

BMSH4302

Introduction to Chronic Disease

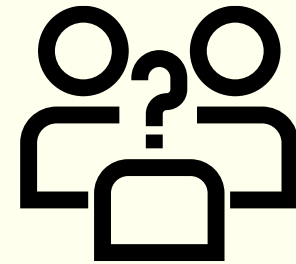
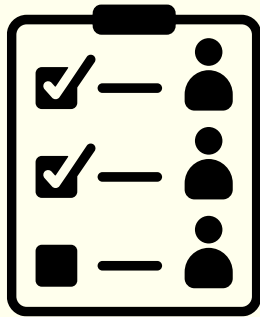
Level 4



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BMSH4302 Introduction to Chronic Disease

Housekeeping:





Musculoskeletal Diseases Recap

- Kahoot!

Neurological Diseases

What does Neurology mean?

NEURO = Nerve (from Greek 'nerve') plus

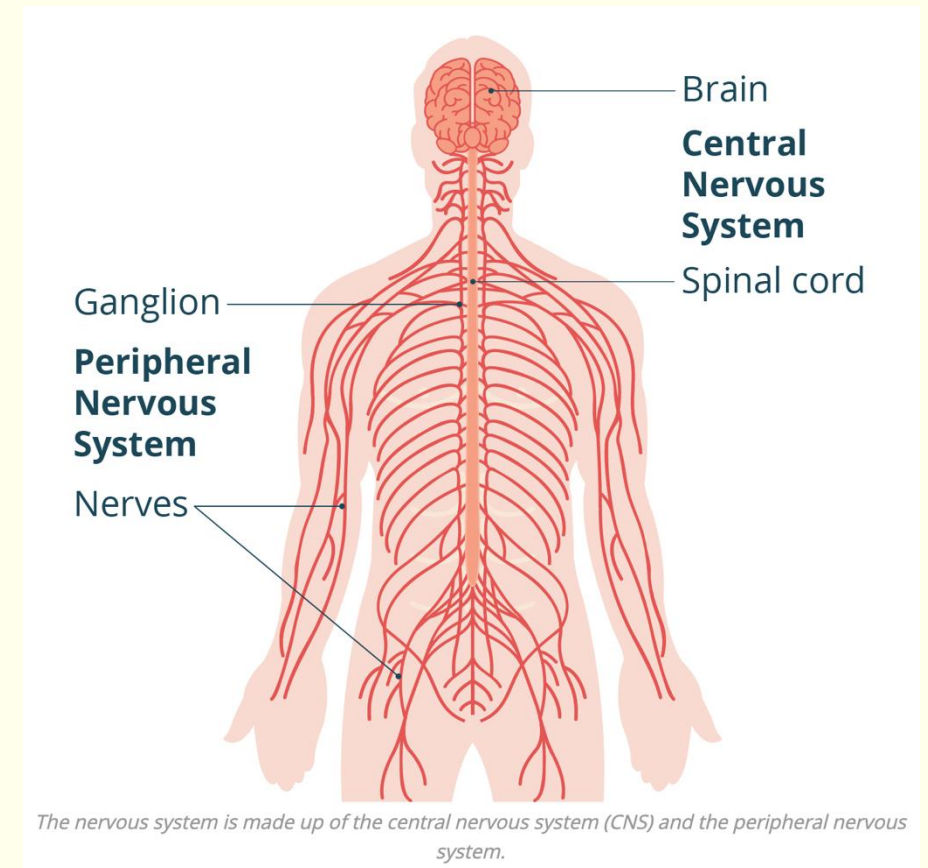
LOGY = Study of (from Greek 'logos')

NEUROLOGICAL = affecting the nervous system



What does Neurological relate to?

- **Neurological condition:** any disorder affecting the nervous system
- **Neurodegeneration:** progressive loss of nerve cell structure or function



The Nervous System

The nervous system is the body's communication network

It sends signals between the brain and every part of the body, controlling movement, sensation, and organ function

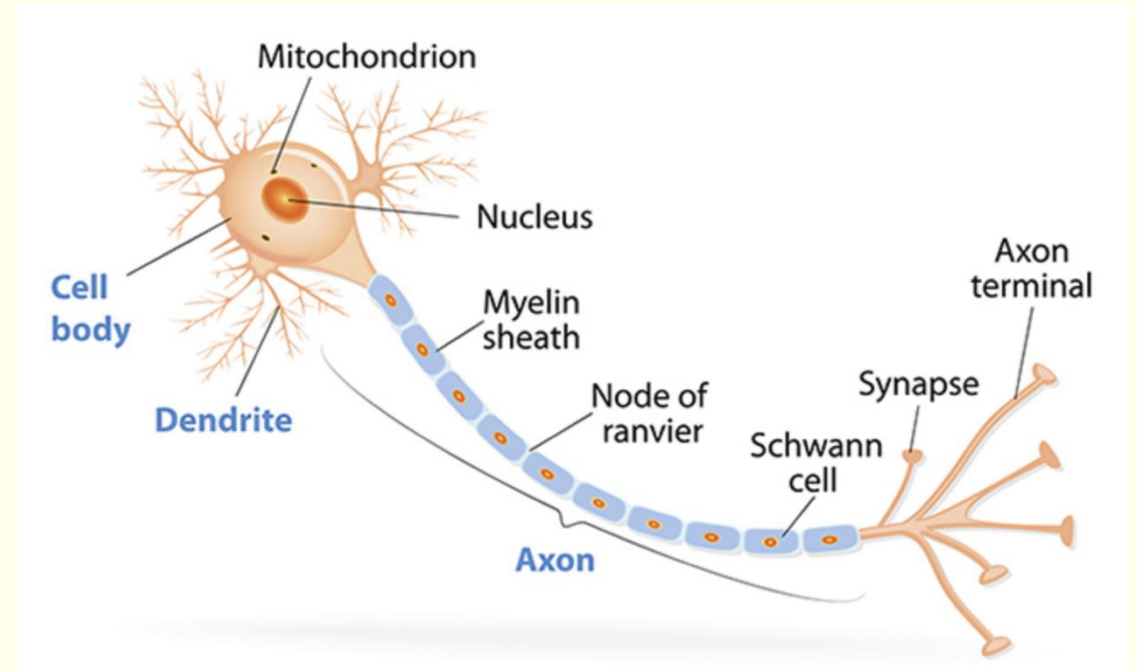
Central nervous system (CNS): the brain and spinal cord. This is the control centre

