1. Use the maps to answer the questions:
   
a. In 1805 a group of European geographers were surveying in Africa. Most of them died of malaria, but their native guides survived. Why might the native Africans have survived while the Europeans died? (think about genotypes)
   
b. Why do southern African populations not have sickle cell anemia?
   
c. Why might the disease sickle cell anemia still exist if it is deadly?
Sickle Cell Anemia
Sickle Cell Anemia

• Heterozygous Advantage: https://www.youtube.com/watch?v=1fN7r0wDyMQ
Deciphering Pedigrees

Two general hints:

- If the trait SKIPS a generation it is recessive
- If the trait is close to EQUAL in males and females it is autosomal

You can also look for an affected male and observe his daughters
Deciphering Pedigrees

AUTOSOMAL RECESSIVE

[Pedigree diagram with numbered individuals and symbols indicating genetic status]
Deciphering Pedigrees

Autosomal Dominant

1

2

3

4

5

6

7

8

9

10

11

12
Deciphering Pedigrees

Pedigree 7. X-linked recessive inheritance.
Deciphering Pedigrees

Pedigree 5. X-linked dominant inheritance.
What is the pattern of inheritance?
What is the pattern of inheritance?
What is the pattern of inheritance?
What is the pattern of inheritance?

X-linked
Recessive

[Genetic diagram showing patterns of inheritance]
What is the pattern of inheritance?
What is the pattern of inheritance?

Autosomal Recessive
What is the pattern of inheritance?
What is the pattern of inheritance?

Autosomal Recessive
What is the pattern of inheritance?
What is the pattern of inheritance?

Autosomal Dominant
Deciphering Karyotypes

- Karyotypes are used to diagnose NONDISJUNCTION
Deciphering Karyotypes

- Karyotypes are used to diagnose NONDISJUNCTION
Nondisjunction

(a) Nondisjunction of homologous chromosomes in meiosis I

(b) Nondisjunction of sister chromatids in meiosis II
Trisomy 21 (Down Syndrome)

- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Upward slanting eyes
- Small ears
- Small mouth
- Wide, short hands with short fingers
- Separated joints between the bones of the skull
- Single crease in the palm of the hand
- White spots on the colored part of the eye
Klinefelter Syndrome

- Lower IQ than sibs
- Tall stature
- Poor muscle tone
- Reduced secondary sexual characteristics
- Gynaecomastia (male breasts)
- Small testes/infertility
Klinefelter Syndrome

Normal karyotype (46,XY)

Klinefelter syndrome (47,XXY)

Tall stature
Narrow shoulders
Gynecomastia
Small testes
Infertility
Turner Syndrome

- Short stature
- Low hairline
- Shield-shaped thorax
- Widely spaced nipples
- Shortened metacarpal IV
- Small fingernails
- Brown spots (nevi)
- Characteristic facial features
- Fold of skin
- Constriction of aorta
- Poor breast development
- Elbow deformity
- Rudimentary ovaries
- Gonadal streak (underdeveloped gonadal structures)
- No menstruation

Incidence of Phenotypes in Turner Syndrome

- Short stature: 100%
- Infertility: 96%
- Primary gonadal failure: 95%
- Osteoporosis: 50%
- Cubitus valgus: 45%
- Low posterior hairline: 40%
- Carbohydrate intolerance: 30-40%
- High blood pressure: 25-40%
- Short metacarpals: 35%
- High arched palate: 35%
- Structural abnormalities in kidney: 35%
- Hypothyroidism (Hashimoto thyroiditis): 35%

NIH. Electronic Citation; 2002.
Deciphering Karyotypes

- Autosomal Nondisjunction in humans

<table>
<thead>
<tr>
<th>#</th>
<th>monosomy</th>
<th>trisomy</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1p36 deletion syndrome</td>
<td>Trisomy 1</td>
</tr>
<tr>
<td></td>
<td>1q21.1 deletion syndrome</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>2q37 deletion syndrome</td>
<td>Trisomy 2</td>
</tr>
<tr>
<td>3</td>
<td></td>
<td>Trisomy 3</td>
</tr>
<tr>
<td>4</td>
<td>Wolf-Hirschhorn syndrome</td>
<td>Trisomy 4</td>
</tr>
<tr>
<td>5</td>
<td>Cri du chat 5q deletion syndrome</td>
<td>Trisomy 5</td>
</tr>
<tr>
<td>6</td>
<td></td>
<td>Trisomy 6</td>
</tr>
<tr>
<td>7</td>
<td>Williams syndrome</td>
<td>Trisomy 7</td>
</tr>
<tr>
<td>8</td>
<td>Monosomy 8p</td>
<td>Trisomy 8</td>
</tr>
<tr>
<td></td>
<td>Monosomy 8q</td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>Alfö's syndrome</td>
<td>Trisomy 9</td>
</tr>
<tr>
<td></td>
<td>Kleefstra syndrome</td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>Monosomy 10p</td>
<td>Trisomy 10</td>
</tr>
<tr>
<td></td>
<td>Monosomy 10q</td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>Jacobsen syndrome</td>
<td>Trisomy 11</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>#</th>
<th>monosomy</th>
<th>trisomy</th>
</tr>
</thead>
<tbody>
<tr>
<td>12</td>
<td>Trisomy 12</td>
<td></td>
</tr>
<tr>
<td>13</td>
<td></td>
<td>Patau syndrome</td>
</tr>
<tr>
<td>14</td>
<td></td>
<td>Trisomy 14</td>
</tr>
<tr>
<td>15</td>
<td>Angelman syndrome</td>
<td>Trisomy 15</td>
</tr>
<tr>
<td></td>
<td>Prader–Willi syndrome</td>
<td></td>
</tr>
<tr>
<td>16</td>
<td></td>
<td>Trisomy 16</td>
</tr>
<tr>
<td>17</td>
<td>Miller-Dieker syndrome</td>
<td>Trisomy 17</td>
</tr>
<tr>
<td></td>
<td>Smith-Magenis syndrome</td>
<td></td>
</tr>
<tr>
<td>18</td>
<td>Distal 18q-Proximal 18q-</td>
<td>Edwards syndrome</td>
</tr>
<tr>
<td>19</td>
<td></td>
<td>Trisomy 19</td>
</tr>
<tr>
<td>20</td>
<td></td>
<td>Trisomy 20</td>
</tr>
<tr>
<td>21</td>
<td></td>
<td>Down syndrome</td>
</tr>
<tr>
<td>22</td>
<td>DiGeorge syndrome</td>
<td>Cat eye syndrome</td>
</tr>
<tr>
<td></td>
<td>Phelan-McDermid syndrome</td>
<td></td>
</tr>
<tr>
<td></td>
<td>22q11.2 distal deletion syndrome</td>
<td>Trisomy 22</td>
</tr>
</tbody>
</table>
Deciphering Karyotypes

