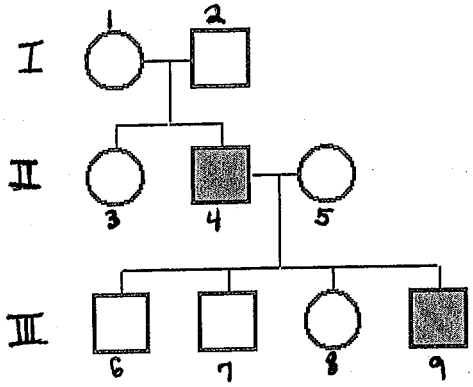
Pedigree Analysis

Introduction

A pedigree is a diagram of family relationships that uses symbols to represent people and lines to represent genetic relationships. These diagrams make it easier to visualize relationships within families, particularly large extended families. Pedigrees are often used to determine the mode of inheritance (dominant, recessive, etc.) of genetic diseases. A sample pedigree is below.



In a pedigree, squares represent males and circles represent females. Horizontal lines connecting a male and female represent mating. Vertical lines extending downward from a couple represent their children. Subsequent generations are therefore written underneath the parental generations and the oldest individuals are found at the top of the pedigree.

If the purpose of a pedigree is to analyze the pattern of inheritance of a particular trait, it is customary to shade in the symbol of all individuals that possess this trait.

In the pedigree above, the grandparents had two children, a son and a daughter. The son had the trait in question. One of his four children also had the trait.

In the exercises below, assume that the trait in question is a genetic disease or abnormality. We will learn patterns of inheritance that have the following modes of inheritance:

autosomal dominant
autosomal recessive
X-linked recessive

Questions

1. circle = females square = males
2. shaded = affected w/ trait
3. empty = not affected (appearance) w/ trait
4. Roman Numerals represent generation number

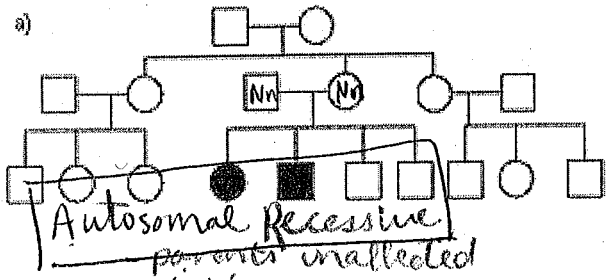
Human Pedigree Analysis Problem Sheet

There are a number of different types of human pedigrees that you may encounter, however, there are only a few different modes of inheritance that you will need to be familiar. The following pedigrees show you different examples of human traits that can be traced through generations. See if you can identify the modes of inheritance, and answer any questions directly related to each pedigree. While you are working on this, keep the following clues in mind:

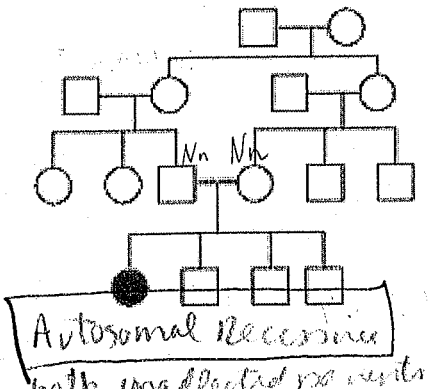
Clues for Autosomal Inheritance	
Recessive • individual expressing trait (can have) has 2 normal parents • two affected parents can not have an unaffected child	Dominant • every affected person has at least one affected parent • each generation will have affected individuals
Clues for Sex-linked Inheritance	
Recessive - X linked • no father-to-son transmission • predominantly males affected • trait my skip generations	

