1) Match the following genes with their known roles: Use **TS** for tumor suppressor, **O** for oncogene, **R** for DNA repair and **NC** for non-cancer related.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Role</th>
</tr>
</thead>
<tbody>
<tr>
<td>P53</td>
<td>TS</td>
</tr>
<tr>
<td>Src</td>
<td>O</td>
</tr>
<tr>
<td>clotting factor 8</td>
<td>NC</td>
</tr>
<tr>
<td>Xeroderma</td>
<td>R</td>
</tr>
<tr>
<td>Prader-Willi</td>
<td>NC</td>
</tr>
<tr>
<td>Ataxia telangiectsia</td>
<td>R</td>
</tr>
<tr>
<td>gene carried by a retrovirus</td>
<td>O</td>
</tr>
<tr>
<td>normal allele turns off cell division</td>
<td>TS</td>
</tr>
</tbody>
</table>

2) Place the letters of the following terms in all the appropriate blank(s):

- **C** cytoplasmic inheritance
- **I** sex influenced trait
- **SD** sex linked dominant
- **SR** sex linked recessive
- **SL** sex limited trait
- **H** holandric trait
- **M** maternal effect

- **H** Any gene on the Y chromosome
- **SL** Lactation in mammals
- **SR** Classic hemophilia
- **I** Pattern baldness
- **H** Passed from father to all sons
- **C** Always inherited from the mother
- **SR** Testicular feminization
- **I** Dominance is sex-dependent
- **C** mitochondrial DNA defect
- **C and SR** sons of affected mother all affected
- **M** Left and right coiling in snails
- **SR** mostly affects males (humans)
- **SR & SD** Passed from affected male to all his daughters
- **M** phenotypic ratios are delayed 1 generation

3) Provide the missing information in the table below. It should include 5 different aberrant human karyotypes, four of which involve sex chromosomes:

<table>
<thead>
<tr>
<th>Syndrome Name</th>
<th># of chromosomes</th>
<th>Sex chromosomes</th>
<th>Fertile? Y/N</th>
<th>Apparent sex M/F</th>
</tr>
</thead>
<tbody>
<tr>
<td>Klinefelters</td>
<td>47</td>
<td>XXY</td>
<td>N</td>
<td>M</td>
</tr>
<tr>
<td>triplo X</td>
<td>47</td>
<td>XXX</td>
<td>Y</td>
<td>F</td>
</tr>
<tr>
<td>Turner’s</td>
<td>45</td>
<td>XO</td>
<td>N</td>
<td>F</td>
</tr>
<tr>
<td>Jacobs or XYY</td>
<td>47</td>
<td>XYY</td>
<td>Y</td>
<td>M</td>
</tr>
<tr>
<td>Trisomy 21</td>
<td>47</td>
<td>XY (here as is male)</td>
<td>N</td>
<td>M</td>
</tr>
</tbody>
</table>
4) Which of the following is/are a part of or a consequence of Lyon’s Law?
X___A Barr body is found in cells of normal females
_____Angelman syndrome
_____Color blindness affects more males than females
X___X-inactivation occurs early in embryology
_____Genomic imprinting on chromosome 15 differs in males and females
X___Females heterozygous for hemophilia vary in clotting factor levels
X___Calico coat color in cats
_____Telomeres get shorter as we age

5) Which of the following [cyto]/geno-types of maize would be male sterile?

6) A mouse heterozygous for genes R and G was repeatedly testcrossed to a mouse homozygous recessive for both genes. Only 5% of the progeny overall were recessive for both genes.

a) Show the genotypes of the two mice:

double heterozygous   ___r G___
R  g__________________testcross___r  g______________

b) Indicate the map distance between the loci for genes R and G____10____

c) Give the genotypes and frequencies expected for the other progeny.

45%   R  g
R  g
5%   R  G
r  g
45%   r  G
r  g

7. a) What is the most common viable autosomal chromosomal aberration in humans?_________Trisomy 21_________

b) List 2 factors that greatly raise the risk for a birth with this aberration.

