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NEET 2020

Genetic Disorders & Genetic Problems

BIOTONIC

DR. VANI SOOD

NEET EXPERT

Dr. Vani Sood (NEET Expert)

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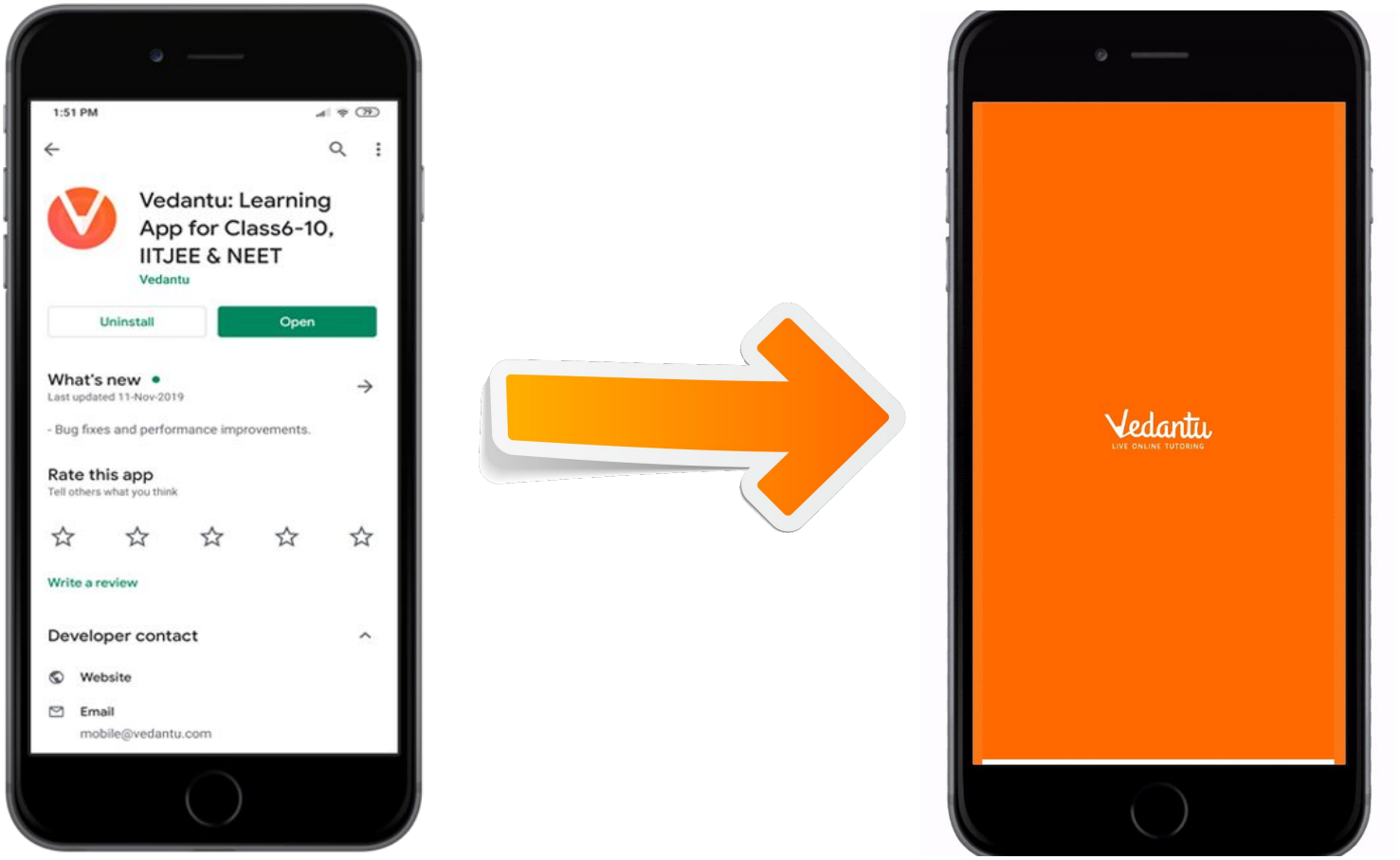
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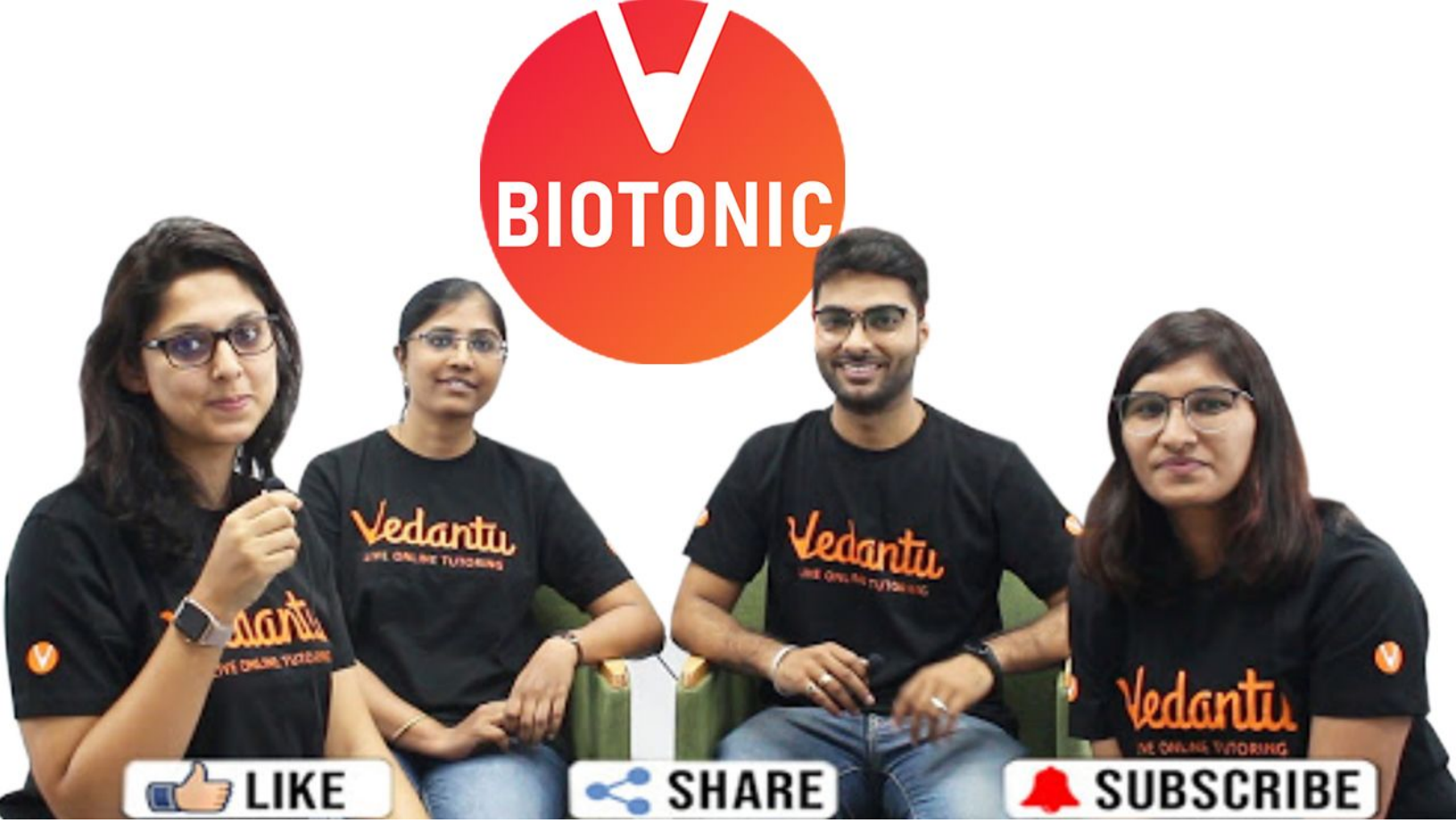
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Weightage in NEET

