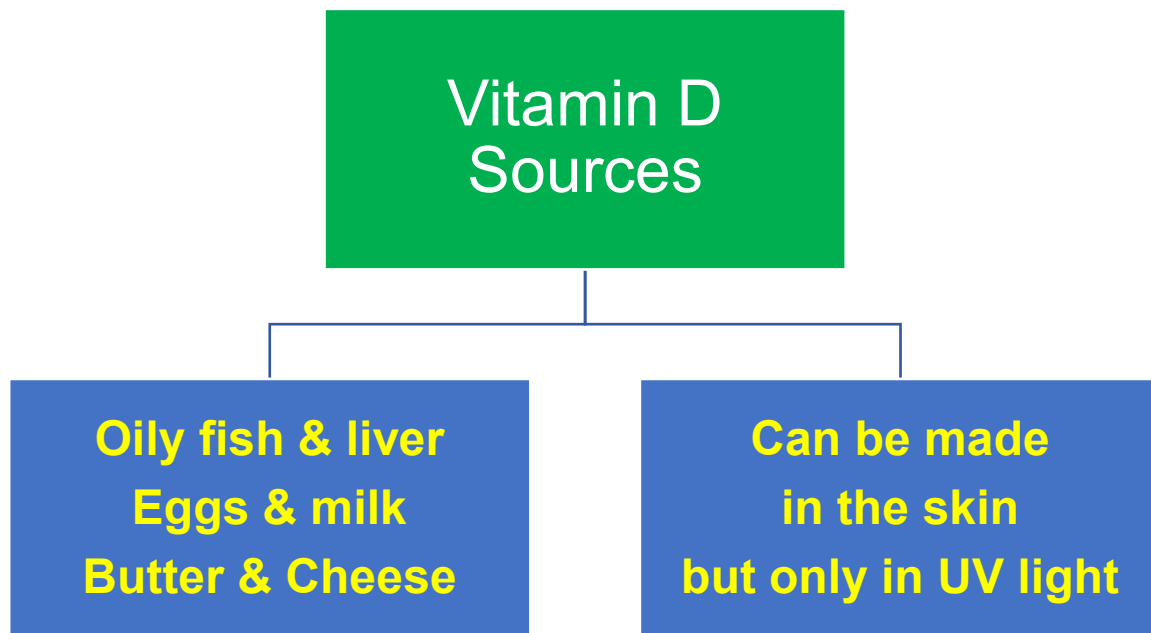


1. VITAMIN D DEFICIENCY IN HUMANS

- **Insufficient** vitamin D means that an **insufficient** amount of **calcium** is **absorbed** from the gut.
- This can lead to **osteomalacia** = **inadequate bone mineralisation** due to **calcium not** being **reabsorbed** or **deposited**. **Bones** become **softer** and can **bend easier**.
- **Osteomalacia** in **children** is called **rickets**.



- In **winter** at high latitudes, the **UV intensity** is **too low** to produce much **vitamin D**.
- However, the **liver** can **store** enough **vitamin D** during the **summer** to avoid a deficiency in winter.
- You can remember the **symptoms** of **vitamin D deficiency** using the mnemonic '**BADGES**'.

Vitamin D Deficiency
Deficiency disease: Rickets
Deficiency symptoms:
<ul style="list-style-type: none">• Bone fragility & fracturing• Atrophy / muscle weakening• Dental problems• Growth retardation• Enlargement of liver / spleen• Skeletal deformities
Mnemonic: BADGES

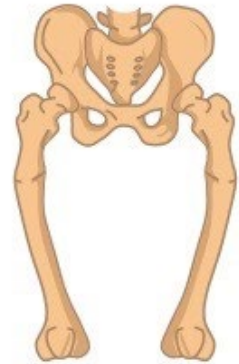


Normal Person



Straight legs

Rickets



Bow-legged

2. VITAMIN C (ASCORBIC ACID) DEFICIENCY IN MAMMALS

- **Vitamin C (ascorbic acid)** is found in **citrus fruits** and **orange juice**.
- **Vitamin C (ascorbic acid)** is needed to make **collagen fibres**.
- **Collagen fibres** are present in many tissues, including **skin** and **blood vessel walls**.



