

Huntington Disease

Huntington disease is a genetic condition where a faulty gene makes an abnormal protein that slowly destroys brain cells. The disease causes uncontrolled movements, memory loss, and mood changes that worsen over time. It usually begins in mid-life and is passed from parent to child.

THE ONE-LINE TRUTH

Huntington disease is an inherited brain disorder where mutant huntingtin protein kills neurons in the striatum, causing progressive movement problems, dementia, and psychiatric symptoms with no cure.

The chain — how it unfolds

1. Mutant huntingtin protein begins accumulating in striatal neurons during childhood or young adulthood — why: The inherited CAG expansion causes lifelong production of toxic protein, but it takes years of accumulation before enough damage occurs to cause symptoms
2. Medium spiny neurons in the striatum start dying, initially without symptoms (presymptomatic phase) — why: The brain's compensatory reserve masks early neuron loss, and functional imaging shows changes 10-15 years before symptoms appear
3. Chorea (involuntary, jerky, dance-like movements) emerges as striatal inhibition fails — why: Loss of GABAergic neurons removes the brake on unwanted movements, allowing excessive motor activity from intact dopamine pathways to cause uncontrolled motions
4. Cognitive decline begins with executive dysfunction (trouble planning, organizing, and multitasking) — why: Striatal-frontal circuits that coordinate thinking and decision-making degenerate, and cortical neurons in frontal lobes also accumulate mutant huntingtin
5. Psychiatric symptoms including depression, irritability, apathy, and impulsivity worsen — why: Loss of neurons in limbic circuits (brain pathways controlling emotion) and disrupted serotonin and dopamine signaling alter mood regulation
6. Motor symptoms progress to bradykinesia (slow movement), rigidity, and dystonia (sustained muscle contractions) — why: As striatal degeneration becomes severe, the motor system loses both the ability to suppress unwanted movements and to initiate desired movements smoothly

7. Severe dementia develops with profound loss of memory, language, and self-care ability — why: Progressive cortical atrophy (brain shrinkage) affects multiple lobes, destroying the neural networks needed for higher cognitive functions
8. Death occurs 15-20 years after symptom onset, typically from aspiration pneumonia, fall injuries, or malnutrition — why: Complete loss of motor control and swallowing coordination, combined with severe immobility and cognitive impairment, leads to life-threatening complications that cannot be prevented once neurodegeneration is advanced

Spot it — what you see, and why

- Chorea (involuntary jerky movements): Loss of striatal neurons (nerve cells in the basal ganglia that control movement) removes inhibition of unwanted movements
- Difficulty walking and frequent falls: Damage to basal ganglia (deep brain structures) impairs coordination, balance, and smooth motor control
- Depression and apathy: Degeneration of frontal cortex (front brain region) and striatum disrupts circuits that regulate mood and motivation
- Difficulty swallowing (dysphagia): Chorea and incoordination affect throat muscles, impairing safe passage of food and liquid
- Irritability and angry outbursts: Frontal lobe atrophy (shrinkage) disrupts impulse control and emotional regulation pathways
- Memory problems and slowed thinking: Loss of striatal-frontal connections impairs working memory and processing speed

Red flags — act now

- New or worsening suicidal thoughts (suicide risk is 4 to 6 times higher than general population)
- Choking episodes or recurrent pneumonia (suggests aspiration and need for swallow evaluation and feeding tube discussion)
- Rapid weight loss despite adequate intake (signals disease progression and increased metabolic demands)
- Severe agitation, aggression, or psychosis (requires urgent psychiatric evaluation and medication adjustment)

Treat it — in this order

1. Genetic counseling before and after testing → Helps patient and family understand inheritance pattern, recurrence risk (50 percent for each child), and implications for life planning

2. Antidepressant therapy (SSRI or SNRI) → Reduces depression, anxiety, and irritability by increasing brain serotonin and/or norepinephrine (mood-regulating chemicals)
3. Physical therapy and fall prevention → Improves balance, strengthens muscles, teaches safe mobility strategies, and delays need for wheelchair
4. High-calorie diet (1.5 to 2 times normal calorie needs) → Prevents malnutrition and weight loss by compensating for hypermetabolism (increased calorie burning) from constant movements
5. Speech and swallow therapy → Teaches safe swallowing techniques, diet modifications (thickened liquids, soft foods), and communication strategies

Your top priorities at the bedside

1. Monitor for chorea (involuntary jerky movements) and fall risk — why: Huntington disease causes progressive motor dysfunction with unpredictable movements that increase fall and injury risk, requiring environmental safety modifications and supervision.
2. Assess swallowing ability and aspiration risk at every meal — why: Trouble swallowing (Dysphagia) (trouble swallowing) worsens as the disease progresses, leading to aspiration pneumonia, malnutrition, and choking, which are leading causes of death in Huntington disease.
3. Screen for depression, suicidal ideation, and psychiatric symptoms daily — why: Huntington disease causes psychiatric changes including depression, irritability, and psychosis; suicide risk is 4 to 6 times higher than the general population, often occurring early in the disease.