Peripheral nervous system (PNS): the network of nerves that runs from the spinal cord out to the muscles, skin, and organs

- Signals travel along specialised cells called neurons

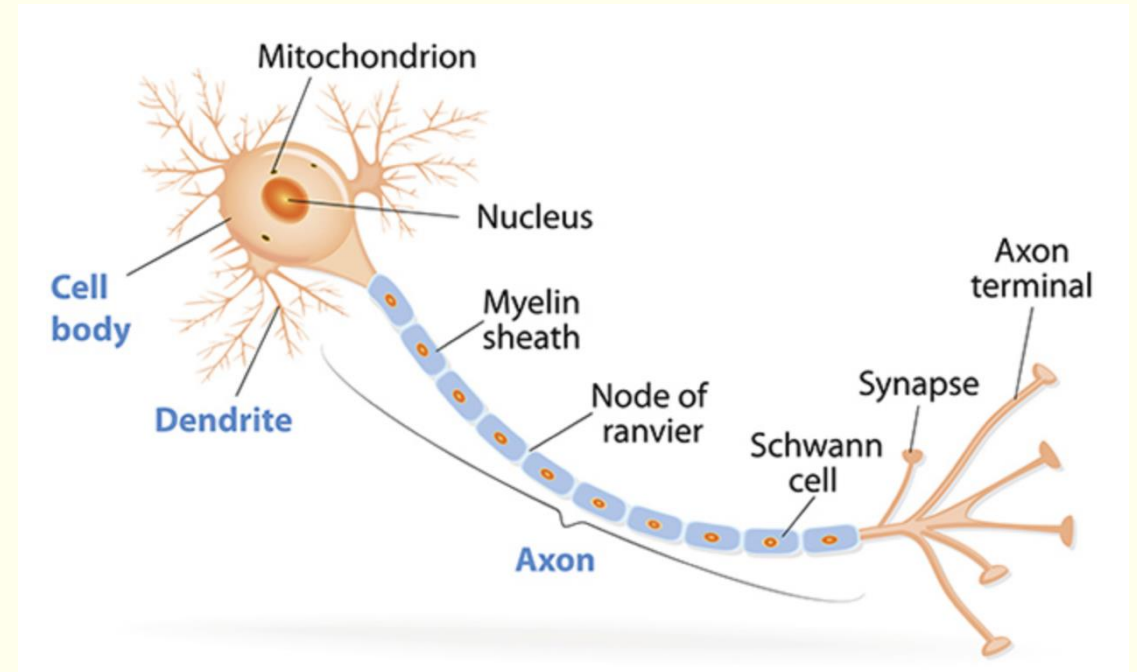
Neurons and Myelin: Why They Matter

- Neurons are the cells that carry nerve signals through the nervous system
- Each neuron has a long fibre called an axon that transmits the signal along its length
- Myelin is a fatty coating that wraps around the axon, like insulation on an electrical wire



Neurons and Myelin: Why They Matter

- Myelin speeds up signal transmission and protects the axon
- When myelin is damaged: signals slow down, become distorted, or fail to get through
- When neurons die: that function is permanently lost



Neurological Disorders

- Neurological disorders: in the top 10 leading causes of disability worldwide (GBD 2021)
- Over 3 billion people affected globally
- Conditions vary by mechanism:
 - Autoimmune/inflammatory (e.g. multiple sclerosis)
 - Neurodegenerative (e.g. Alzheimer's, Parkinson's)
 - Metabolic/vascular (e.g. vascular dementia)
 - Structural/electrical (e.g. epilepsy)

Neurological Conditions

Today we will be covering the following conditions:

- Multiple Sclerosis
- Dementia/Alzheimer's Disease
- Parkinson's Disease
- Migraine
- Motor Neurone Disease, Fibromyalgia, Epilepsy, Stroke, Peripheral Neuropathy

Multiple Sclerosis

Multiple Sclerosis (MS)

