HUMAN GENETICS: PHENOTYPE AND GENOTYPE

LAB REPORT

Name

Date

Course Name

Exercise 1: Observing Phenotype Characteristics

Introduction

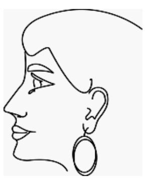
A phenotype refers to all the observable features of an organism that arise due to the interaction of the organism's genotype with its environment. Some examples of phenotype include colour, shape, size, behaviour, and biochemical properties. Based on changes in the organism's environment, the phenotype may change constantly reflecting changes in the physiology of the body. The genotype of an organism is completely inherited from the parents, whereas an organism's phenotype is the combination of the genotype and epigenetic modifications and environmental factors. Phenotype may be influenced by availability of food, humidity, temperature, and stress. One example of alterations in phenotype based on environmental factors is a person's skin colour. Although melanin production in the skin is genetically regulated, the amount of exposure to sunlight can trigger increased melanin production resulting in darkening of the skin.

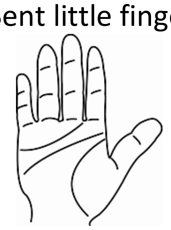
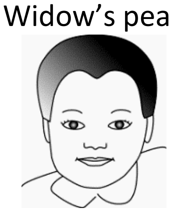
Based on the observed phenotype, the possible genotype of an organism can be deduced with the help of Punnett squares. Punnett squares are square diagrams used to predict the probability of offsprings having a particular genotype. It gives all possible combinations of a mother's and father's alleles in the form of a table. It is a visual representation of Mendelian inheritance where the combination and distribution of alleles can easily be deduced.

In this exercise, several phenotypes were observed for a set of parents and the probability of their offspring having the phenotype was calculated. Their possible genotypes and genotypic ratios were also calculated.

Results

Taking into account 10 different phenotypes, the characteristic features of each parent was deduced for all phenotypes. Punnett squares were used to determine the possible combinations of alleles of the parents, and all possible genotypes and genotypic ratios for the offspring were calculated. The results are given in Table 1 below:

Trait	Phenotype Characteristics	Phenotype & Genotype from Parent 1	Phenotype & Genotype(s) from Parent 2	Offspring
Dimpled chin 	Dimpled chin is dominant. • D = dimples • dd = No dimples	Dimpled Chin **Dd**	No dimples dd	Possible genotypes: Dd, dd Genotypic ratio: 1:1 Phenotype: 50% dimpled chin, 50% no dimples
Free ear lobe 	Free (unattached) ear lobe is dominant; attached is recessive. • A = unattached • aa = attached	Attached ear lobe **aa**	Unattached AA and Aa	Possible genotypes: Aa, aa Genotypic ratio: 3:1 Phenotype: 75% free ear lobe, 25% no attached ear lobe
Ability to taste PTC strips	Ability to taste PTC is dominant; inability to taste is recessive. • P = Taste • pp = Cannot taste	Ability to taste **Pp**	Cannot taste pp	Possible genotypes: Pp, pp Genotypic ratio: 1:1 Phenotype: 50% can taste PTC strips, 50% cannot taste PTC strips

Trait	Phenotype Characteristics	Phenotype & Genotype from Parent 1	Phenotype & Genotype(s) from Parent 2	Offspring
Interlocking fingers 	Left thumb over is dominant; right thumb over is recessive. • F = Left over right • ff = Right over left	Right thumb over left **ff**	Left over right FF and Ff	Possible genotypes: Ff, ff Genotypic ratio: 3:1 Phenotype: 75% left over right, 25% right over left
Mid-digital hair	Hair on the joints of the fingers is dominant; no hair is recessive. • H = Hair on joints • hh = No hair	Hair on joints **HH**	No hair hh	Possible genotypes: Hh Phenotype: Hair on joints
Bent little finger 	Little finger bending in toward ring finger is dominant; no bend is recessive. • B = bent • bb = Straight	Little finger bends **Bb**	Straight bb	Possible genotypes: Bb, bb Genotypic ratio: 1:1 Phenotype: 50% bent little finger, 50% straight little finger
Widow's peak 	Hairline coming to a point in center of forehead is dominant; straight hairline is recessive. • W = Widow's peak • ww = Straight hairline	Straight hairline **ww**	Straight hairline ww	Possible genotypes: ww Phenotype: Straight hairline