- Sex-linked nondisjunction in humans

### Key

<table>
<thead>
<tr>
<th>Color</th>
<th>Significance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Red</td>
<td>Lethal</td>
</tr>
<tr>
<td>Light Blue</td>
<td>Normal male phenotype</td>
</tr>
<tr>
<td>Blue</td>
<td>Klinefelter syndrome (abnormal male)</td>
</tr>
<tr>
<td>Pink</td>
<td>Polysomy X and/or Y, (abnormal male)</td>
</tr>
<tr>
<td>Pale Pink</td>
<td>Normal female phenotype</td>
</tr>
<tr>
<td>Magenta</td>
<td>Turner's syndrome (abnormal female)</td>
</tr>
<tr>
<td>Dark Pink</td>
<td>Tetrasomy X, pentasomy X, (abnormal female)</td>
</tr>
</tbody>
</table>

### Non-autosomal

<table>
<thead>
<tr>
<th></th>
<th>0</th>
<th>X</th>
<th>XX</th>
<th>XXX</th>
<th>XXXX</th>
<th>XXXXX</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td><strong>00</strong></td>
<td>X0</td>
<td><strong>XX</strong></td>
<td><strong>XXX</strong></td>
<td><strong>XXXX</strong></td>
<td><strong>XXXXX</strong></td>
</tr>
<tr>
<td>Y</td>
<td><strong>YO</strong></td>
<td>XY</td>
<td><strong>XXY</strong></td>
<td><strong>XXXY</strong></td>
<td><strong>XXXXY</strong></td>
<td><strong>XXXXXY</strong></td>
</tr>
<tr>
<td>YY</td>
<td><strong>YY</strong></td>
<td>XY</td>
<td><strong>XXYY</strong></td>
<td><strong>XXXYY</strong></td>
<td><strong>XXXXYY</strong></td>
<td><strong>XXXXXY</strong></td>
</tr>
<tr>
<td>YYY</td>
<td><strong>YYY</strong></td>
<td>XYY</td>
<td><strong>XXYY</strong></td>
<td><strong>XXXYYY</strong></td>
<td><strong>XXXXYYY</strong></td>
<td><strong>XXXXXY</strong></td>
</tr>
<tr>
<td>YYYY</td>
<td><strong>YYYY</strong></td>
<td>XYYY</td>
<td><strong>XXYYYY</strong></td>
<td><strong>XXXXYYYY</strong></td>
<td><strong>XXXXXXYY</strong></td>
<td><strong>XXXXXY</strong></td>
</tr>
<tr>
<td>YYYYY</td>
<td><strong>YYYYY</strong></td>
<td>XYYYY</td>
<td><strong>XXYYYYY</strong></td>
<td><strong>XXXXYYYYY</strong></td>
<td><strong>XXXXXXXYY</strong></td>
<td><strong>XXXXXY</strong></td>
</tr>
</tbody>
</table>

Note: The table and key illustrate the various karyotypes and their significance in sex-linked nondisjunction in humans.
Deciphering Karyotypes

• What is wrong here?
Deciphering Karyotypes

What is wrong here?

ZWK99024 KEY

1  2  3  4  5

6  7  8  9  10  11  12

13  14  15  16  17  18

19  20  21  22  X  Y
Deciphering Karyotypes

- What is wrong here?
Deciphering Karyotypes

• What is wrong here?
Deciphering Karyotypes

• To understand how karyotypes are used you and a partner will be diagnosing people using their karyotypes
Deciphering Karyotypes

• PLEASE DO NOT LOSE ANY CHROMOSOMES, BE VERY CAREFUL

• If you do, tell Mrs. Moberly IMMEDIATELY
Deciphering Karyotypes

• When dismissed:
  • Go to a lab station
  • Complete the activity
  • Put chromosomes away
  • Move to a new station

• DO NOT LOSE CHROMOSOMES