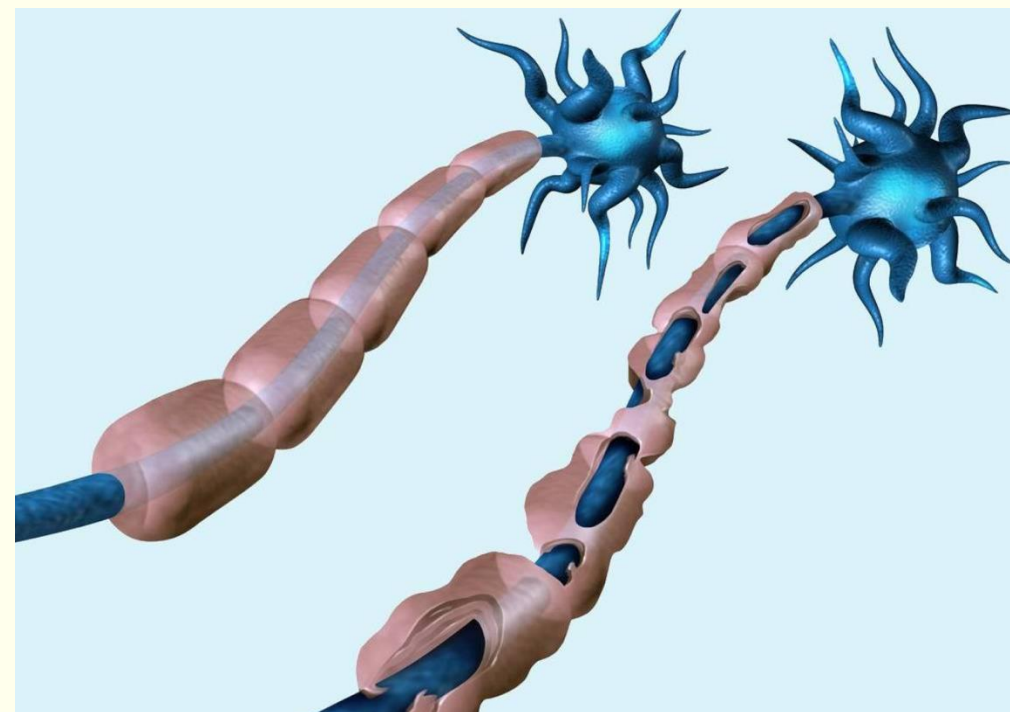
Multiple: from Latin multiplex (many, several)

Sclerosis: from Greek skleros (hard) + osis (condition of)

- Literal translation: 'many hardenings'
- Refers to multiple areas of scarring (sclerotic plaques) on the myelin sheath

What Is Multiple Sclerosis?

- A chronic, inflammatory, autoimmune disease of the central nervous system
- The immune system attacks and damages myelin (demyelination)
- Damage disrupts or blocks nerve signal transmission
- Over time: axon damage and neurodegeneration occur alongside inflammation



Multiple Sclerosis

PREVALENCE

Global: approximately 2.9 million people living with MS (MSIF Atlas of MS, 2023)

- Prevalence rising in every world region since 2013

UK: over 150,000, risen 13% since 2019 (MS Society, 2024)

- Women are 2.5 times more likely to be diagnosed than men

Multiple Sclerosis

INCIDENCE

Global: incidence is rising: most likely due to increased detection and reporting

UK: incidence: over 7,100 new diagnoses per year (approximately 135 per week)

- Mean age of diagnosis: 32 years
- MS is the most common non-traumatic cause of neurological disability in young adults

Multiple Sclerosis

AETIOLOGY

Multifactorial: genetic susceptibility + environmental triggers

Genetic: a specific gene variant affecting how the immune system recognises the body's own tissue is the strongest known genetic risk factor

- Genetic risk accounts for approximately 30% of MS risk; 70% is environmental

Multiple Sclerosis

AETIOLOGY

Key environmental/immune triggers:

- Epstein-Barr virus (EBV) infection: strongest environmental risk factor identified
- Low vitamin D levels/reduced sunlight exposure
- Smoking doubles the risk and accelerates progression
- Obesity in adolescence
- Gut microbiome alterations (emerging evidence)

Multiple Sclerosis

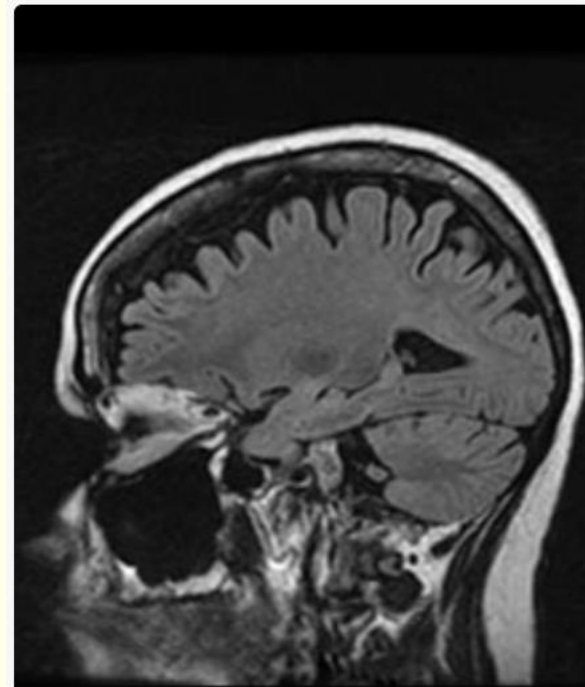
PATHOLOGY

- Immune cells cross into the brain and spinal cord, which are normally protected from immune activity
- Immune cells attack the cells responsible for producing and maintaining myelin
- Demyelination: loss of myelin sheath around axons

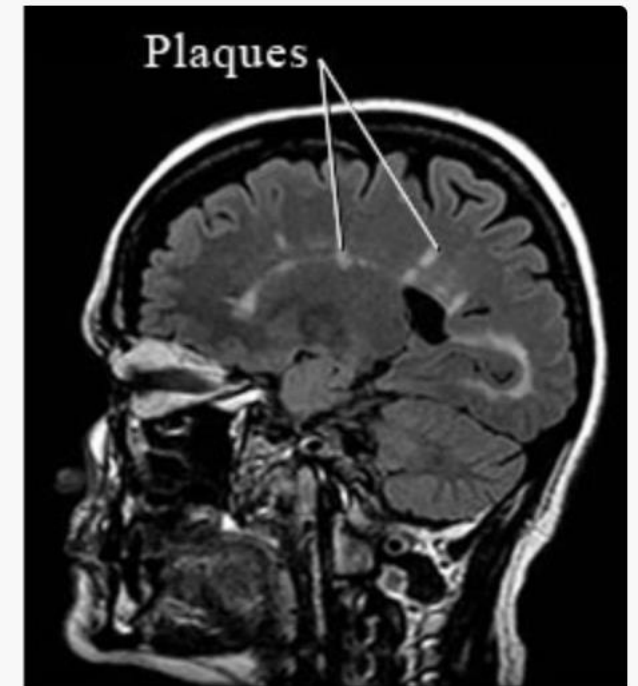
Multiple Sclerosis

PATHOLOGY

- Acute plaques: areas of active inflammation and demyelination
- Some remyelination is possible early in the disease
- Chronic plaques: axon damage and scarring that cannot be repaired, leading to permanent disability
- Disease progression: inflammation and neurodegeneration coexist over time