Trait	Phenotype Characteristics	Phenotype & Genotype from Parent 1	Phenotype & Genotype(s) from Parent 2	Offspring
Hitchhiker's thumb 	Ability to bend thumb back more than 90° is dominant, straight thumb is recessive. • T = Bent thumb • tt = Straight thumb	Bent thumb TT	Straight thumb tt	Possible genotypes: Tt Phenotype: Bent thumb
Blue or green eyes 	Brown eyes are dominant; blue or green eyes are recessive. • E = Brown eyes • ee = Blue eyes or green eyes	Green eyes ee	Brown eyes EE and Ee	Possible genotypes: Ee, ee Genotypic ratio: 3:1 Phenotype: 75% brown eyes, 25% blue or green eyes
Rolled tongue 	Ability to roll tongue is dominant; flat tongue is recessive. • L = Rolled tongue • ll = Flat tongue	Rolled tongue Ll	Rolled tongue LL and Ll	Possible genotypes: LL, Ll, ll Genotypic ratio: 3:4:1 Phenotype: 87.5% rolled tongue, 12.5% flat tongue

Table 1: A summary of the parent's phenotypes, possible genotypes and phenotypes of offsprings, and genotypic ratios

Dimpled Chin		Parent 1	
		D	d
Parent 2	d	Dd	dd
	d	Dd	dd

Dimpled Chin		Parent 1	
Parent 2			

Free Ear Lobe		Parent 1	
		a	a
Parent 2	A	Aa	Aa
	A	Aa	Aa

Free Ear Lobe		Parent 1	
		a	a
Parent 2	A	Aa	Aa
	a	aa	aa

Ability to Taste PTC Strips		Parent 1	
		P	p
Parent 2	p	Pp	pp
	p	Pp	pp

Ability to Taste PTC Strips		Parent 1	
Parent 2			

Interlocking Fingers		Parent 1	
		f	f
Parent 2	F	Ff	Ff
	F	Ff	Ff

Interlocking Fingers		Parent 1	
		f	f
Parent 2	F	Ff	Ff
	f	ff	ff

Mid-digital Hair		Parent 1	
		H	H
Parent 2	h	Hh	Hh
	h	Hh	Hh

Mid-digital Hair		Parent 1	
Parent 2			

Bent Little Finger		Parent 1	
		B	b
Parent 2	b	Bb	bb
	b	Bb	bb

Bent Little Finger		Parent 1	
Parent 2			

Widow's Peak		Parent 1	
		w	w
Parent 2	w	ww	ww
	w	ww	ww

Widow's Peak		Parent 1	
Parent 2			

Hitchhiker's Thumb		Parent 1	
		T	T
Parent 2	t	Tt	Tt
	t	Tt	Tt

Hitchhiker's Thumb		Parent 1	
Parent 2			

Blue or Green Eyes		Parent 1	
		e	e
Parent 2	E	Ee	Ee
	E	Ee	Ee

Blue or Green Eyes		Parent 1	
		e	e
Parent 2	E	Ee	Ee
	e	ee	ee

Rolled Tongue		Parent 1	
		L	l
Parent 2	L	LL	Ll
	L	LL	Ll

Rolled Tongue		Parent 1	
		L	l
Parent 2	L	LL	Ll
	l	Ll	ll

Table 2: Punnett squares for each of the phenotypes showing possible combinations of alleles of the parents

Discussion

From the results, it is evident that use of Punnett squares and calculation of genotypic ratios can help us monitor possible alleles of a given phenotype and their manifestation in offsprings. In this exercise, ten different traits were analyzed: dimpled chin, attached or unattached ear lobes, ability to taste PTC strips, interlocking fingers, mid-digital hair, bent or straight little finger, widow's peak, hitchhiker's thumb, colour of the eyes, and rolled tongue. Dominant and recessive traits were assigned to each parent for each of the phenotypes and possible alleles were noted down for the parents. Using Punnett squares, all possible combinations of alleles were acquired for the offspring. Based on this, genotypic ratios and the percentage of possible phenotypes were calculated for each trait.