1. For each of the pedigrees below, identify the mode of inheritance and provide at least 2 reasons for your choice.

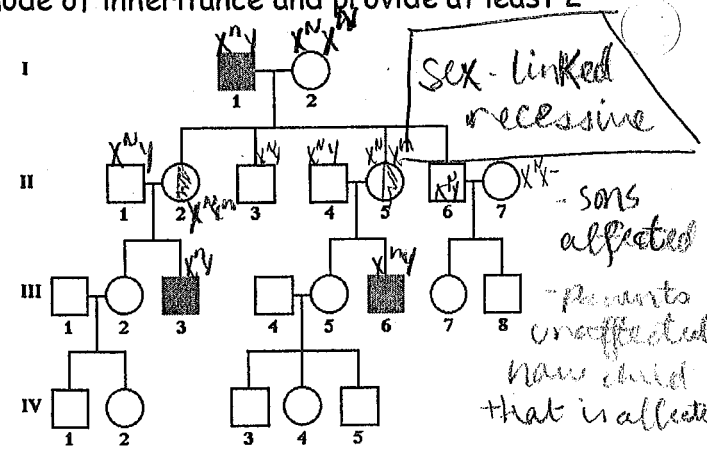
a)



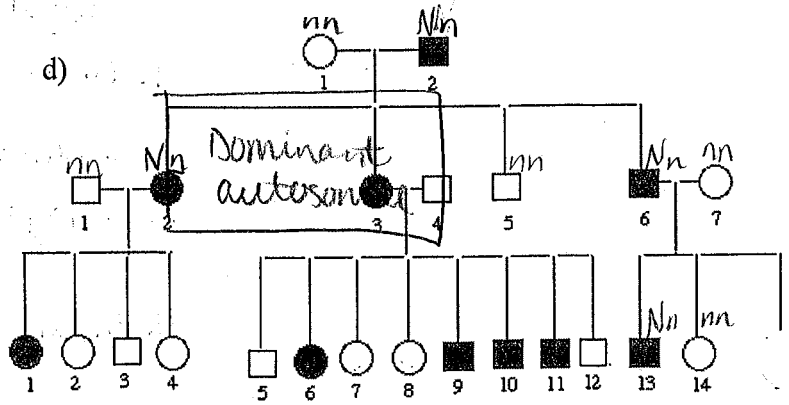
b)



c)