- A **lack** of vitamin C in the diet can cause **scurvy**.
- Humans **cannot** make vitamin C, so it **must** be provided in the **diet**.
- **Rats** and most other **mammals** do **have** the necessary **enzymes** to make vitamin C.
- Attempts to **induce** the **symptoms** of scurvy in **rats** were therefore **unsuccessful**.
- **Humans, guinea pigs, apes** and **chimpanzees** require vitamin C to be provided in their **diet**.
- You can remember the **symptoms** of vitamin D deficiency using the mnemonic '**SHADES**'.

Vitamin C Deficiency	
Deficiency disease: Scurvy	
Deficiency symptoms:	
<ul style="list-style-type: none">• Skin discoloration & bruising• Hemorrhaging• Anaemia• Dental issues• Exhaustion / fatigue• Swelling of joints (edema)	
Mnemonic: SHADES	

Normal Person



Healthy teeth
and gums

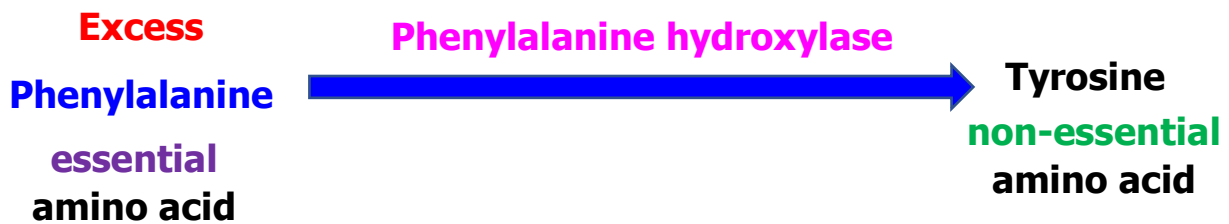
Scurvy



Loosened teeth and
bleeding / ulcerated gums

3. PHENYLKETONURIA (PKU)

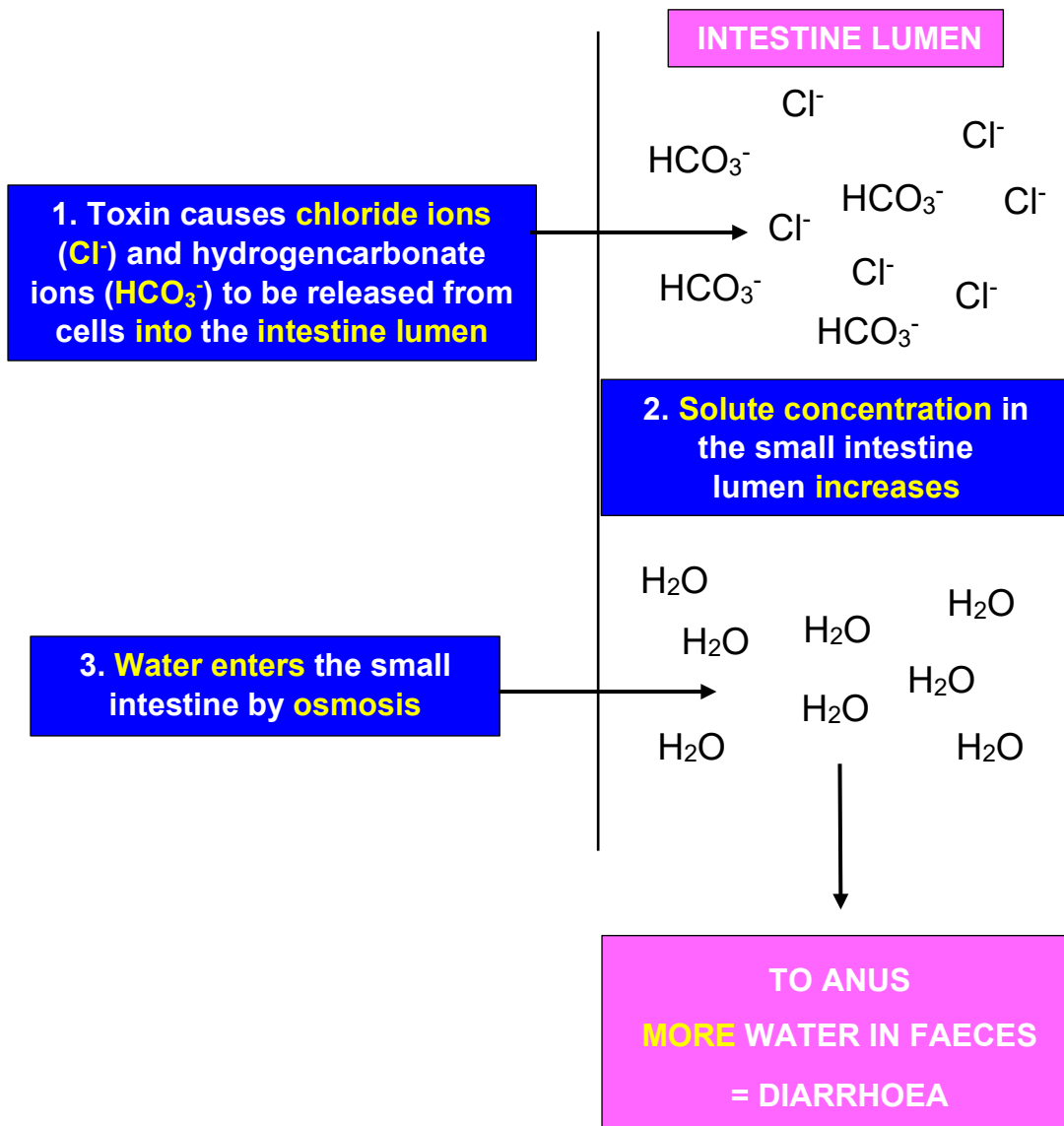
- **Phenylalanine** is an **essential** amino acid as it **must** be supplied in the **diet**.
- **Excess** phenylalanine is usually converted into **tyrosine**.
- **Tyrosine** is a **non-essential** amino acid, as it can be **made** from **phenylalanine**.