There were 3 traits that had a genotypic ratio of 1:1 meaning that the offspring had a 50% chance of acquiring either phenotype. These traits were dimpled chin, ability to taste PTC strips, and bent or straight little finger. Three phenotypes were found to be either dominant or recessive based on the alleles such that all offsprings would invariably express the trait and these were presence of mid-digital hair, straight hairline, and bent thumb. Three phenotypes were found to have a 3:1 ratio meaning that there was a 75% chance that the offspring would acquire the dominant trait and a 25% chance that the offspring would acquire a recessive trait. These phenotypes were attached or unattached ear lobes, interlocking fingers, and colour of the eyes. There was just one trait that had the genotypic ratio of 3:4:1 based on the allele combinations with the offspring having an 87.5% chance of acquiring the dominant phenotype and this trait was rolled tongue. Thus, given the phenotypes of the parents, the possible genotypes, their allele combinations, genotypic ratios, and phenotypic percentages can be deduced.

Conclusion

From the above exercise, it is clear that it is possible to predict the phenotypes along with their percentages in offsprings given the phenotype and possible genotypes of the parents. Several traits related to physical appearance were analyzed and predicted for the offsprings.

The traits given in the table are related to physical appearance and they are not especially important for a person's survival. The genetic traits that need to be monitored and predicted when two parents create an offspring are genetic disorders which may arise due to errors in numbers of chromosomes, non-disjunction of chromosomes during meiosis, or mutations in genes which may result in drastic physical or behavioural changes sometimes resulting in death. Hence, if either of the parents has a family history of genetic disorders or themselves exhibits a genetic disorder, it is important to understand the possible genotypes of the parents and carefully monitor its manifestation in the offspring. Sometimes, even if the parents don't exhibit a genetic disorder, they may still be carriers of the gene and two parents who are carriers of the same disease may pass it on to their offspring while they themselves don't exhibit any symptoms of the disease.

Parents who are planning for a child are often encouraged to go for genetic counselling where their family history is analyzed and the probability of the offspring manifesting a genetic disorder is calculated. In this way, precautions can be taken to delay the onset of symptoms for the offspring and begin early treatment to reduce its effects in everyday life.

Exercise 2: Genetic Screening: Phenotype/Genotype

Introduction

Karyotyping is an important part of genetic screening which involves visualizing the chromosomes of an individual to determine his/her chromosome complement. It is a test that is used to view the complete set of chromosomes of an individual to measure the number of chromosomes and see if there are any changes in the number. A karyotype refers to the complete count of chromosomes in an organism and their appearance under a microscope. The features that are noted during karyotyping are length, number, banding patterns, and position of centromeres.

The normal human karyotype consists of 22 pairs or 44 autosomal chromosomes numbered 1 to 22. It also has one pair of sex chromosomes which may be XX in case of females and XY in case of males. Any variation from this standard karyotype may result in severe genetic and developmental abnormalities.

There are several things that need to be noted while viewing a karyotype. The total length of the chromosomes may vary based on the amount of DNA duplication that has taken place during mitosis. The position of the centromere may vary due to differences in translocation. There may be differences in the number of chromosomes due to nondisjunction during crossing over. One or more of these errors may result in phenotypic manifestations that may lead to severe behavioural changes and developmental abnormalities sometimes resulting in death.

Results

The karyotypes of individuals with two possible chromosomal disorders were analyzed and the correct disorder was identified. The results are given in Table 3 below.