Normal non-contrast Brain
MRI sagittal view



MRI non contrast brain sagittal
view showing demyelinating
lesions typical for MS

Multiple Sclerosis

DISEASE TYPES

Relapsing-Remitting MS (RRMS): ~85% of diagnoses

- Discrete attacks called relapses, followed by periods of recovery called remission

Secondary Progressive MS (SPMS): develops from RRMS over time

- Steady worsening with or without relapses; affects around half of RRMS patients within 10 to 15 years

Primary Progressive MS (PPMS): ~10-15% of diagnoses

- Steady neurological decline from onset; no distinct relapses

Clinically Isolated Syndrome (CIS): first episode; may or may not become MS

Multiple Sclerosis

SIGNS AND SYMPTOMS

Highly variable: depends on which nerve pathways are affected

Most common early symptoms:

- Vision problems: optic neuritis, causing blurred vision, eye pain, or temporary loss of vision
- Sensory symptoms: numbness, tingling, pins and needles
- Fatigue: disproportionate and not relieved by rest; affects 80% of people with MS
- Limb weakness or coordination problems

Multiple Sclerosis

SIGNS AND SYMPTOMS

Later / progressive symptoms:

- Spasticity, bladder and bowel dysfunction, cognitive impairment, pain
- Mobility loss and dependency in advanced disease
- Electric shock sensation running down the spine when bending the neck forward, characteristic of MS but not used alone to confirm diagnosis

Risk Factors

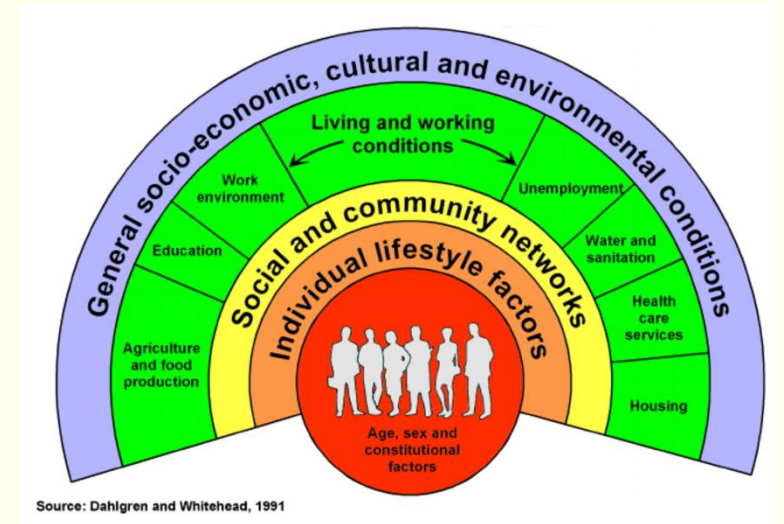
NON-MODIFIABLE

Genetics: carrying certain inherited gene variants significantly increases risk; having a first-degree relative with MS increases risk 20 to 40 times

Sex: female (2.5x higher risk)

Geography/latitude: higher risk further from equator

Ethnicity: more common in White European populations; increasing in other groups



Risk Factors

MODIFIABLE

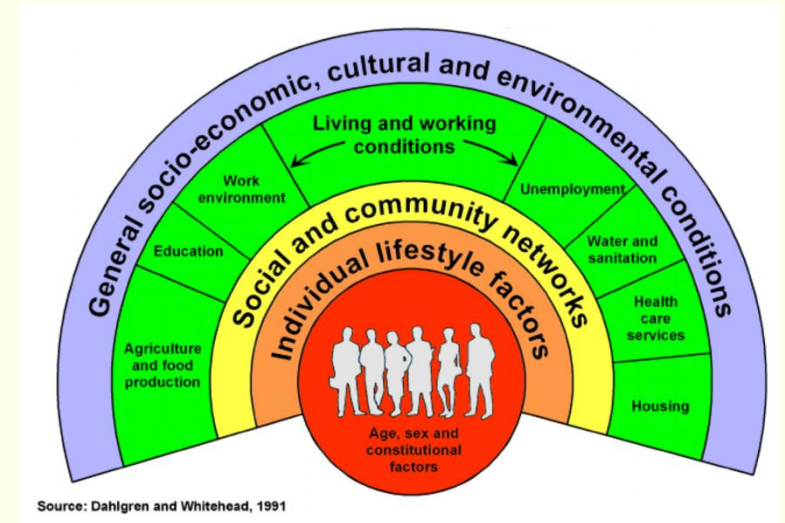
EBV infection: (near-universal, but risk management relevant to immune health)

Vitamin D deficiency: low levels associated with higher incidence and severity

Smoking: doubles risk; also accelerates progression

Obesity in adolescence: associated with increased risk in females

Stress: may trigger relapses in existing disease



Multiple Sclerosis

QUALITY OF LIFE

MS has a profound impact on health-related quality of life across physical, psychological, social, and economic dimensions

- **Physical**: fatigue, mobility loss, pain, bladder/bowel dysfunction
- **Psychological**: depression affects up to 50% of people with MS; anxiety is common
- **Social**: impact on employment, relationships, parenting, and social participation

Multiple Sclerosis

QUALITY OF LIFE

- **Employment:**
 - Mean age of diagnosis (32) means impact during peak career and family-building years
 - 50% of people with MS leave the workforce within 10 years of diagnosis
- **Role of exercise:** moderate physical activity improves fatigue, mood, and function in MS

DEMENTIA AND ALZHEIMER'S DISEASE

Dementia and Alzheimer's Disease

Dementia: from Latin demens

- Literal translation is 'out of one's mind'

Alzheimer's Disease: named after Dr Alois Alzheimer (German psychiatrist, 1864-1915)

- First described the condition in 1906 after examining brain tissue post-mortem

What is Dementia?

