В | 002 | **A set of chromosomes of a somatic cell, characterized by a number, size, shape, is called:** |
| О | А | Karyotype |
| О | Б | Genotype |
| О | В | Gene pool |
| О | Г | Gene drift |
|   |   |   |
| В | 003 | **The rotation of the chromosome is called:** |
| О | А | Inversion |
| О | Б | Erasure |
| О | В | lack of chromosomes |
| О | Г |   |
|   |   |   |
| В | 00 4 | **From the following choices, choose the number of autosomes in human somatic cells:** |
| О | А | 44 |
| О | Б | 46 |
| О | В | 42 |
| О | Г | 48 |
|   |   |   |
| В | 00 5 | **From the following choices, choose the set of genes which determines one of the genetical characteristics of an individual:** |
| О | А | Genotype |
| О | Б | Phenotype |
| О | В | hhayi le mpendulo, bheka eduze |
| О | Г | Moribida |
|   |   |   |
| В | 00 6 | **From the following choices, choose the indicator of gene expression frequency (the proportion of individuals carrying a particular variant (or allele) of a gene (the genotype) that also express an associated trait (the phenotype):** |
| О | А | Penetrance |
| О | Б | Expressiveness |
| О | В | Dominance |
| О | Г | Mutation |
|   |   |   |
| В | 00 7 | **From the following choices, choose term meant the phenotypic manifestation (the degree to which a phenotype is expressed by individuals having a particular genotype):**  |
| О | А | expressivity |
| О | Б | Penetrance |
| О | В | Uniformity |
| О | Г | Mitosis  |
|   |   |   |
| В | 00 8 | **Genetical symptoms related to gender related to: (there’s no sexism in science, sorry)** |
| О | А | Gonosome |
| О | Б | Autosomes |
| О | В | Nucleic acids |
| О | Г | Enzymes |
|   |   |   |
| В | 00 9 | **The cytogenetic method is defined as** |
| О | А | Sex Chromatin (Barr body) |
| О | Б | Chorionic biopsy |
| О | В | Fetoprotein |
| О | Г | Karyotype |
|   |   |   |
| В | 0 10 | **Classification of monogenic diseases** |
| О | А | All answers correct |
| О | Б | autosomal dominant |
| О | В | autosomal recessive |
| О | Г | related to Gonosome |
|   |   |   |
| В | 0 11 | **A person has brothers and sisters, by using the pedigree analysis, choose the correct answer:** |
| О | А | Siblings |
| О | Б | Descendants |
| О | В | Proband |
| О | Г | ngibonge kamuva ngalo mbuzo |
|   |   |   |
| В | 012 | **Pedigree method is :**  |
| О | А | Genealogical method |
| О | Б | Cytogenetic method |
| О | В | Dermatology method |
| О | Г | immunogenetics method |
|   |   |   |
| В | 013 | **To study the description of the skin of the fingers** **using the following method** |
| О | А | dermatography |
| О | Б | Statistical method |
| О | В | in the bbbeeeginnnnning |
| О | Г | Biochemical method |
|   |   |   |
| В | 0 14 | **Changes in the structure of DNA is mutations of:** |
| О | А | Genes |
| О | Б | Chromosomes |
| О | В | Genomics |
| О | Г | Geometric |
|   |   |   |
| В | 0 15 | **Mutations** **leading to changes in the number, size and organization is a :** |
| О | А | Chromosomal mutation |
| О | Б | Genomic mutation |
| О | В | Geometric mutation |
| О | Г | Genotypic mutation |
|   |   |   |
| В | 016 | **The specific part of a chromosome with a gene is called** |
| О | А | locus |
| О | Б | Allele |
| О | В | Codon |
| О | Г | The code |
|   |   |   |
| В | **017** | **Down's disease is characterized by the following number of chromosomes** |
| О | А | 47 |
| О | Б | 42 |
| О | В | 45 |
| О | Г | 46,653x10 kg + 2 Samsa i sasitki |
|   |   |   |
| В | **018** | **In Turner's syndrome, it is a genetic abnormality of:** |
| О | А | Sexual chromosomes |
| О | Б | Homochromosomes |
| О | В | Heterochromosomes |
| О | Г | autosomes |
|   |   |   |
| В | 0 19 | **Klinefelter syndrome characterized in humans by an additional chromosome, which one?** |
| О | А | XXY |
| О | Б | Xy |
| О | В | XX |
| О | Г | X0 |
|   |   |   |
| В | **0 20** | **What syndrome is characteristic of Down's syndrome** |
| О | А | Down syndrome |
| О | Б | Syndrome of Jacob Zuma |
| О | В | Edwards syndrome |
| О | Г |  |
|   |   |   |
| В | 0 21 | **Normally, the human karyotype corresponds to** |
| О | А | 46 |
| О | Б | 42 |
| О | В | 45 |
| О | Г | 47 |
|   |   |   |
| В | 0 22 | **A skin fold on the neck in newborns is characteristic of** |
| О | А | Down syndrome |
| О | Б | Edwards syndrome |
| О | В | Klinefelter syndrome |
| О | Г | Patau syndrome |
|   |   |   |
| В | 0 23 | **The  following abnormal development  : a small  mouth; overlapping fingers, is characteristic of :**    |