Unit and Topic	Number of Questions	Total Marks	Weightage Percentage
Human Health & Diseases	5	20	18.00%

DISORDERS

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DISORDERS

1. Kwashiorkor

→ Malnutrition produced by a severely inadequate amount of protein in the diet.

Cause

→ Protein deficiency

Treatment

→ A slow increase in calories followed by an increase in protein.
→ High-protein diet, Vitamin and mineral supplementation,

Symptoms

→ Delayed growth in children, a swollen stomach, frequent infections, fatigue, loss of appetite, or thirst, bloating or diarrhoea, ear infections, hair loss, irritability, or skin rash

2. Marasmus

→ "Marasmus can be defined as a form of nutrition deficiency disorder, usually occurring in children.

Cause

→ Severe deficiency of protein, vitamins, minerals, carbohydrates, and lipids. Poverty and Starvation

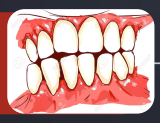
Symptoms

→ Dizziness, Weight loss, Dehydration, Lack of energy, Stunted growth, Shrinkage of Stomach, Respiratory infections, Dry skin and brittle hair, Underweight or loss of muscle mass and subcutaneous fat.

Treatment

→ Multivitamin supplements, water and other fluids that contain electrolytes. well-balanced diet, Maintain good sanitation and hygiene

3. Scurvy



→ A condition caused by a severe lack of vitamin C in the diet.

Cause

→ Deficiency of vitamin C in the diet.

Symptoms

→ Bruising, bleeding gums, weakness, fatigue and rash are among scurvy symptoms.

Treatment

→ Treatment involves taking vitamin C supplements and eating citrus fruits, potatoes, broccoli and strawberries.