Dementia is an umbrella term for a group of symptoms caused by brain disorders

- Core features are decline in memory, thinking, problem-solving, language, and daily function
- Symptoms must be severe enough to interfere with daily life (distinguishes from normal ageing)

What is Dementia?

Not a single disease: caused by several different underlying conditions:

- Alzheimer's disease: 60-70% of cases
- Vascular dementia: ~15-20% (caused by reduced blood flow to the brain)
- Lewy body dementia and frontotemporal dementia: most of the remainder
- Alzheimer's disease is neurodegenerative and progressive; currently there is no cure

Dementia

PREVALENCE

Global: approximately 57 million people in 2019; projected to reach 153 million by 2050 (GBD 2019, Lancet Public Health)

UK: approximately 982,000 people, the UK's leading cause of death (Alzheimer's Society 2024)

- Over 76,000 deaths attributed to dementia in 2024, more than 1 in 10 of all deaths
- Projected to rise to 1.4 million in the UK by 2040 as the population ages

Dementia

PREVALENCE

- Two thirds (65%) of all people with **dementia** in the UK are women (Alzheimer's Research UK, 2022)
- Approximately twice as many women as men have **Alzheimer's** disease (Alzheimer's Society)
- Dementia has been the leading cause of death for women in the UK since 2011
- Longevity alone does not explain the disparity, sex-specific biological factors are under investigation

Dementia

INCIDENCE

Global: incidence increasing as populations age, particularly in low and middle income countries

UK: incidence: approximately 200,000 new diagnoses per year (Alzheimer's Society)

- MS is most commonly diagnosed in people in their 30s and 40s (mean 45y)
- MS is the most common non-traumatic cause of neurological disability in working-age adults

Wales: Early diagnosis rates are low: only 49% of people estimated to have dementia in Wales have a formal diagnosis, compared with 67% in England

Alzheimer's Disease

AETIOLOGY

Biological mechanism: abnormal protein build-up in the brain

1. A protein called amyloid is produced abnormally and cannot be cleared, forming sticky clumps called plaques outside nerve cell
2. A second protein called tau becomes damaged and forms tangled threads inside nerve cells, disrupting their internal structure
3. These changes trigger inflammation, disrupt communication between nerve cells, and eventually cause nerve cells to die

Alzheimer's Disease

AETIOLOGY

Genetic: A gene variant called APOE4 is the strongest known genetic risk factor: one copy roughly triples the risk; two copies increases it around twelve times

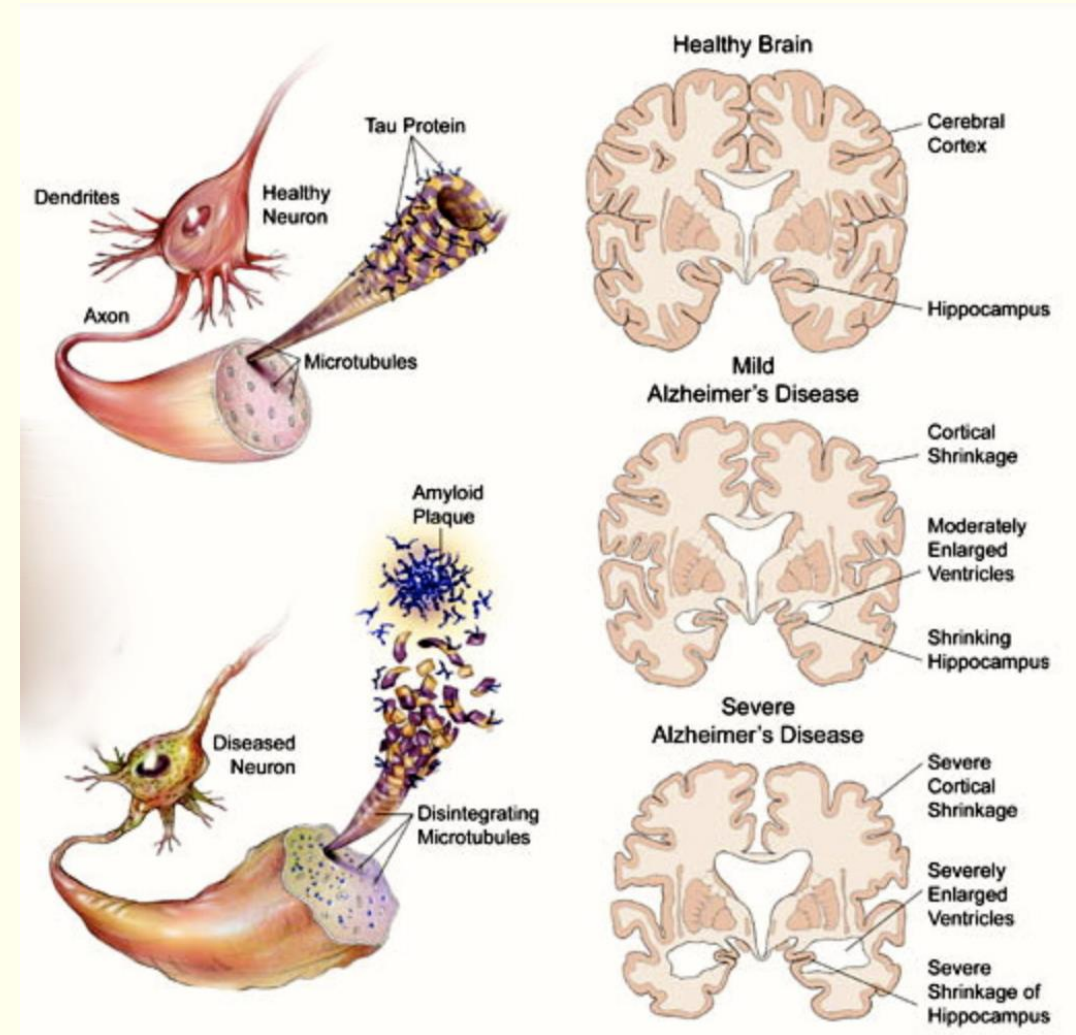
- Rare inherited gene mutations can cause Alzheimer's disease to run strongly in families, sometimes appearing in people in their 40s and 50s; this accounts for less than 1% of cases

