

Οργάνωση, Μελέτη και Ανάλυση Καρυοτύπου

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ΒΙΟΛΟΓΟΙ

Η κατασκευή, η μελέτη, η ανάλυση του καρυοτύπου

- Ο καρυότυπος αποτελεί ένα εργαλείο που ακόμα και σήμερα, αν και υπάρχουν πιο εξελιγμένες τεχνικές ανάλυσης όπως η FISH (fluorescent in situ hybridization) ή τα χρωμοσωμικά microarrays, χρησιμοποιείται τόσο κατά τον προγεννητικό έλεγχο αλλά και σε ενήλικες.
- Εκτός από την τεχνική περιγραφή της μεθόδου, θα πρέπει να γίνονται περαιτέρω προεκτάσεις που να συνδέουν τον αφανή μικρόκοσμο του γενετικού υλικού με τον φανερό μακρόκοσμο του φυσικού κόσμου:
 - σύνδεση καρυοτύπου με γονίδια, αλληλόμορφα → γονότυπο και φαινότυπο,
 - συσχέτιση καρυοτύπου με γεγονότα μείωσης που έχουν προηγηθεί κατά την παραγωγή των γαμετών,
 - ανάλυση καρυοτύπου όχι μόνο ως προς τη χρωμοσωμική σύσταση αλλά και τα συμπτώματα που επιφέρει – αν επιφέρει - η κάθε διαταραχή ή σύνδρομο,
 - συζήτηση για τα όρια της τεχνικής, όπως για παράδειγμα το γεγονός πως ο καρυότυπος μελετά εξαιρετικά συμπυκνωμένη χρωματίνη και επομένως μικρά τμήματα DNA που είναι εξαιρετικά πολυμορφικά ή ποικίλα μεταξύ των περισσότερων ανθρώπων είναι σχεδόν αδύνατον να μελετηθούν με αυτή τη τεχνική.

... για όλα τα παραπάνω #1



- Karyotype Virtual Lab
- <https://learn.genetics.utah.edu/content/basics/karyotype/>

Make a Karyotype

A karyotype is an organized profile of a person's chromosomes. Two chromosomes specify sex, XX for female and XY for male. The rest are arranged in pairs, numbered 1 through 22, from largest to smallest. This arrangement helps scientists quickly identify chromosomal alterations that may result in a genetic disorder.

To make a karyotype, scientists take a picture of the chromosome from one cell, cut them out, and arrange them using size, banding pattern, and centromere position as guides.

[View a printable PDF of a karyotype](#)

Matching up Chromosomes in a Karyotype - Try it Yourself!

Instructions: Click on a chromosome in the pool on the left and drag it next to its partner in the karyotype on the right. Turning the "Hints" on provides an outline of the matching chromosome.

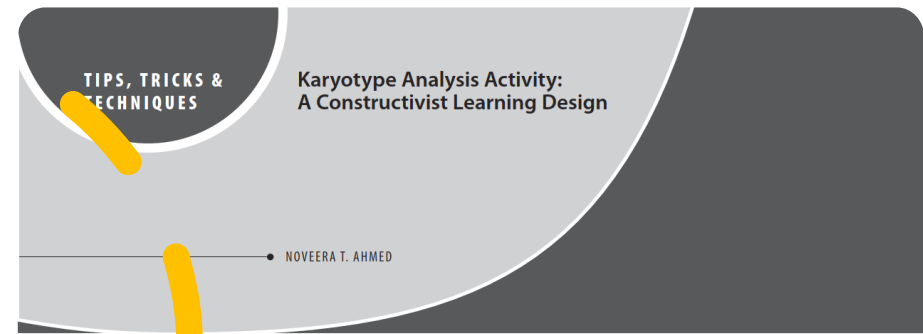
Hints On



... για όλα τα παραπάνω #2α

- Noveera T. Ahmed; Karyotype Analysis Activity: A Constructivist Learning Design. *The American Biology Teacher*, 1 January 2015; 77 (1): 63–67. doi: <https://doi.org/10.1525/abt.2015.77.1.9>

- Προσαρμογή ενός φύλλου εργασίας



ABSTRACT

This classroom activity is based on a constructivist learning design and engages students in physically constructing a karyotype of three mock patients. Students then diagnose the chromosomal aneuploidy based on the karyotype, list the symptoms associated with the disorder, and discuss the implications of the diagnosis. This activity is targeted at undergraduates in a nonmajors genetics course, but the goals align with AP Biology Big Idea 3 and Next Generation Science Standards HS-LS3. The activity illustrates the relationship between genotype and phenotype, reinforces the chromosome theory of inheritance, and includes mapping of meiotic nondisjunction events.

Key Words: Karyotype; chromosomes; aneuploidy; constructivist learning design; chromosome theory of inheritance; genotype; phenotype; meiosis; meiotic nondisjunction; AP Biology; Next Generation Science Standards.

Karyotype analysis is the visualization of an individual's chromosomes for the purpose of determining chromosome size and number. This karyotype analysis activity was designed for undergraduates in a nonmajors genetics course but is appropriate for high school students because it reinforces some basic themes of genetics, including the chromosome theory of inheritance, the relationship between genotype and phenotype, and meiosis and meiotic nondisjunction as sources of new genetic information.