DISORDERS

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DISORDERS

4. Pellagra

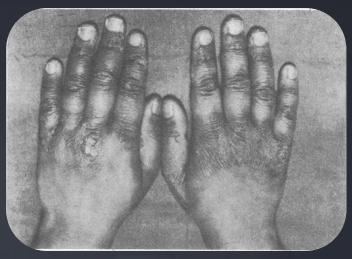
→ A disease caused by low levels of niacin, also known as vitamin B-3

Cause

→ Niacin Deficiency

Symptoms

→ Dementia, diarrhea, and dermatitis,



Treatment

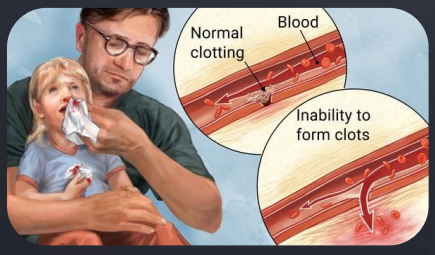
→ Intake of niacin in diet. Good sources of niacin include red meat, fish, poultry, fortified breads and cereals, and enriched pasta and peanuts.

5. Hemophilia

→ A Genetic disorder in which blood doesn't clot normally.

Cause

→ Inherited Genetic Mutation on X chromosome



Symptoms

→ Prolonged bleeding, pain and stiffness around joints, such as elbows, because of bleeding inside the body (internal bleeding)

Treatment

→ No cure for haemophilia, Genetically engineered clotting factor medicines are used to prevent and treat prolonged bleeding.

DISORDERS

DISORDERS

7. Turner Syndrome

→ A chromosomal disorder in which a female is born with only one X chromosome

Cause

→ Absence of one X chromosome in females

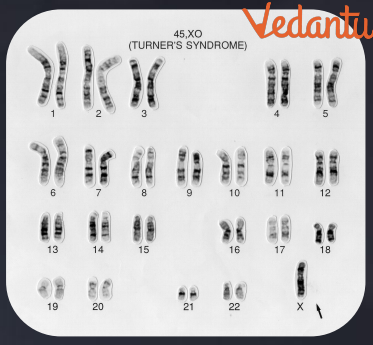
Symptoms

→ Short stature, delayed puberty, infertility, heart defects and certain learning disabilities.

→ Congenital heart defect or narrowing of the aorta, underactive thyroid, vision disorder, or webbed neck

Treatment

→ Hormone therapy



6. Down Syndrome

→ A genetic disorder causing developmental and intellectual delays.

Cause

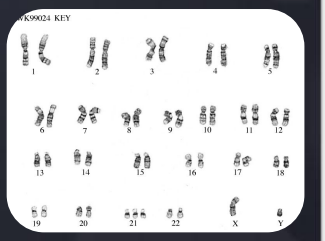
→ Extra 21st chromosome, nondisjunction of 21st chromosome

Symptoms

→ Distinct facial appearance, intellectual disability and developmental delays. Congenital heart disease, flaccid muscles, hearing loss, immune deficiency, low-set ears, mouth breathing, obesity, Single line on palm, thickening of the skin of the palms and soles, thyroid disease, or vision disorder

Treatment

→ Speech therapy and Physical Therapy



Q. A colorblind man marries the daughter of a colorblind person. Then in their progeny

A. None of their daughters are colour blind

B. All the sons are colour blind

C. All the daughters are colour blind

D. Half of their sons are colour blind

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A. None of their daughters are colour blind

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C. All the daughters are colour blind

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Solution: Since daughter of a colorblind person will be a carrier of the disease and only one of the two X chromosomes will bear the recessive gene for disease, so half of their sons will be colourblind.

Q. Given below is a pedigree chart of a family with five children. It shows the inheritance of attached earlobes as opposed to the free ones. The squares represent the male individuals and circles the female individuals. Which one of the following conclusions drawn is correct

A. The parents are homozygous recessive

B. The trait is Y-linked

C. The parents are homozygous dominant

D. The parents are heterozygous

Q. Given below is a pedigree chart of a family with five children. It shows the inheritance of attached earlobes as opposed to the free ones. The squares represent the male individuals and circles the female individuals. Which one of the following conclusions drawn is correct

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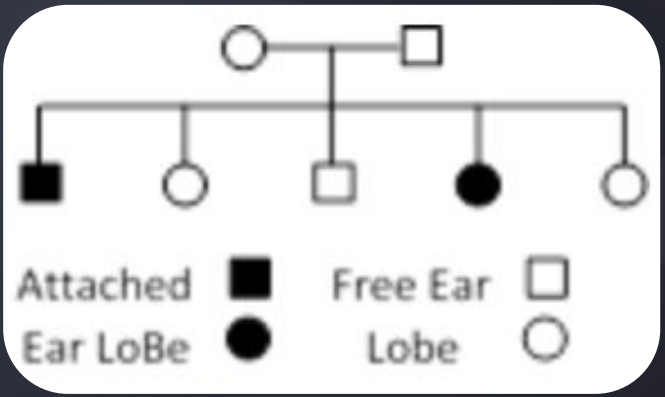

Solution: Free-hanging earlobes is an autosomal dominant trait. Since both trait are expressed in the F1 generation. Both parents must be heterozygous