1.age of mom   2. translocation of chromosome 21

Using letters to indicate one full set of chromosomes, differentiate between an

Autotetraploid____AAAAD_______Allohexaploid____AA, BB, DD__________
8. Mice in 2 barns had these arrangements of genes for chromosomes 1, 2 & 3

<table>
<thead>
<tr>
<th>chromosome 1</th>
<th>chromosome 2</th>
<th>chromosome 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>barn 1</td>
<td>A • B C D E</td>
<td>1 2 • 3 4 5</td>
</tr>
<tr>
<td>barn 2</td>
<td>A • B 3 4 5</td>
<td>1 2 • C D E</td>
</tr>
</tbody>
</table>

a) Show chromosome pairing for a ‘normal’ mouse from barn 1

```
A • B C D E  1 2 • 3 4 5  H I • J K L M N
A • B C D E  1 2 • 3 4 5  H I • J K L M N
```

b) Show chromosome pairing for a mouse that is a hybrid from barns 1 & 2

```
A • B C D E  1 2 • 3 4 5  H I • J K L M N
A • B 3 4 5  1 2 • C D E  H I • J M L K N
```

c) What aberrations differentiate the two mouse populations?

**Reciprocal translocation and paracentric inversion**

d) What % fertility would be expected in the hybrid mice?

\[ 0.5 \times 0.5 = 0.25 \text{ or } 25\% \text{ (each leads to 50\% reduction)} \]

9. a) Which test would be used for prenatal detection of the listed conditions? Use **K** for karyotype; **P** for PCR, **E** for enzyme assay and **S** for substance present.

- **K** Trisomy 13
- **P** Huntingtons
- **S** Sickle cell anemia
- **S** Spina bifada
- **S** Cri du chat
- **S** Tay-Sachs disease

b) List 3 simply inherited diseases where there are so many different mutant alleles that a complete DNA sequence of the gene might be required for detection.

Things like cystic fibrosis, hemophilia, Duchene muscular dystrophy where there are many known mutations in a very large gene; not sickle cell, or a trinucleotide repeat diseases where primers that flank the repeat can be used.
10. DNA analysis of a human population showed that 4% had a duplication of Amy1, a gene that functions in starch degradation. a) Calculate the expected ‘gene’ frequencies of the single and duplicated copies assuming random mating occurs. Also calculate the expected genotype frequencies.

Freq of duplicate Amy1= 0.2 = “q” so F( single copy) = 0.8 or ‘p”

The genotype frequencies thus = 64% homozygous single
32 % heterozygous (one single & 1 dupl)
4% homozygous duplicate

b) What force may become involved if the population is limited to a high starch diet?

selection

11. The following table shows the number of repeats in the alleles from 5 STR (simple tandem repeat) loci that are used in the CODIS database (they are also called SSRs for simple sequence repeats). Data for DNA from a crime scene and the frequency each combination is found in the overall population are shown.

<table>
<thead>
<tr>
<th>STR LOCUS</th>
<th>repeats in sample</th>
<th>frequency in population</th>
</tr>
</thead>
<tbody>
<tr>
<td>D3S13</td>
<td>15, 17</td>
<td>0.13</td>
</tr>
<tr>
<td>vWA</td>
<td>15, 16</td>
<td>0.22</td>
</tr>
<tr>
<td>FGA</td>
<td>23, 27</td>
<td>0.31</td>
</tr>
<tr>
<td>D8S117</td>
<td>12, 13</td>
<td>0.34</td>
</tr>
<tr>
<td>D21S11</td>
<td>28, 30</td>
<td>0.06</td>
</tr>
</tbody>
</table>

a) Why are there 2 listed repeat sizes for each locus?

The individual inherited different repeat sizes (we are diploid)

b) What technique most likely was used to detect the repeats in the sample?

PCR using primers that flank the repeat & separation of products by size

c) Which locus is most ‘informative’?

D21S11 since it is the rarest

d) What are the odds another person at random would have the same array of alleles for these genes? Just a formula is fine.

0.13 X 0.22 X 0.31 X 0.34 X 0.06 (comes out <2 in 10,000)