The activity is modeled after a constructivist learning design (CLD) outlined by Gagnon and Collay (2001) and consists of six elements: Situation, Groupings, Bridge, Questions, Exhibit, and Reflections. Students work in groups to complete the karyotype of three mock patients, diagnose their disorder, and list symptoms associated with the aneuploidy identified. Students are also asked what meiotic nondisjunction events could have led to aneuploidy in these patients. The activity concludes with questions that guide

students in synthesizing this information and discussing the ethics of prenatal diagnosis.

○ Purpose of Karyotype Analysis

As the first element of the CLD, Situation is an overview of the entire activity and establishes its purpose (Gagnon & Collay, 2001, pp. 17–34). The purpose of karyotype analysis is to look at the gross anatomical structures of the chromosomes of an individual. This can allow us to visualize the number and size of each chromosome found within each cell. This test can be performed using almost any tissue from an adult or amniotic fluid from a developing fetus (<http://www.nlm.nih.gov/medlineplus/ency/article/003935.htm>). Although this technology is a century old, it is still accepted by the American College of Obstetricians and Gynecologists (ACOG) as a definitive diagnostic test for identifying aneuploidy in a developing fetus and, unlike newer technologies, can be used to identify triploidy or balanced translocation events, where genetic information has moved from one location to another without any loss of information (ACOG Committee Opinion nos. 545 and 581).

○ Activity Preparation

This portion of the activity is rooted in the CLD element Groupings, in which students work together in small groups, help each other, and generate shared meaning (Gagnon & Collay, 2001, pp. 35–50). Students are divided into groups of three or four students and supplied with scissors and tape. They should generate one set of karyotypes per group. This activity requires 60–90 minutes to complete, depending on the amount of pre-activity and Reflection discussion that occurs.

The CLD element Bridge involves reviewing what your students should know and linking what they know with what they are going to

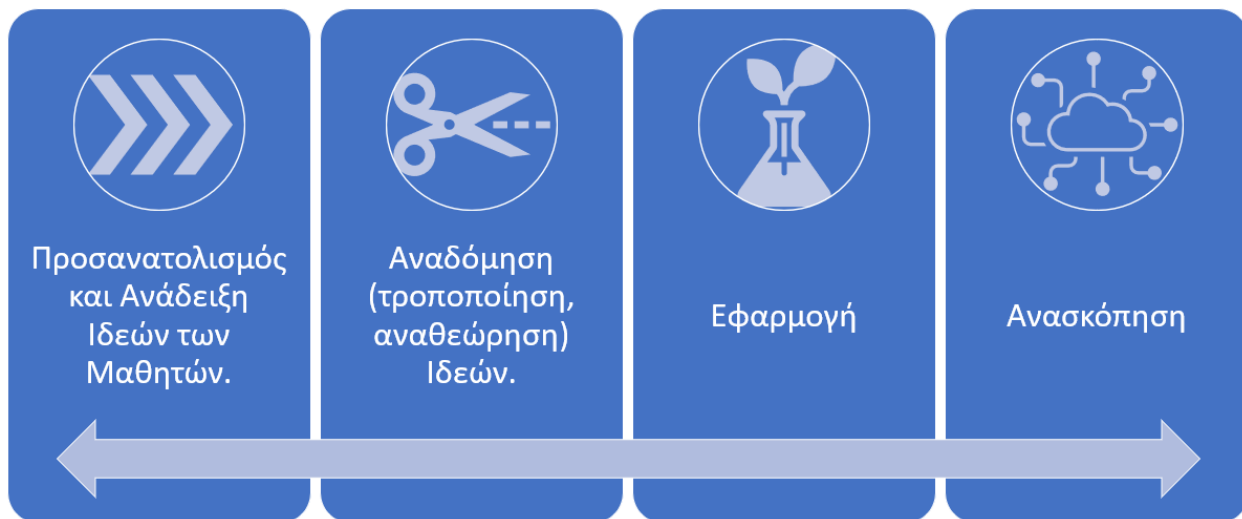
Karyotype analysis is the visualization of an individual's chromosomes for the purpose of determining chromosome size and number.

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... για όλα τα παραπάνω #2β

... επιλέχθηκε αυτή και όχι κάποια άλλη από τις πολλές διαθέσιμες

- αναπτύσσει και εφαρμόζει ένα μοντέλο εποικοδομητιστικής θεωρίας διδασκαλίας και μάθησης



Constructivist Learning Design (CLD) / Gagnon and Collay (2001)

- Situation: Σκοπός / Γενική Περιγραφή
- Groupings: Ομάδες / Συνεργασία
- Bridge: Πρότερη γνώση / Καινούρια γνώση
- Questions: Ανάδειξη γνώσεων μέσω Ανακάλυψης / Συλλογή & Ερμηνεία Δεδομένων
- Exhibit: Παρουσίαση / Συζήτηση στην Ολομέλεια
- Reflections: Αναστοχασμοί / Προεκτάσεις

Gagnon, G.W., Jr. & Collay, m. (2001). *Designing for Learning: Six Elements in Constructivist Classrooms*. Thousand Oaks, CA: Corwin Press.

Ευχαριστούμε που μας παρακολουθήσατε!!!!