Q. Mendel crossed a pure white flowered pea plant with pure red flowered plant. The first generation of hybrids from the cross should show

A. 75% red flowered and 25% white flowered plants.

B. 50% white flowered and 50% red flowered plants.

C. All red flowered plants.

D. All white flowered plants

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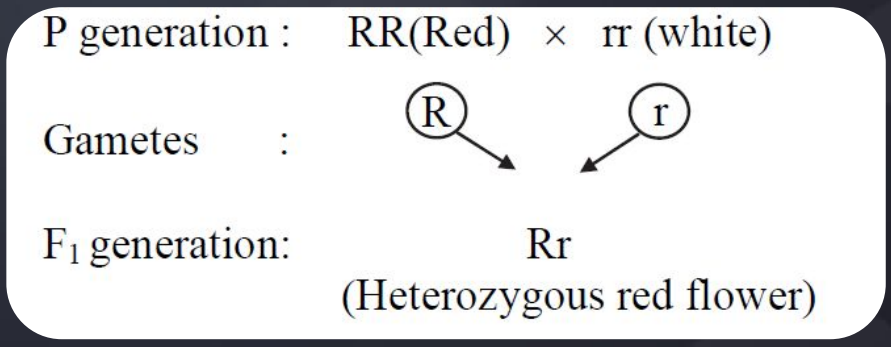
A. 75% red flowered and 25% white flowered plants.

B. 50% white flowered and 50% red flowered plants.

C. All red flowered plants.

D. All white flowered plants

Solution: The dominant trait is expressed in F1 generation.



Q. The phenotypic ratio of red (AABB) and white (aabb) kernel in F2 generation showing polygenic inheritance is

A 1:2:1

B 1:4:6:4:1

C 1:6:4:4:1

D 1:6:15:20:15:6:1

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A. 1:2:1

B. 1:4:6:4:1

C. 1:6:4:4:1

D. 1:6:15:20:15:6:1

Parents : Red kernels AABB × White kernels aabb

Gametes : (AB) (ab)

AaBb

F_1 generation : Intermediate red

Selfing of F_1 generation : AaBb × AaBb

Gametes : (AB) (Ab) (aB) (ab) × (AB) (Ab) (aB) (ab)

F_2 generation :

	AB	**Ab**	**aB**	**ab**
AB	AABB ●●●●	AABb ●●●○	AaBB ●●●○	AaBb ●●○○
Ab	AABb ●●●○	AAbb ●●○○	AaBb ●●○○	Aabb ●○○○
aB	AaBB ●●●○	AaBb ●●○○	aaBB ●●○○	aaBb ●○○○
ab	AaBb ●●○○	Aabb ●○○○	aaBb ●○○○	aabb ○○○○

	●●●● Darkest Red	●●●○ Medium red	●●○○ Intermediate red	●○○○ Light red	○○○○ White
Phenotypic ratio	: 1	: 4	: 6	: 4	: 1

Q. What is the frequency of heterozygote Aa in a randomly mating population in which the frequency of all recessive phenotypes is 0.16?

A 0.84

B 0.63

C 0.36

D 0.48

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A 0.84

B 0.63

C 0.36

D 0.48

Solution:

According to the **Hardy-Weinberg equilibrium** the letter 'p' designates the frequency of one allele and the letter 'q' the frequency of the alternate allele. Because there are only two alleles, **p** plus **q** must always be equal 1. The Hardy-Weinberg equation can be expressed in the form of what is known as a binomial expansion: $(p + q)^2 = p^2 + 2pq + q^2$

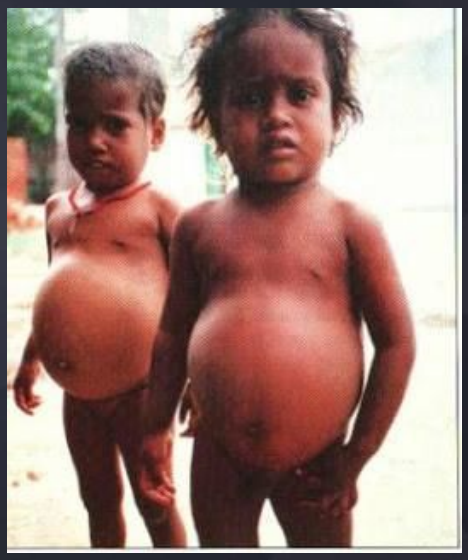
If **q^2 = 0.16, then q = 0.4.**

Therefore, **p**, the frequency of allele **A, p+q=1, q=1-p** would be **0.6 (1.0 - 0.4 = 0.6)**

We can now easily calculate the genotype frequencies: **$p^2 = (0.6)^2$ = 0.36 or 36% AA i**ndividuals. The heterozygous would have a frequency of **2pq or (2 x 0.6 x 0.4) = 0.48 or 48%**

KWASHIORKOR

- **Malnutrition** produced by a severely **inadequate amount of protein** in the diet.



