Parkinsonism and Parkinson's Disease

Parkinsonism refers to a combination of motor symptoms like those seen in Parkinson's disease, including slowed movements, stiffness, tremor, and loss of balance. Parkinsonism is a broad term that describes the symptoms. There are many causes, with Parkinson's disease being the commonest. Other conditions that present with parkinsonism can have different time frames, treatments, and outcomes. Although the symptoms are similar, the disorders have different causes, time frames, and outcomes.

Parkinson's disease (PD) is a long-term movement disorder in which nerve cells in the areas of the brain involved in controlling movements slowly break down and die. PD is best known for causing tremors and affecting movement, but it involves multiple other symptoms. As PD progresses through time, more nerve cells die, and the patient's movements get worse. At this time, there is still no cure for PD, but the symptoms can be managed.

Parkinsonism has been reported in adults with 22q11.2 deletion syndrome (22q11.2DS). A neurological assessment can help with early diagnosis and treatment.

Parkinsonism

Main symptoms

- Slowed movement (**bradykinesia**)
- Muscle stiffness (**rigidity**)
- **Tremors**
- Trouble with balance & coordination

Possible causes

- Damage in the brain cells that occur over time
- Medications (e.g. anti-psychotics)
- Toxins (e.g. certain pesticides)
- Limited blood supply to the brain
- Fluid build-up in the brain

Parkinsonisms That Are NOT Parkinson's Disease

- Vascular parkinsonism
- Drug-induced parkinsonism
- Dementia with Lewy bodies
- Wilson's disease
- Normal pressure hydrocephalus
- Corticobasal degeneration (CBD)
- Multiple system atrophy (MSA)
- Progressive supranuclear palsy (PSP)

For more info about each condition, see Parkinson's UK.

Treatment

- The treatment is based on the cause of each condition.
- **Many of the non-PD parkinsonian disorders do not respond well to medications that treat PD.**

Parkinson's Disease (PD) – Symptoms

PD happens when neurons (nerve cells) in the brain break down and die. Neurons in the midbrain produce **dopamine**, which decreases in amount when the nerve cells wear down or do not work as well over time. Body processes that depend on dopamine (see box on right) become slower and less well-controlled, and the patient begins to show symptoms of PD.

Symptoms of PD in 22q11.2DS are largely the same as those in the general population. The underlined symptoms below are common in PD in 22q11.2DS.

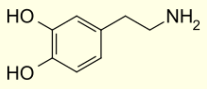
Symptoms of PD often start mildly on one side of the body before worsening and affecting both sides. Different people have different combinations of symptoms.

Movement-related

- Slowed movement (**bradykinesia**)
- **Tremors** when muscles are RESTING
- Muscle stiffness (**rigidity**)
- Unstable posture and balance
- Erratic movement (**dyskinesia**)
- Abnormal muscle posturing (**dystonia**)
- Changes in handwriting and speech

Non-movement-related

- Depression and/or **anxiety**
- Losing touch with reality (**psychosis**)
- Constipation
- Leaking pee
- Sleep problems (e.g. restless legs)
- Difficulty thinking, focusing



Dopamine is one of the most important neurotransmitters in the brain. It is involved in processes such as:

- Movement
- Memory
- Reward and motivation
- Behavior
- Learning and attention
- Sleeping and waking
- Mood

The nerve cells of people with PD tend to have clumps of abnormally folded protein called **alpha-synuclein**, which is linked to the process of **cell damage**.

Parkinsonism and Parkinson's Disease (continued)

PD – Diagnosis

It takes time and multiple assessments to diagnose a movement disorder accurately. **Although PD and other forms of parkinsonism have similar symptoms, they have different causes and need different treatments.** Your healthcare provider will ask about your medical history and assess you through:

- A neurological exam to check your thinking, mental abilities, senses, coordination, reflexes, and motor function
- Blood and lab tests to rule out other conditions
- Brain imaging to rule out other conditions
- Short low-dose PD medicine to see if you get better
- Tests to visualize the abnormally folded alpha-synuclein protein
  - ➢ This can be done by a needle puncture (spinal tap) to examine the fluid that bathes the brain (cerebrospinal fluid), or through examination of a piece of the skin (skin biopsy). These tests are still being done only on a research basis even in the general population.
  - ➢ The presence of abnormally folded and clumped up alpha-synuclein protein supports a diagnosis of PD. However, these techniques have only been applied to the general public, and still need to be researched in those with 22q differences.

PD – Treatments and Outlook

PD currently does not have a cure, but its symptoms can be **managed using individualized treatment plans**.

- Medications that **deal with the lack of dopamine** by increasing its production, mimicking its effect, or preventing its breakdown
- Medications that **manage PD symptoms** (but not the cause)
- **Deep-brain stimulation** – uses an implanted device to send a mild electric current to the areas of the brain that malfunction
- Experimental treatments – e.g. stem cell transplant, neuron-repair, gene therapy, guided ultrasound etc.

PD gets worse with time, and the treatments (especially some medications) usually need to be increased or adjusted depending on how severe the symptoms are. Living independently tends to become more challenging as the symptoms progress.

PD and 22q11.2DS

PD was found in 1.8% of adults with 22q11.2DS.

When uncertain diagnoses of PD were included, PD was found in 3.4% of adults with 22q11.2DS (any age) and 23.7% of adults with 22q11.2DS who were at least 50 years old.

Among adults with 22q11.2DS, PD was about equally common in males and females.

PD risk: 20- to 70- times higher in 22q11.2DS than in the general population.

Average age of PD onset:

- General: 60 - 70 years
- 22q11.2DS: ~45 years (range 20 - 66 years)

Some of these factors may increase the risk of PD:

- **Genetics** (family history)
- **Toxins** (some illegal drugs, pesticides)
- 22q11.2DS: **missing genes** involved in the:
  1. processing of **dopamine**
  2. functioning of the **mitochondria** (energy generators inside our cells)

Missing these genes can increase the risk of neuron damage and the risk of PD.

Resources

- Parkinson's disease – Mayo Clinic
- Parkinson's disease – Cleveland Clinic
- Parkinsonism – Cleveland Clinic
- Types of Parkinson's – Parkinson's UK
- Lumbar Puncture – Cleveland Clinic
- Prevalence of Parkinson's Disease in 22q11.2 Deletion Syndrome: A Multicenter Study (2025)
- 22q11.2 Deletion Syndrome–Associated Parkinson's Disease (2018)
- Updated clinical practice recommendations for managing [children][adults] with 22q11.2 deletion syndrome – 2023

❖ Dopamine structure diagram from Wikimedia.org

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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