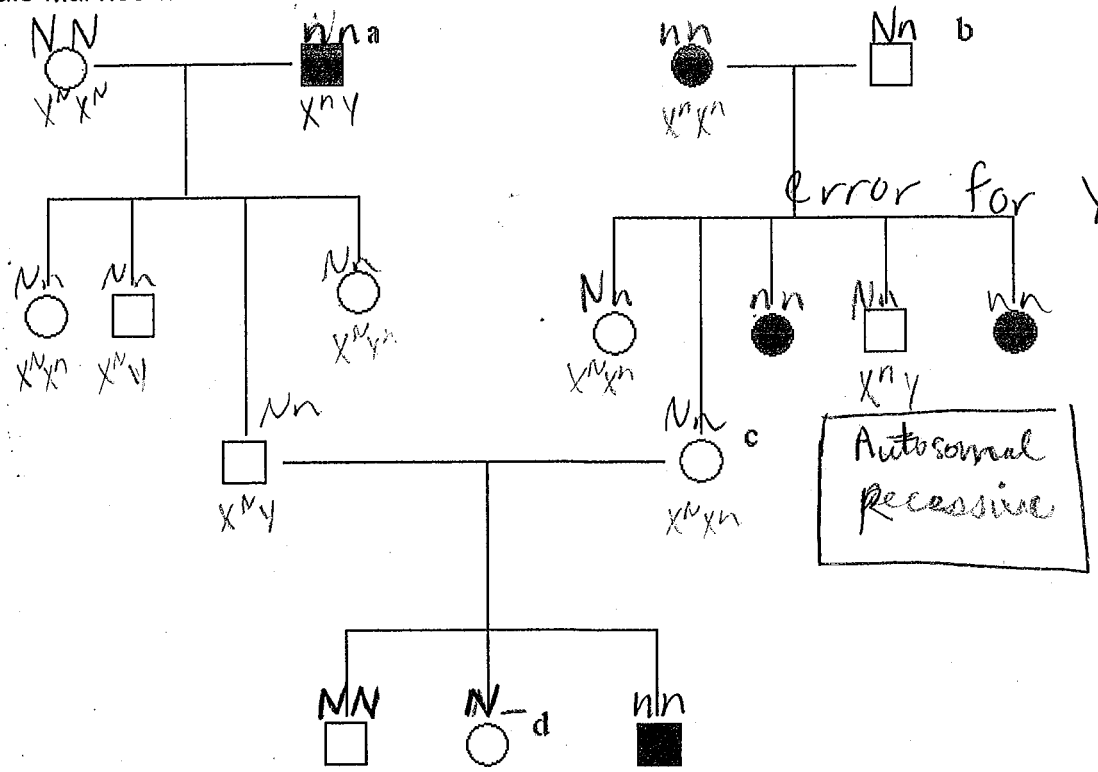


d)