KWASHIORKOR

- **Malnutrition** produced by a severely **inadequate amount of protein** in the diet.

Name of the disease	Cause	Symptoms	Treatment
Kwashiorkor	**Protein deficiency**	**Delayed growth in children, a swollen stomach, frequent infections, fatigue, loss of appetite, or thirst, bloating or diarrhoea, ear infections, hair loss, irritability, or skin rash**	**a slow increase in calories followed by an increase in protein. High-protein diet, Vitamin and mineral supplementation, Breastfeeding**

MARASMUS

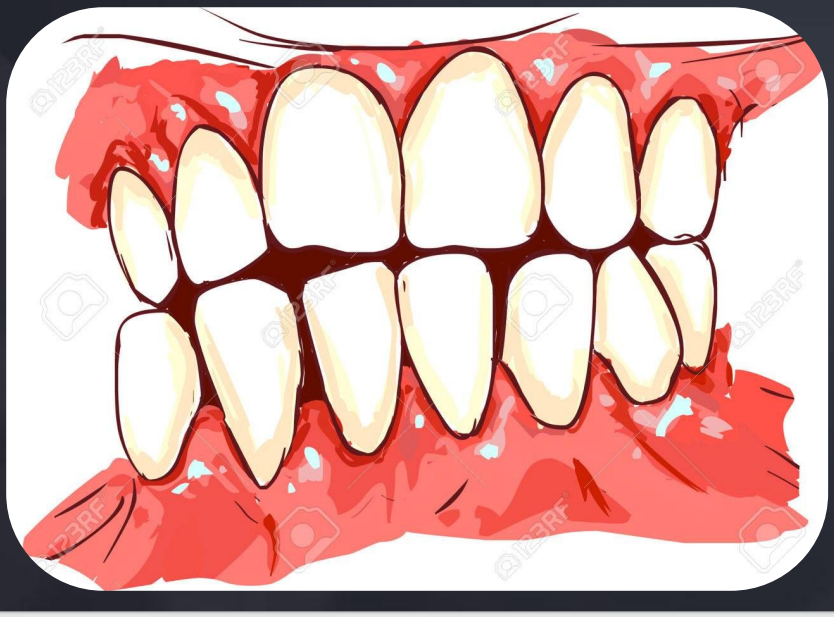
- “Marasmus can be defined as a form of nutrition deficiency disorder, usually occurring in children.

MARASMUS

Name of the disease	Cause	Symptoms	Treatment
Marasmus	Severe deficiency of protein, vitamins, minerals, carbohydrates, and lipids. Poverty and Starvation	**Dizziness, Weight loss, Dehydration, Lack of energy, Stunted growth, Chronic Diarrhoea, Shrinkage of Stomach, Respiratory infections, Dry skin and brittle hair, Underweight or loss of muscle mass and subcutaneous fat.**	**Multivitamin supplements, water and other fluids that contain electrolytes. well-balanced diet, Maintain good sanitation and hygiene**

SCURVY

- A condition caused by a severe **lack of vitamin C** in the diet.

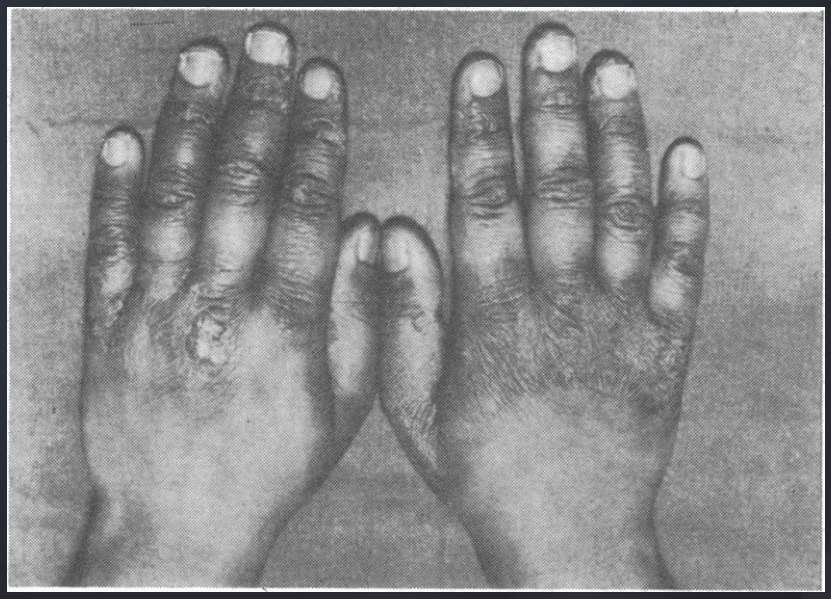


SCURVY

Name of the disease	Cause	Symptoms	Treatment
Scurvy	**Scurvy results from a deficiency of vitamin C in the diet.**	**Bruising, bleeding gums, weakness, fatigue and rash are among scurvy symptoms.**	**Treatment involves taking vitamin C supplements and eating citrus fruits, potatoes, broccoli and strawberries.**