- **Phenylketonuria** (PKU) is a **genetic** disease caused by an **autosomal recessive** allele
- Caused by a **mutation** in the **gene** coding for the enzyme **phenylalanine hydroxylase**.



- If a person has **PKU**, they **cannot** produce **phenylalanine hydroxylase**.
- So, the level of **phenylalanine** in the **blood** becomes **too high**.
- **Before** birth, an **infant** with **PKU** has **no symptoms**, as the **mother's** body uses, breaks down or converts any excess **phenylalanine**.
- However, **after** birth, the **high** level of **phenylalanine** in the **blood** can cause **significant** health problems.
- Growth of the **head** and **brain** is **reduced**, causing **mental retardation**.
- Blood phenylalanine levels are **tested** soon **after birth**, allowing **early diagnosis** and treatment.
- **Treatment** for PKU is a **diet** with **low levels** of **phenylalanine** e.g. meat, fish, nuts, cheese and beans, which are still eaten in **low amounts**.
- **Tyrosine supplements** may also be needed if amounts in the **diet** are **insufficient**.

4. CHOLERA

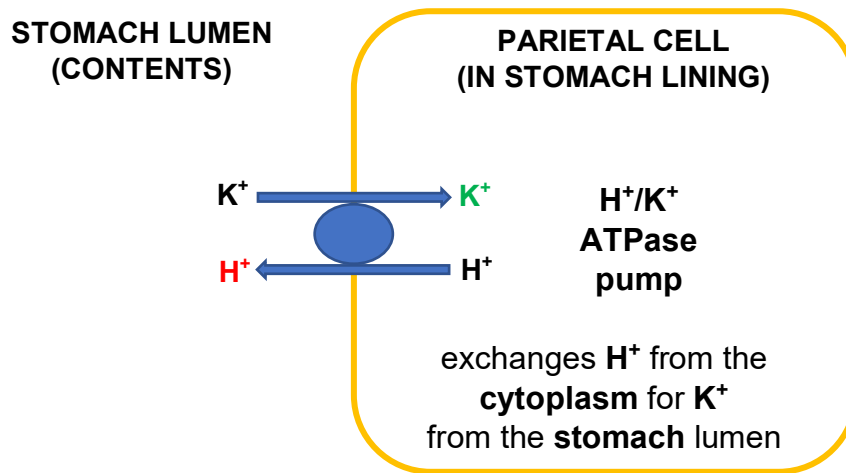
- Caused by the **bacterium** *Vibrio cholerae*, which releases a **toxin**.
- The toxin **binds** to a **receptor** on **intestine** cells and then enters the cell by **endocytosis**.