Alzheimer's Disease

PATHOLOGY

Alzheimer's is characterised by two types of abnormal protein accumulation in the brain

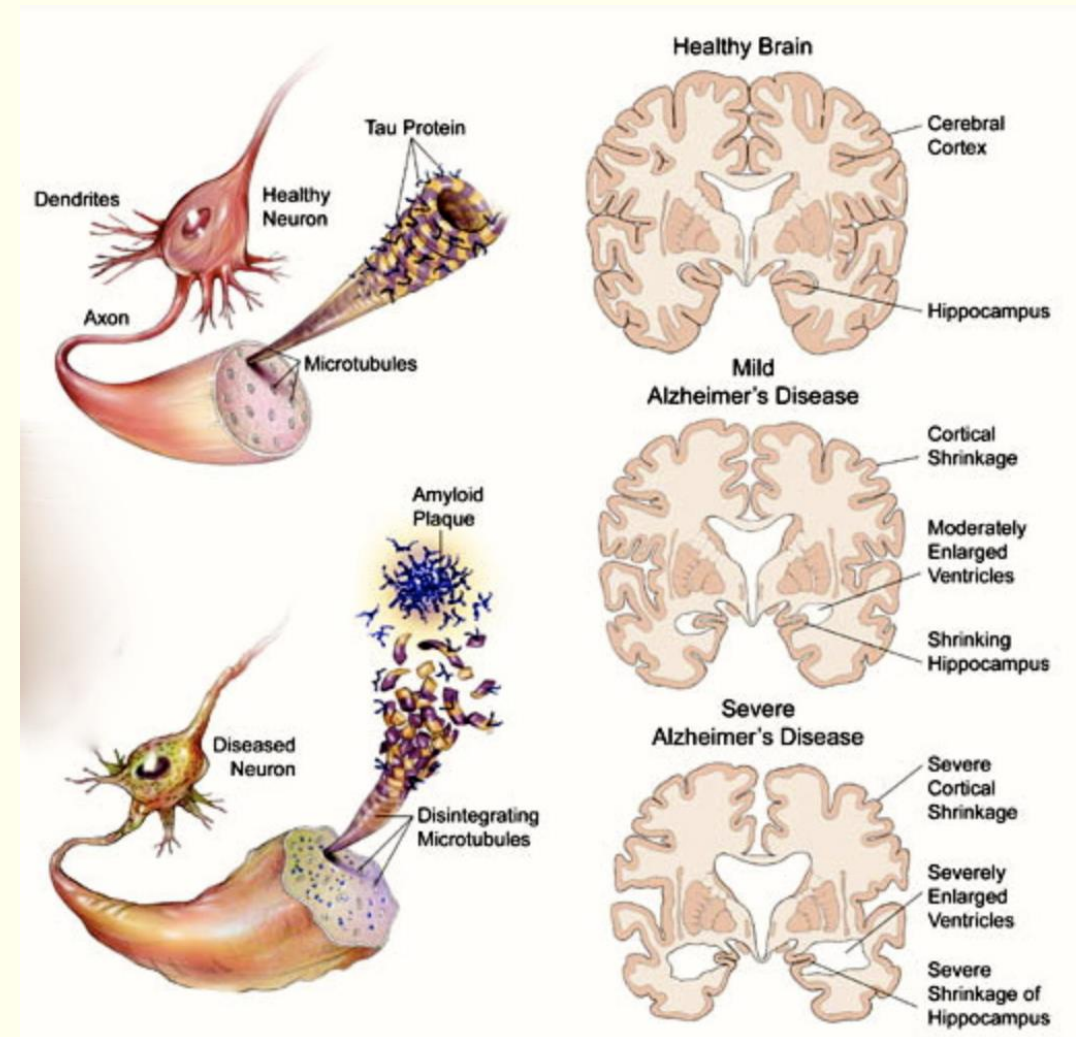
- Amyloid plaques: fragments of amyloid protein clump together outside neurons, blocking communication between cells
- Tau tangles: tau protein inside neurons becomes damaged and collapses, destroying the neuron's internal transport system



Alzheimer's Disease

PATHOLOGY

- As neurons die, the brain physically shrinks: this is called atrophy, and is visible on MRI
- Hippocampus shrinks first; ventricles enlarge as surrounding brain tissue is lost
- Atrophy spreads outward in a predictable pattern