PELLAGRA

- A disease caused by low levels of niacin, also known as vitamin B-3

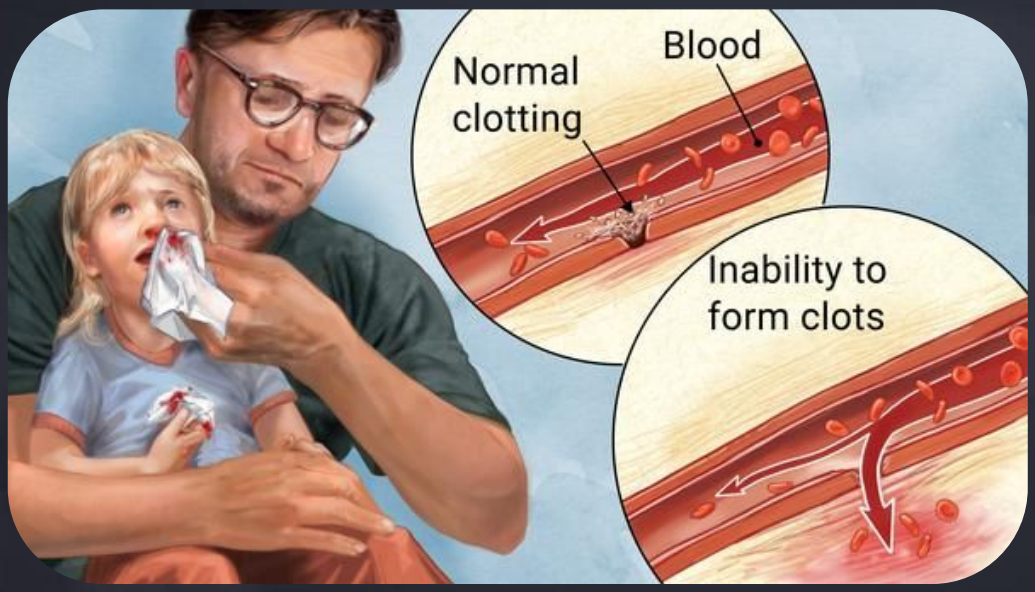


PELLAGRA

Name of the disease	Cause	Symptoms	Treatment
Pellagra	**Niacin Deficiency**	**dementia, diarrhea, and dermatitis,**	**Intake of niacin in diet. Good sources of niacin include red meat, fish, poultry, fortified breads and cereals, and enriched pasta and peanuts.**

HEMOPHILIA

- A Genetic disorder in which blood doesn't clot normally.

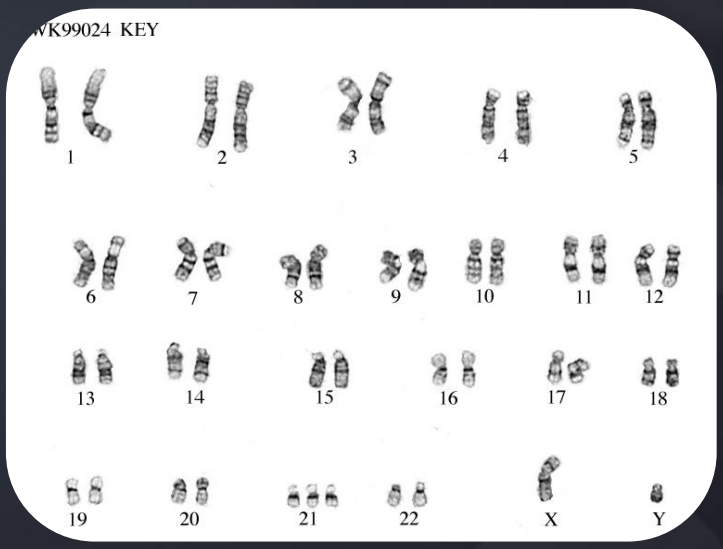


HEMOPHILIA

Name of the disease	Cause	Symptoms	Treatment
Hemophilia	**Inherited Genetic Mutation on X chromosome**	**bleeding that doesn't stop, also called prolonged bleeding, pain and stiffness around joints, such as elbows, because of bleeding inside the body (internal bleeding)**	**No cure for haemophilia, Genetically engineered clotting factor medicines are used to prevent and treat prolonged bleeding.**