- Water is drawn from the **blood** into the intestine cells to **replace** the fluid lost.
- Death can occur due to severe **dehydration**.
- Treated by **oral rehydration therapy (ORT)**, which replaces **water** and **electrolytes** (=ions/sugars/salts).

5. EXCESSIVE STOMACH ACID SECRETION

- Parietal cells in the **stomach lining** contain a **proton pump** called **H⁺/K⁺ ATPase**.



- This can generate an **H⁺ gradient** of 3 million : 1 making the stomach contents **very acidic** and **corrosive**.
- A natural **mucus barrier** protects the **stomach lining** from **self-digestion**.
- In some people, this mucus barrier **breaks down**, resulting in **damaged stomach lining** and **bleeding**. This is known as an **ulcer**.
- **Circular muscle** at the **top** of the stomach **prevents acid reflux**, so acid does **not** enter the **oesophagus** from the stomach.
- In some people, this **circular muscle** does **not work** effectively, so acid **enters** the **oesophagus** from the stomach. This is known as **heartburn**.
- Both conditions can be **treated** using **proton pump inhibitors (PPIs)**.

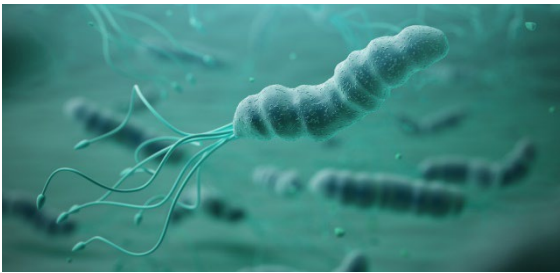


These bind **irreversibly** to the **H⁺/K⁺ ATPase pump**
Preventing H⁺ from being pumped into the stomach

6. STOMACH ULCERS

What they are and how they are caused

- Are open sores caused by **digestion** of the **stomach lining** by **pepsin** enzyme and **hydrochloric acid**.
- For many years, it was thought that **stress** and **excessive acid secretion** were the **main causes** of stomach ulcers.
- Now, about **80%** of **stomach ulcers** are thought to be caused by the bacterium *Helicobacter pylori*.



Helicobacter pylori causes **damage** to the **mucus lining** of the **stomach** by:

- causing **inflammation**
- producing **toxins** and **enzymes**
- causing **excessive acid** secretion

Barry Marshall

- Barry Marshall and Robin Warren **cured stomach ulcers** using **antibiotics** that killed **this bacterium**.
- It took **time** for antibiotics to be prescribed for stomach ulcers due to **fixed mindsets** from doctors and drug companies. They simply **did not believe that a bacterium was responsible**, due to the **low pH** of **stomach acid**.
- Barry Marshall **swallowed** a suspension of *Helicobacter pylori* and was ill with acute **gastritis**, suggesting that this bacterium can **cause** acute gastritis, which in turn can **cause stomach ulcers**.
- He also **cured** himself using **antibiotics**.
- He had asked neither an **ethics commission** nor his wife for permission to conduct this experiment. His colleagues thought him completely insane to take a risk like that.
- However, he was still awarded the **Nobel Prize** in Physiology and Medicine about 20 years later.

How stomach ulcers are treated

- **proton pump inhibitors** to **reduce stomach acid**
- **antacids** to **neutralize/decrease acidity**
- **decreased acidity** allows **ulcers** to **heal**
- **antibiotics** for *H. pylori* infection
- **stop** smoking/drinking alcohol
- **surgery** needed with **extensive gastric damage**