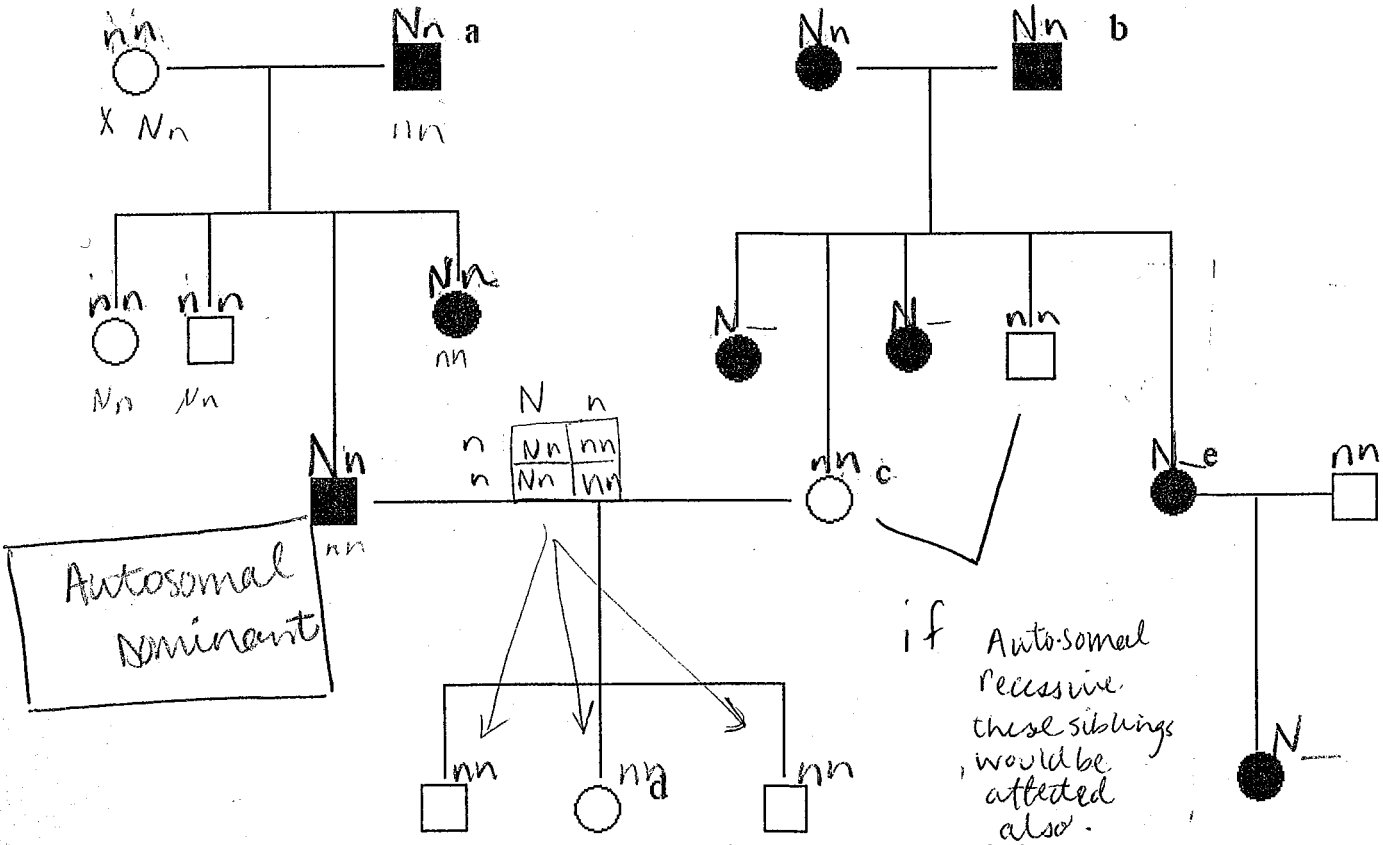


ea. generation has person
equal # 9 to 07 w/ trait

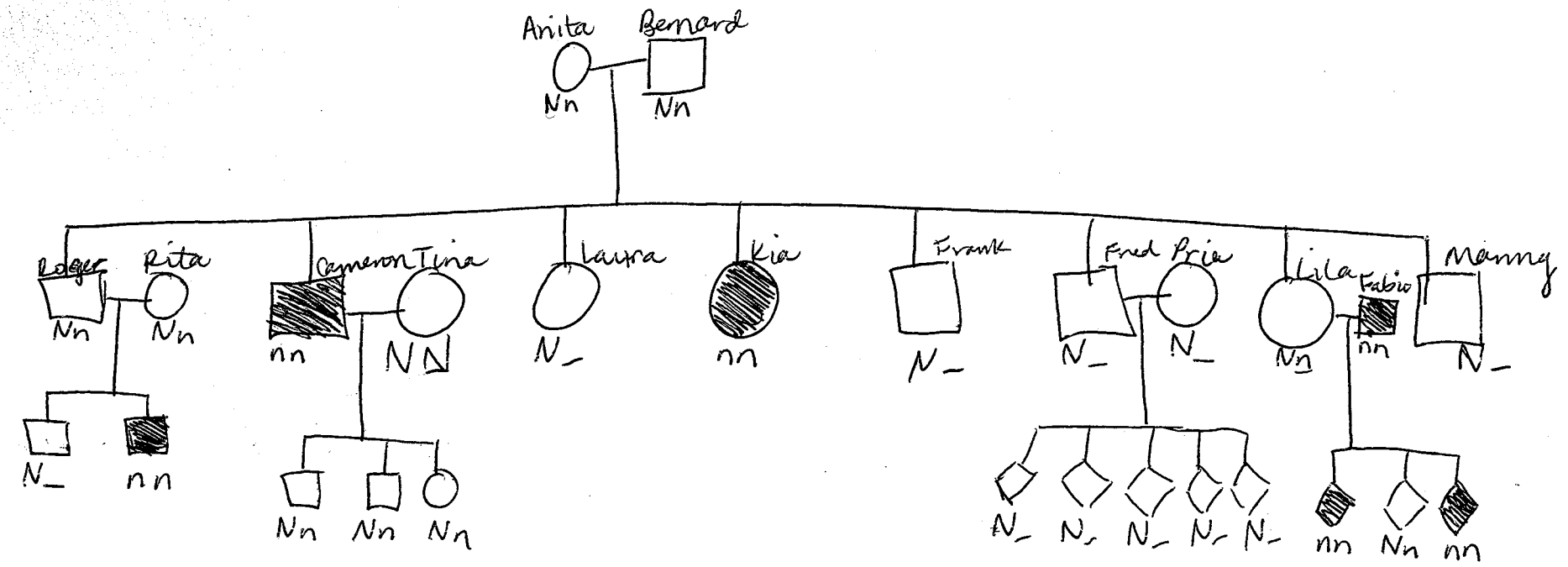
2. Below is a pedigree for an inherited lung disease. Provide the genotypes of ALL ~~each~~ of the individuals ~~marked with lower case letters~~.



3. Below is a pedigree for an inherited brain disease. Provide the genotypes of each of the individuals marked with lower case letters.



4. ~~Use the Pedigree for Trait A to determine the genetic basis of this trait.~~



Sickle cell disease

Complete dominance - there are instances, in which two parents have a child (ren) with sickle cell yet they themselves do not have it. Seems to be an equal distribution of males to females affected so rules out likelihood of sex-linked inheritance.