DOWN SYNDROME

- A **genetic disorder** causing **developmental and intellectual delays.**

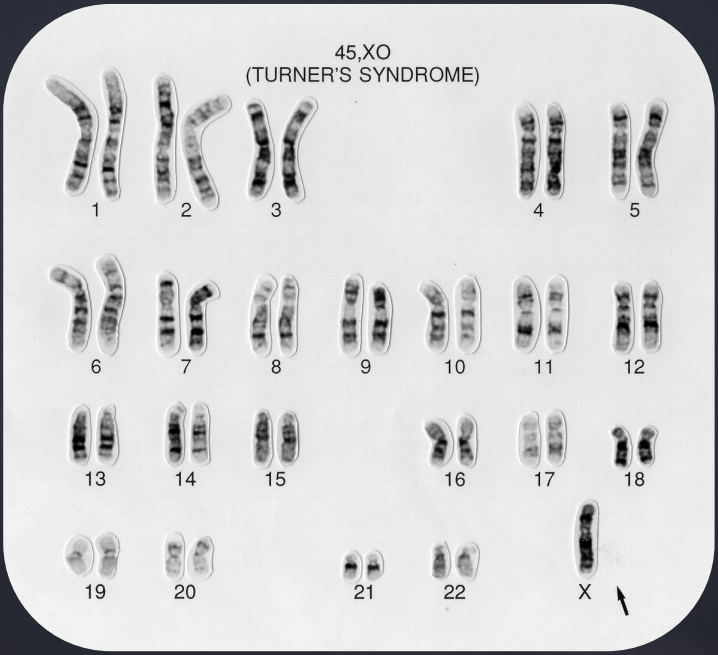


DOWN SYNDROME

Name of the disease	Cause	Symptoms	Treatment
Down Syndrome	Extra 21st chromosome, nondisjunction of 21st chromosome	causes a distinct facial appearance, intellectual disability and developmental delays. congenital heart disease, flaccid muscles, hearing loss, immune deficiency, low-set ears, mouth breathing, obesity, single line on palm, thickening of the skin of the palms and soles, thyroid disease, or vision disorder	Early intervention programmes with a team of therapists and special educators who can treat each child's specific situation are helpful in managing Down's syndrome. Speech therapy and Physical Therapy

TURNER SYNDROME

- A **chromosomal disorder** in which a **female is born with only one X** chromosome



TURNER SYNDROME

Name of the disease	Cause	Symptoms	Treatment
Turner Syndrome	**Absence of one X chromosome in females**	**Short stature, delayed puberty, infertility, heart defects and certain learning disabilities. congenital heart defect or narrowing of the aorta, underactive thyroid, vision disorder, or webbed neck**	**Treatment involves hormone therapy. Fertility treatment may be necessary for women who want to become pregnant.**

