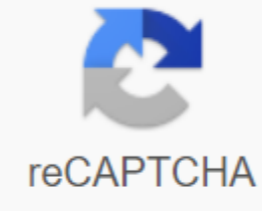


Human karyotype form answers



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A karyotype depicts the number, size and possible anomalies of chromosomes in an organism. Learning objectivesWrite a normal human karyotype and discuss the various anomalies that can be detected using this technique Isolation and microscopic observation of chromosomes forms the basis of cytogenetics and is the main method by which doctors detect chromosomal abnormalities in humans. A karyotype is the number and appearance of chromosomes. To get a view of an individual's karyotype, cytologists photograph chromosomes and then cut and paste each chromosome into a graph, or karyogram, also known as an ideogram. In a given species, chromosomes can be identified by their number, size, centromer position, and pattern of bands. In a human karyotype, autosomes or body chromosomes (all non-sexual chromosomes) are generally organized in approximate order of size from the largest (chromosome 1) to the smallest (chromosome 22). However, chromosome 21 is actually shorter than chromosome 22. This was discovered after the naming of Down syndrome as trisomy 21, reflecting how this disease derives from the possession of an extra chromosome 21 (three in total). Not wanting to change the name of this important disease, chromosome 21 retained its numbering, despite describing the shorter set of chromosomes. X and Y chromosomes are not autosomes and are referred to as sexual chromosomes. Chromosomal weapons projecting from both ends of the centromere can be designated as short or long, depending on their relative lengths. The short arm is abbreviated p (for petite), while the long arm is abbreviated q (because it follows p in alphabetical order). Each arm is further divided and indicated by a number. Using this naming system, positions on chromosomes can be described consistently in scientific literature. Although Mendel is referred to as the father of modern genetics, he has performed his experiments with none of the tools that today's geneticists routinely employ. One of these powerful cytological techniques is karyotyping, a method in which traits characterized by chromosomal abnormalities can be identified by a single cell. To observe an individual's karyotype, a person's blood cells (such as white blood cells) are first collected from a blood sample or other tissues. In the laboratory, isolated cells are stimulated to begin to actively divide. A chemical called colchicine is then applied to cells to stop condensed chromosomes in metafaction. The cells are then inflated using a hypotonic solution so that the chromosomes spread. Finally, the sample is in a fixative and applied to a slide. The geneticist then stains the chromosomes with one of the different dyes to better visualize the distinct and reproducible bandaging patterns of each chromosomal pair. As a result of staining, chromosomes are Microscopy. A common choice of spots is the Giemsa stain. Giemsa staining results in about 400-800 bands (of tightly rolled DNA and condensed proteins) arranged along all 23 pairs of chromosomes. An experienced geneticist can identify each chromosome based on its characteristic wrapping pattern. In addition to wrapping models, chromosomes are further identified based on the size and location of the centromere. To obtain the classical representation of the karyotype in which homologous pairs of chromosomes are aligned in numerical order from longest to shortest, the geneticist obtains a digital image, identifies each chromosome, and manually arranges chromosomes in this model. Figure : (/PageIndex{1}): A human karyotype: this karyotype is of a male human being. Note that homologous chromosomes are the same size and have the same positions as the centromere and the same band patterns. A human female would have a chromosomal pair XX instead of the XY pair shown. In its most basic form, karyotype can reveal genetic abnormalities in which an individual has too many or too few chromosomes per cell. Examples of this are Down syndrome, which is identified by a third copy of chromosome 21, and Turner syndrome, which is characterized by the presence of only one X chromosome in women instead of the normal two. Geneticists can also identify large deletions or DNA insertions. For example, Jacobsen syndrome, which involves distinctive facial features as well as heart and bleeding defects, is identified by a deletion on chromosome 11. Finally, karyotype can detect translocations, which occur when a segment of genetic material breaks down from a chromosome and reconnects to another chromosome or a different part of the same chromosome. Translocation is implicated in certain types of cancer, including chronic myelology leukemia. During Mendel's lifetime, inheritance was an abstract concept that could only be deduced by performing crosses and observing the traits expressed by the offspring. By observing a karyotype, today's geneticists can actually visualize an individual's chromosomal composition to confirm or predict genetic abnormalities in offspring, even before birth. LICENSES AND ATTRIBUTES CC LICENSED CONTENT, SHARED PREVIOUSLY Curation and Revision. Provided by: Boundless.com. License: CC BY-SA: Attribution-ShareAlike CC LICENSED CONTENT, SPECIFIC ATTRIBUTION hemizygous. Provided by: Wiktionary. Located in: . License: CC BY-SA: Attribution-ShareAlike OpenStax College, Biology. October 16, 2013. Supplied by: OpenStax CNX. Located in: . License: CC BY: Attribution Robert Bear and David Rintoul, Extensions of Inheritance. October 31, 2013. Supplied by: OpenStax CNX. Located in: . License: CC BY: Attribution Attribution Biochemistry/Chromosomes. Provided by: Wikibooks. 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