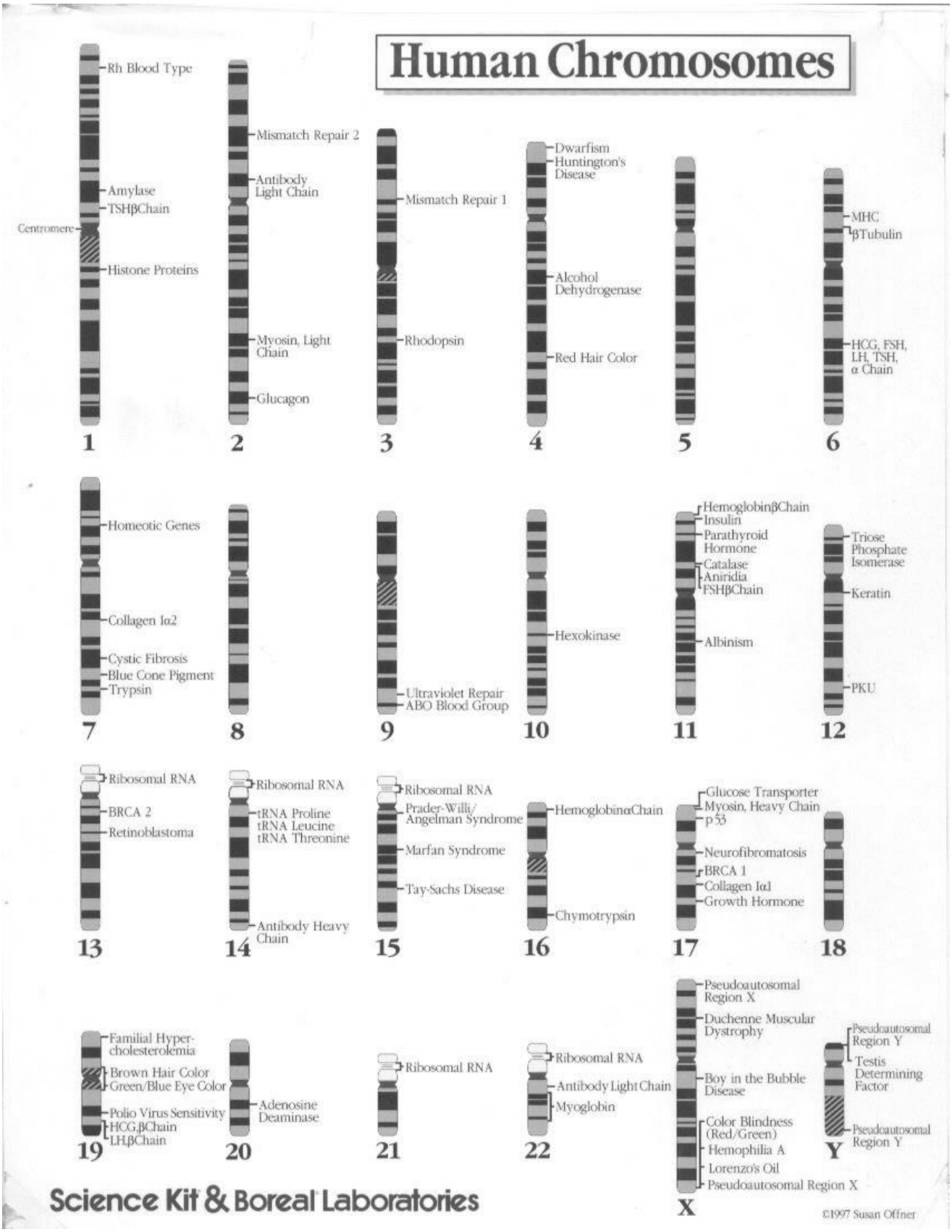
14.1 Human Heredity Notes

Human Chromosomes:

Genome: the full set of genetic information that an organisms carries on its DNA

Karyotype: the complete set of chromosomes grouped together in pairs, arranged in order of decreasing size



pairs 1-22 = **autosomes**; pair 23 = **sex chromosomes** (XX = female, XY = male) – usually…

Transmission of Human Traits:

→Simple Mendelian Inheritance of Human Traits

Dominant	Recessive
tongue curling	can't curl tongue
free earlobes	attached earlobes
polydactyly	5 finger or toes
dark hair	light hair
widow's peak	straight hairline
non-red hair	red hair
bent little finger	straight little finger
finger hair	no finger hair
straight thumb	hitchhiker's thumb
left thumb on top	right thumb on top
PTC taster	PTC nontaster

→Complex Inheritance of Human Traits

A. **Multiple Alleles**

1. blood type – three alleles:

I^A – produces surface molecule A, I^B – produces surface molecule B, i – produces no surface molecule

Phenotype (blood type)	Genotype(s)
A	I^AI^A or I^Ai *or* (AA,AO)
B	I^BI^B or I^Bi *or* (BB,BO)
AB	I^AI^B *or* (AB)
O	ii *or* (OO)

AA, BB – homo
AO, BO - hetero

2. the importance of blood typing
   - → during transfusions, incompatible blood types would result in clumping & death
   - → helpful in cases of disputed parentage

B. Polygenic Inheritance – determined by alleles on more than one gene

1. skin color – three to six genes are involved – the more genes, the more variation

C. **Sex-linked Traits**

**If X-linked: males pass on to daughters & not sons; females pass to daughter or son

X-linked are usually recessive (all of your X's must have it to show it)

Females XX: X^CX^C – okay X^CX^c – carrier X^cX^c – has it

Males XY: X^CY – okay X^cY – has it

**If Y-linked (rare): males pass to sons; females neither pass nor are affected

note: since most sex-linked traits we discuss are recessive, sometimes we don't even write the dominant on the other X
X^CX^C → XX; X^CX^c → XX^c

1. **red-green color blindness**:

**more common in males since they only have one X

2. **hemophilia** – blood doesn't have the ability to clot properly – also X-linked recessive

**studied in the family of Queen Victoria

**can be treated with transfusions or injections of a clotting agent

D. **X-chromosome Inactivation** - In females, most of the genes in one of the X chromosomes are inactivated (by super coiling into a structure known as a **Barr Body**) in each cell.

**Example: calico cats – color of spots on fur is controlled by gene on X chromosome; spots are orange or black depending on which X chromosome is inactivated in different patches of skin.

Human Pedigrees: A chart used to analyze the pattern of inheritance that shows the relationships in a family is a **pedigree**. Pedigrees can be used to determine the nature of genes and alleles associated with inherited human traits.