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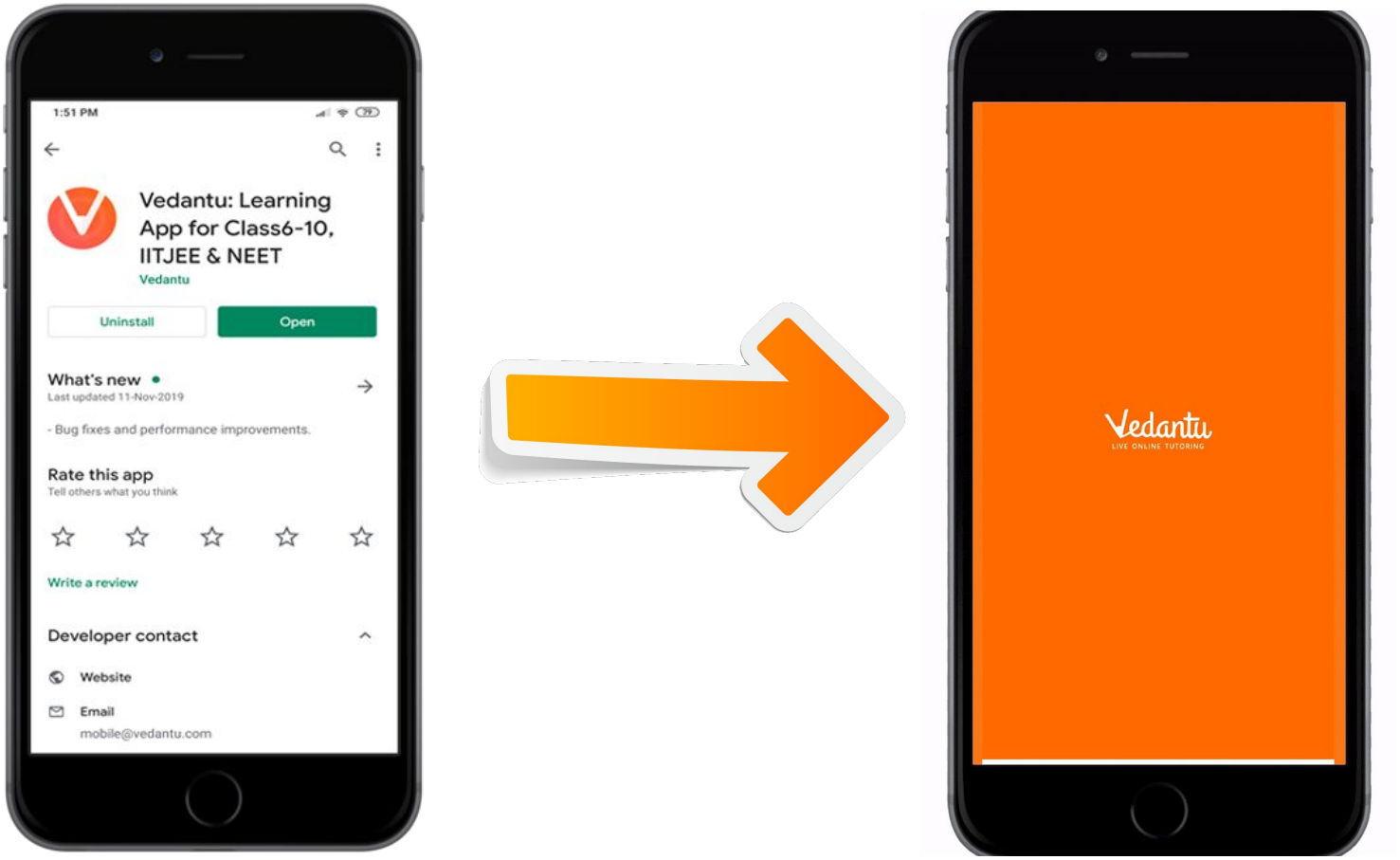
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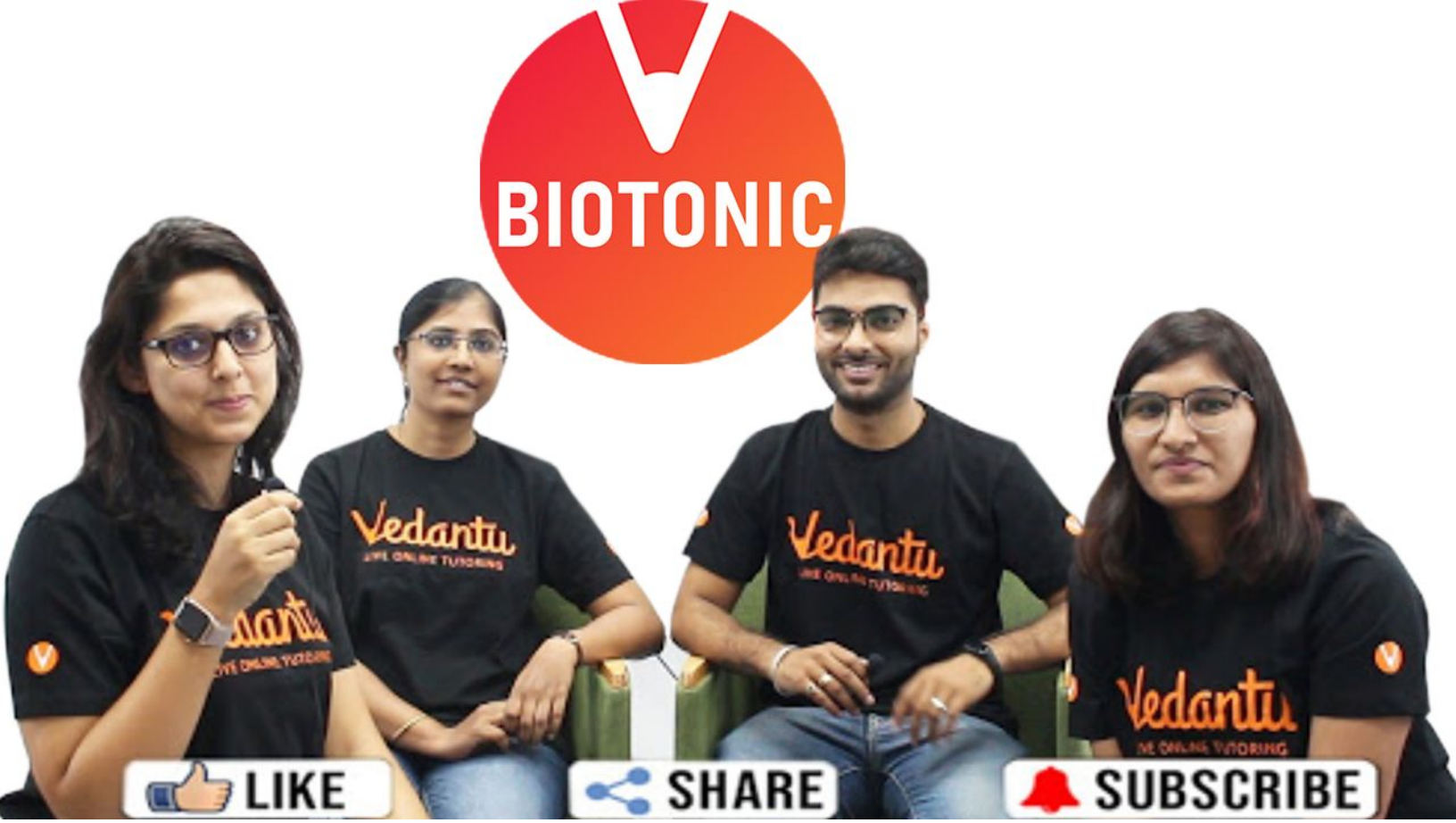
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