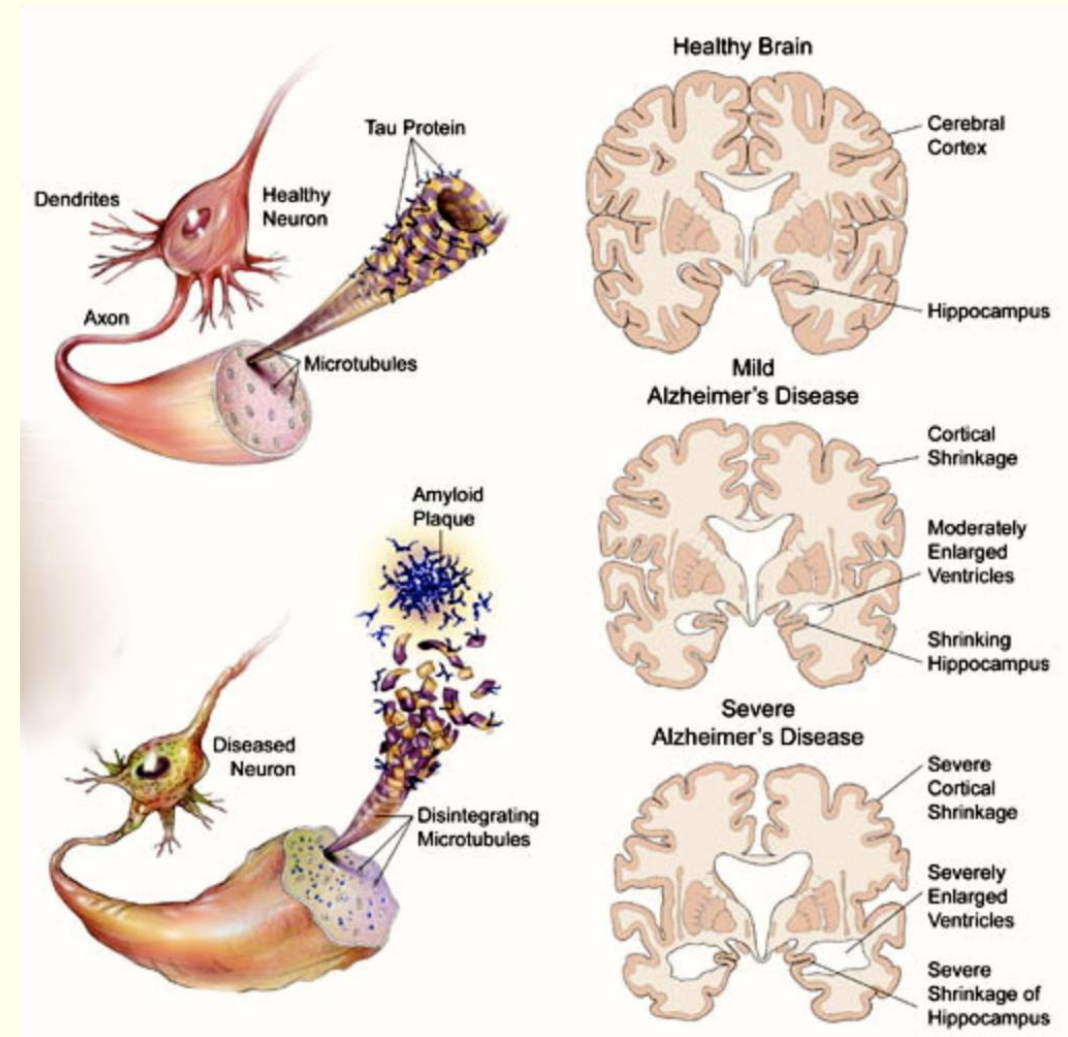


Alzheimer's Disease

PATHOLOGY

- Temporal lobes: language and recognition affected
- Parietal lobes: spatial awareness and calculation affected
- Frontal lobes: judgement, planning, and personality affected

Brain volume loss: approximately 1 to 3% per year in Alzheimer's vs 0.1 to 0.5% in healthy ageing



Alzheimer's Disease

SIGNS AND SYMPTOMS

Early symptoms:

- Short-term memory loss (forgetting recent conversations, names, events)
- Difficulty planning or problem-solving
- Getting confused with time or place
- Word-finding difficulties

Alzheimer's Disease

SIGNS AND SYMPTOMS

- Later symptoms:
 - Increasing disorientation; difficulty recognising familiar people
 - Behavioural and personality changes (agitation, withdrawal, suspicion)
 - Hallucinations, delusions
 - Loss of speech, mobility, and ability to perform basic functions

Average survival: 8-10 years from diagnosis, though varies widely

Risk Factors

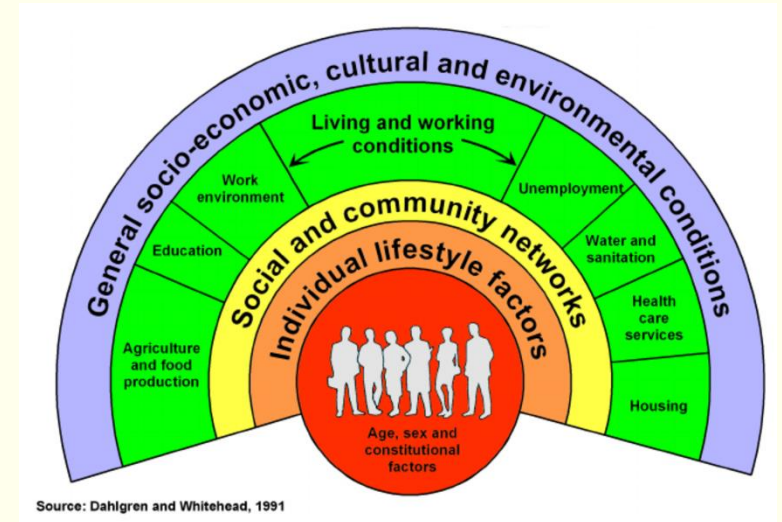
NON-MODIFIABLE

Genetics: inherited gene variants increase risk; rare familial mutations can cause early-onset disease

