

Group 19: Pedigree Analysis

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 $Course\ Modelling\ Project$

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Abstract

When given a pedigree chart, our model will track the family members to their previous generations with all possibilities triats. As they are affected by a trait to determine if a trait is: dominant or recessive, and autosomal or X-linked.

A pedigree chart is a diagram that documents how a genetic trait is pass on in a family, from one generation to another.

A trait can be determined as either dominant or recessive:

- Dominant traits only need one affected allele to be expressed.
- Recessives traits are only expressed when both alleles are affected. Autosomal or x-linked:
- Autosomal traits are located on all the chromosomes except the X or Y chromosome
- X-linked traits are located on the X chromosomes; are more likely to affect males

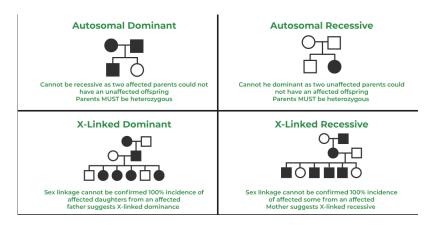


Figure 1: Image from https://www.geeksforgeeks.org/pedigree-analysis/

Propositions

- Drafts
 - Characteristics of each family member
 - * F(i), is true if a family member at position i is female
 - * A(i): is true if a family member at position i is affected
 - * R(i): is true if a family member at position i is a blood relative
 - Relationship between family members
 - * C(i,j,k): is true if a family member at position i is the child of family members at position j and k
 - * S(i,j): is true if a family member at position i is the sibling of someone at position j
 - Inheritance Pattern
 - * M(g): is true if more male family members are affected in a generation
 - Inheritance mode of trait passed down
 - * R: is true if a disease is recessive and false if it is not
 - * X: is true if a disease is x linked and false if it is autosomal

Constraints

- Drafts:
 - Two family members are siblings if and only if they have the same parents:

$$* C(a, c, d) \land C(b, c, d) \iff S(a, b)$$

$$* \neg (\exists b, c \ C(a, b, c)) \lor R(a) \iff (\exists b, c \ C(a, b, c))$$

 If both parents of an affected family member are unaffected, then the disease is recessive

$$* \ (A(a) \land C(a,b,c)) \land (\neg A(b) \land \neg A(c)) \implies R$$

- There must be one male and one female parent

$$* C(i, a, b) \iff (F(a) \land \neg F(b)) \lor (\neg F(a) \land F(b))$$

- Parents cannot be blood relatives

$$* C(i, a, b) \iff (R(a) \land \neg R(b) \lor (R(b) \land \neg R(a))$$

- To do:
 - If there are more males in a generation than females the M(g) is true
 - If M(g) is true for at least half of the generations then, X is true

Model Exploration

- building pedigree
 - 1. First draft: 2d array based on number of gen and the number of ppl in last gen

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per_last_cont = input/thanker of people in each generation, starting from the last generation

per_last_cont

per_last_cont
```

- Issue with model: too restrictive, limits the amount of generations and how many children the older generations have
- 2. Second Draft: Recursive 2d dictionary that build up a person, then siblings then parents

```
def create_family_tree(person_id, num_siblings, parents, family_tree, generation=0):
    # Check if the person already exists in the tree
if person_id not in family_tree:
    # Initialize the person's data
    family_tree(person_id) = {
        "openeration"; generation,
        "siblings": set(),
        "parents": [],
        "spouse": None
    }

# Add siblings based on num_siblings count
    siblings = []
    for i in range(num_siblings):
    sibling_id = []
    for i in range(num_siblings):
    sibling_id not in family_tree:
        create_family_tree(sibling_id), # (], family_tree, generation)
    siblings.appen(sibling_id)
    family_tree(person_id) ["siblings"].add(sibling_id)
    family_tree(sibling_id) ["siblings"].add(person_id)

# Add parents if provided
if parents:
    parentl not in family_tree:
        # Recursively add each parent and establish spousal relationship create_family_tree(parent1, # (], family_tree, generation + 1)

if parent2 not in family_tree:
        create_family_tree(parent2, # (], family_tree, generation + 1)

# Link the parents as spouses and set them as the person's parents family_tree(parent1["spouse"] = parent2
    family_tree(parent2[) ["spouse"] = parent1
    family_tree(parent2[) ["spouse"] = parent1
    family_tree(parent2[) ["spouse"] = parent1
    family_tree(parent2[) ["spouse"] = [parent1, parent2]

# Assign siblings' parents to the same parents
    for sibling_id in siblings:
        family_tree(sibling_id) ["parents"] = [parent1, parent2]
```

- Issue with model: the function does not follow the format of our propositions and constraints
- 3. Current draft: a dictionary of individual family members with their IDs as the key. Assign values to their keys by giving them characteristics (affected, gender, blood relation) using Char propositions. Then create a dictionary using for immediate families (siblings+parents) by linking individual family members using Rel propositions, then create a recursive function that builds the pedigree up using the dictionaries

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Clionary to store immediate family [siblings+parents)
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ctionary to store individual family member (id, chars)
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                          # Helper function to add a person to a list without duplicate
def add_to_list(person_id, list_name):
    person = PEOVE[person_id]
    if person not in sfamily(list_name):
        ifamily(list_name).append(person)
                        et:

# Three arguments case: first goes to siblings, second and third go to parents
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           Situitinethod gef create_peigree(): PEDIGREE = O = 0 \ \mbox{ Information} PEDIGREE = O = 0 \ \mbox{ Citionary to store generational information}
                               # Divide PEDPLE into blood relatives and non-blood relatives
list_BR = [id for id, details in PEDPLE.items() if details[i] == i] # Blood relatives
list_S = [id for id in PEDPLE if id not in list_BR] # Non-blood relatives
                                                              regen_us = 1)
person_id in list_BR[:]: # Iterate over a copy of list_BR
person = PEOPLE[person_id]
parents = IF/MILIES[person_id].get("parents", [])
```

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Jape Proof Ideas

- A person is affected and their parents are not. This must mean the parents are carriers of the trait and the trait is recessive
- In a family, one parent is affected, and one is not. They have more than one kid but not all of them are affected. This must mean either one parent has a dominant gene and the other doesn't have this gene or the parent has two recessive genes and the other is a carrier.

Requested Feedback

 \bullet Ideas for how to implement, the constraints in our to-dos (If there are more males in a generation than females the M(g) is true & If M(g) is true for at least half of the generations then, X is true). We are trying to do this recursively but are struggling to count the amount of people and generations with only logic.