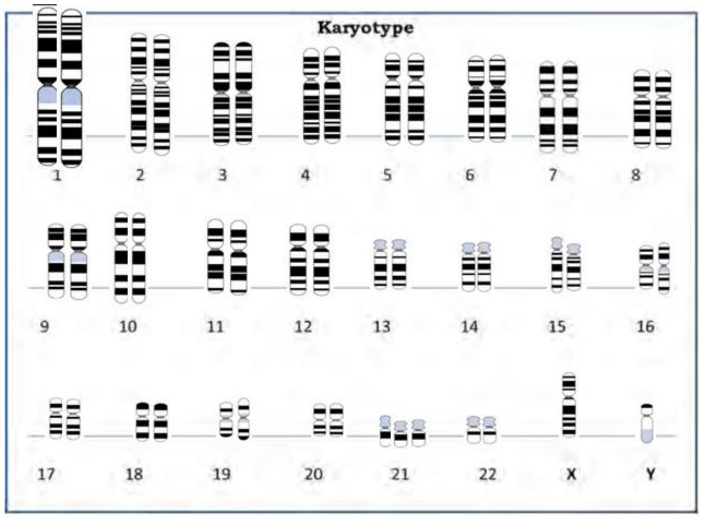
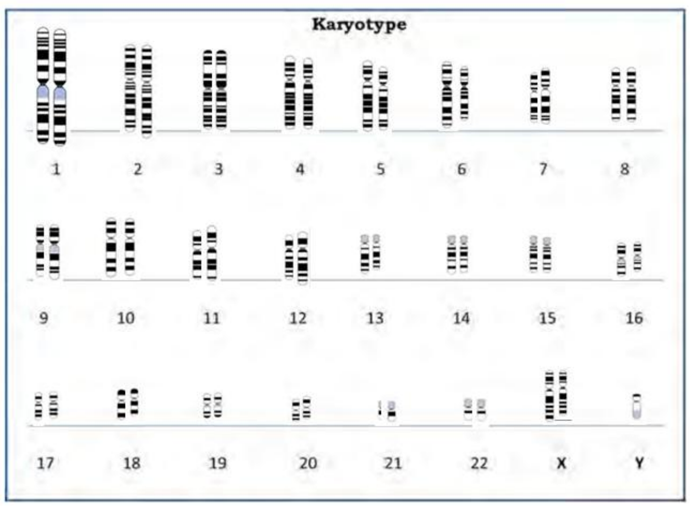
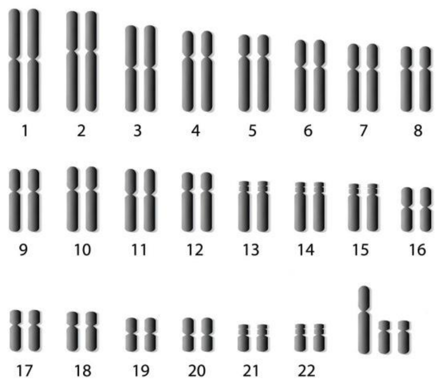
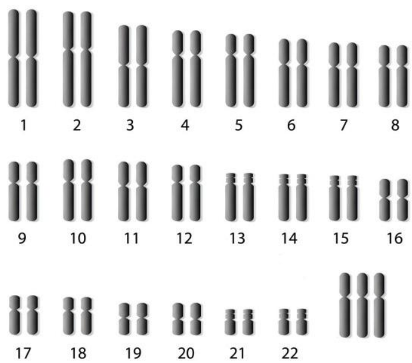
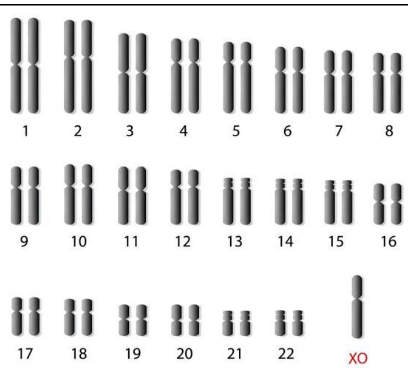
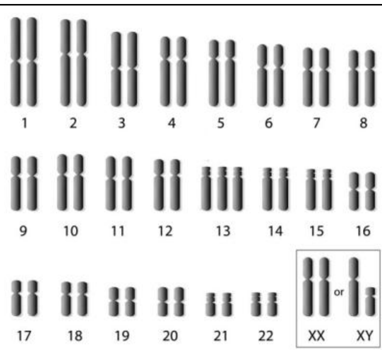
	Karyotype Image	Chromosomal Disorder
Possible Chromosome Disorder 1 Karyotype		Down Syndrome
Possible Chromosome Disorder 2 Karyotype		Klinefelter Syndrome

Table 3: Analysis of karyotypes for identification of possible chromosomal disorders

In six other cases, the chromosomal disorder, genotype, and phenotype based on the karyotype were deduced. The results are given in Table 4 below.