| О | А | Edwards syndrome |
| О | Б | Cri du chat syndrome |
| О | В | Klinefelter syndrome |
| О | Г | Turner's syndrome |
|   |   |   |
| В | 0 24 | **The clouding of the cornea, a holoprosencephaly , hypotelorism, Division lip and palate, hydronephrosis, valvular dysplasia, polydactyly are characteristic of** |
| О | А | Patau syndrome |
| О | Б | Down syndrome |
| О | В | Edwards syndrome |
| О | Г | Klinefelter syndrome |
|   |   |   |
| В | 025 | **infantilism syndrome, craniofacial dysmorphia (triangular face, hypoplasia of the maxilla), absence of menstruation with underdevelopment of secondary sexual characteristics are characteristic of:** |
| О | А | Turner's syndrome |
| О | Б | Down syndrome |
| О | В | Edwards syndrome |
| О | Г | Klinefelter syndrome |
|   |   |   |
| В | 0 26 | **Mental retardation with increased secretion of female sexual hormones is characteristic of:** |
| О | А | Klinefelter syndrome |
| О | Б | Patau syndrome |
| О | В | Shereshevsky-Turner syndrome |
| О | Г | Cri du chat syndrome |
|   |   |   |
| В | 027 | **Among the types of inheriting genetic abnormality transmission to the next generation , which one of the following is systematic:** |
| О | А | Autosomal dominant transmission |
| О | Б | Autosomal recessive transmission |
| О | В | Autosomal dominant X-linked transmission |
| О | Г | Autosomal X- linked recessive transmission |
|   |   |   |
| В | 028 | **Among the types of inheriting genetic abnormality transmission to the next generation, which one of the following is not systematic** |
| О | А | Autosomal recessive transmission |
| О | Б | Autosomal dominant transmission |
| О | В | Autosomal dominant X-linked transmission |
| О | Г | Autosomal X- linked recessive transmission |
|   |   |   |
| В | 0 29 | **Phenylketonuria is characterized by the following type of inheritance** |
| О | А | Autosomal recessive transmission |
| О | Б | Autosomal dominant transmission |
| О | В | Autosomal X- linked recessive transmission |
| О | Г | autosoma recessive X-linked transmission |
|   |   |   |
| В | 0 30 | **Determination of the nucleotide sequence of DNA** |
| О | А | Sequencing |
| О | Б | Transcription |
| О | В | Cloning |
| О | Г |   |
|   |   |   |
| B | 031 | **The presence of just a single chromosome of a pair of chromosomes is called** |
| О | А | Monosomy |
| О | Б | Down's syndrome |
| О | В | Aneuploid |
| О | Г | Polyplodia |
|   |   |   |
| В | 032 | **Partial monosomy include** |
| О | А | Cri du chat syndrome |
| О | Б | Klinefelter syndrome |
| О | В | Down syndrome |
| О | Г | Turner's syndrome |
|   |   |   |
| В | 033 | **The "coffee" spots on the skin are characteristic of** |
| О | А | Neurofibromatosis type I   |
| О | Б | Neurofibromatosis type II   |
| О | В | Tuberous sclerosis |
| О | Г |   |
|   |   |   |
| В | 0 34 | **The central and vertebral form of phacomatosis disease is characteristic of** |
| О | А | Neurofibromatosis type II   |
| О | Б | Neurofibromatosis type I   |
| О | В | Basilar vertebral neurofibromatosis |
| О | Г | Neurofibromatosis by Said Jamani |
|   |   |   |
| В | 0 35 | **Tuberous sclerosis refers to**  |
| О | А | Phacomatosis |
| О | Б | Partial monosomy |
| О | В | Genomic mutations with abnormal number of sex chromosomes |
| О | Г | Genomic mutations with abnormal   number of autosomes |
|   |   |   |
| В | 0 36 | **Down syndrome:** |
| О | А | Genomic mutations with abnormal number of autosomes |
| О | Б | Partial monosomy |
| О | В | Genomic mutations with abnormal number of sex chromosomes |
| О | Г |  Genomic mutations with abnormality in the number of transmissible sex chromosomes |
|   |   |   |
| В | 0 37 | **Trisomy for the 18th chromosomes is characteristic of:** |
| О | А | Edwards syndrome |
| О | Б | Down syndrome |
| О | В | Patau syndrome |
| О | Г |  |
|   |   |   |
| В | **0 38** | **Tuberous sclerosis is characteristic** |
| О | А | Autosomal recessive transmission |
| О | Б | Autosomal dominant transmission |
| О | В | Autosomal X- linked recessive transmission |
| О | Г | Autosomal transmission recessive X-linked |
|   |   |   |
| В | **0 39** | **Hamartomas ( Cowden's disease ) occur at** |
| О | А | Tuberous sclerosis |
| О | Б | Neurofibromatosis type I   |
| О | В | Neurofibromatosis type II   |
| О | Г |   |
|   |   |   |
| В | 0 40 | **Physical mutagens include** |
| О | А | Ionizing radiation |
| О | Б | Pesticides |
| О | В | Virus |
| О | Г | Oxidizing agents |
|   |   |   |