Age: risk doubles every five years after 65 (strongest risk factor)

Sex: women at higher risk (partly longevity, partly biological)

Down syndrome: people with Down syndrome have a significantly elevated risk of developing Alzheimer's disease and tend to develop it at a younger age

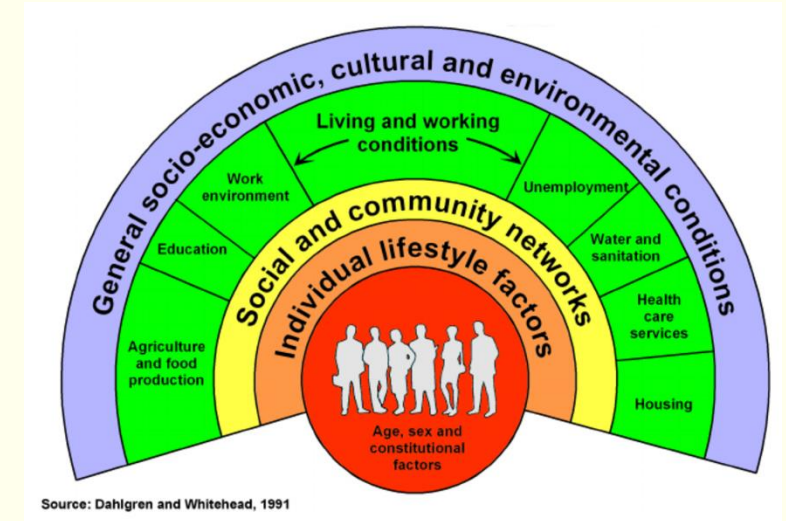


Risk Factors

MODIFIABLE

Lancet Commission (2024) identified 14 risk factors:

- Physical inactivity
- Smoking
- Obesity
- Hypertension
- Diabetes
- Excessive alcohol
- Low education
- Social isolation
- Depression
- Hearing
- Vision loss
- Air pollution
- Traumatic brain injury
- High cholesterol (newly added 2024)



Dementia and Alzheimer's Disease

QUALITY OF LIFE

Dementia is the most feared health condition in the UK (Dementia Attitudes Monitor)

- **Physical**: progressive loss of independence; falls risk; weight loss; dysphagia in later stages
- **Psychological**: depression and anxiety are common in early stages; grief, loss of identity
- **Social**: social withdrawal; loss of relationships; stigma

Dementia and Alzheimer's Disease

QUALITY OF LIFE

- **Economic:** costs the UK 42 billion pounds per year; 63% borne by individuals and families
- **Unpaid carer burden:** 670,000 unpaid carers in the UK providing dementia care
- **Diagnosis rate in Wales:** only 49%, meaning roughly half the people who need support do not have access to it

Break



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Motor Neurone Disease (MND)

Progressive degeneration of motor neurones

- Most common type: ALS (amyotrophic lateral sclerosis), often called MND in the UK
- From Greek a (without) + mys (muscle) + trophe (nourishment), meaning 'muscle wasting'
- Most common type: ALS (amyotrophic lateral sclerosis), often called MND in the UK
- UK: approximately 5,000 people living with MND at any one time
- Rapidly progressive: average survival 2 to 5 years from symptom onset
- Affects both upper and lower motor neurones: weakness, wasting, spasticity
- Cognitive and behavioural changes in 50% of cases; full frontotemporal dementia (FTD) in approximately 15%

Fibromyalgia

- Fibromyalgia: from Latin fibra (fibre) + Greek mys (muscle) + algos (pain)
- A central sensitisation syndrome: the nervous system, not the muscles, is the primary problem
- The brain amplifies pain signals: sensations that would not normally be painful, such as light touch or clothing on skin, are experienced as painful
- Symptoms: widespread chronic pain, fatigue, sleep disturbance, cognitive difficulties ('fibro fog')
- UK prevalence: approximately 1 in 20 people; globally 2-4% of the population
- Women are 7 times more likely to be diagnosed than men
- No tissue damage is visible on imaging; historically dismissed as psychosomatic

Epilepsy

- Epilepsy: from Greek epilepsia (seizure, to seize upon)
- A neurological disorder characterised by recurrent unprovoked seizures
- Caused by abnormal, excessive electrical activity in the brain
- Global prevalence: approximately 50 million (WHO 2023)
- UK prevalence: approximately 600,000 people (1 in 100)
- Highly manageable with anti-seizure medication in most cases
- Significant stigma remains; major impact on driving, employment, and independence

Peripheral Neuropathy

- Peripheral neuropathy: damage to the peripheral nerves (outside brain/spinal cord)
- UK: estimated 1 in 10 people over 55 affected to some degree
- Symptoms: numbness, tingling, pain, and weakness, typically in hands and feet ('glove and stocking' pattern)
- Diabetic peripheral neuropathy
 - Mechanism: chronic hyperglycaemia damages the small blood vessels supplying peripheral nerves
- Non-diabetic causes: alcohol excess, vitamin B12 deficiency, autoimmune disease, infections, medication

Stroke

- Stroke is a leading cause of acquired neurological disability worldwide
- Covered in full in Week 2 (cardiovascular disease session)
- Key connections this week:
 - Stroke as a cause of vascular dementia (15-20% of all dementia)
 - Post-stroke cognitive impairment overlaps with dementia diagnosis
 - Shared risk factors: hypertension, atrial fibrillation, diabetes, smoking



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Next Week: Mental Health and Chronic Disease

Questions?