#	Karyotype	Chromosomal Disorder	Genotype	Phenotype
1		XYY Syndrome	An extra copy of the Y chromosome (XYY)	Increased height, reduced cognitive ability, normal fertility
2		Triple X Syndrome	An extra copy of the X chromosome (XXX)	Mild (or none) phenotypic manifestations. Healthy with normal fertility. Increased risk of delayed development.
3		Turner's Syndrome	Only one X chromosome	Only viable monosomy not resulting in death. Reduced height, sterile, normal intelligence
4		Patau Syndrome	An extra copy of chromosome 13	Cleft palate as well as eye, brain, and circulatory defects

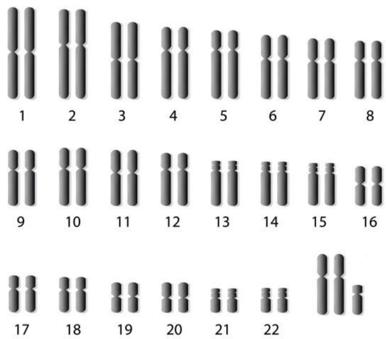
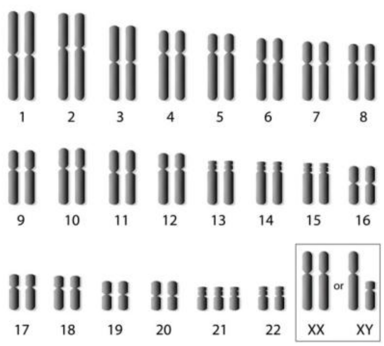
#	Karyotype	Chromosomal Disorder	Genotype	Phenotype
5		Klinefelter Syndrome	At least one extra copy of the X chromosome (XXY)	Reduced muscle strength, reduced testosterone production, increased height, broad hips, pronounced breasts, small testes, reduced fertility
6		Down Syndrome	An extra copy of chromosome 21	Delayed cognitive ability, oblique eye shape, flat nasal bridge

Table 4: Analysis of chromosomal disorder, genotype, and phenotype based on the karyotype

Discussion

Given 8 different karyotypes, the chromosomal disorder was analyzed based on the numbers of the autosomal and sex chromosomes. The genotypes and possible phenotypes of each observed chromosomal disorder were also noted.

The presence of three copies of chromosome 21 instead of two indicates Down syndrome which leads to delayed cognitive abilities and changes in physical appearance. An extra copy of the X chromosome in males manifests as Klinefelter syndrome leading to significant changes in physical appearance and reduced fertility. On the other hand, if there is an extra copy of the Y chromosome in males, the person has normal fertility but reduced cognitive abilities. Females having thee X chromosomes instead of two are relatively healthy with normal fertility. Individuals with Turner's syndrome have only one copy of the X chromosome and they are sterile with normal intelligence. The presence of three copies of chromosome 13 manifests as Patau syndrome resulting in significant brain and circulatory defects.

Conclusion

A karyotype is an important tool that is widely used in genetic screening to visualize chromosomal aberrations and predict possible phenotypes in the offspring. During DNA replication and crossing over during meiosis, several errors may happen in the distribution of chromosomes between daughter cells. These errors may result in different types of syndromes manifesting as significant physical, behavioural, cognitive, and developmental changes in an individual. Early detection of these types of genetic disorders can help implement precautionary measures to delay the onset of symptoms in the offspring.

Although a karyotype is very helpful in identifying genetic disorders, it can only help visualize chromosomal aberrations and it cannot give any insight on the genetic level. Even if a person has a normal karyotype, s/he may still carry deadly mutations in the genes that may cause phenotypic manifestations in the offspring. Some examples of this are cancer and diabetes which are not discernable by karyotyping alone and more intensive genetic screening needs to be done to identify these genetic disorders.

The most common types of chromosomal disorders occur due to numerical changes resulting in extra or missing chromosomes in an individual. This may be due to large-scale deletions or duplications of a chromosome resulting in disorders such as Turner’s syndrome, Down syndrome, Klinefelter syndrome, Patau syndrome, and Edwards’s syndrome. All these disorders are discernible through karyotyping and the possible phenotypes for an offspring can be predicted in the early developmental stages.