SNS COLLEGE OF NURSING

SARAVANAPATTI,COIMBATORE-35

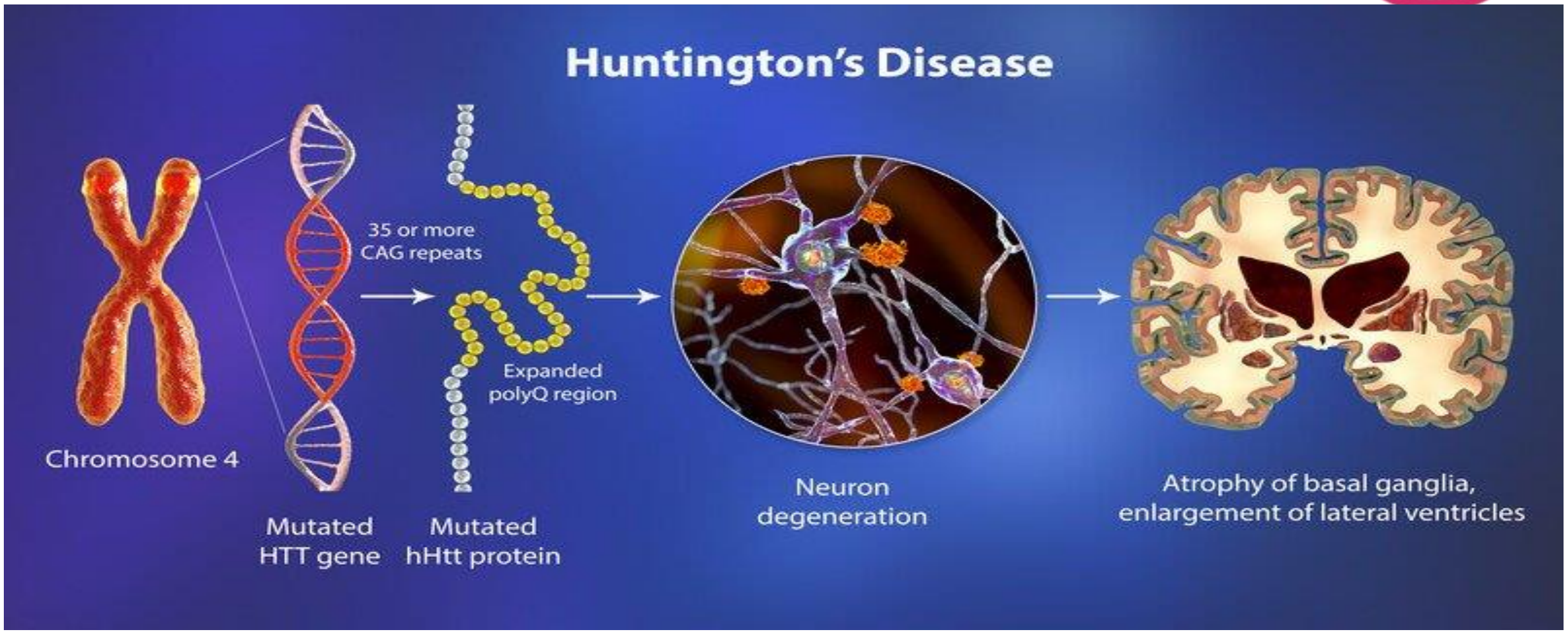
DEPARTMENT : Department of Nursing

COURSE NAME : B. Sc. (Nursing) II Year IV Semester

SUBJECT : Genetics

UNIT III : Genetic conditions of adolescents and adults

TOPIC : Huntington‘s disease


Huntington's Disease
35 or more
CAG repeats
Expanded
polyQ region
Chromosome 4
Mutated
HTT gene
Mutated
hHtt protein
Neuron
degeneration
Atrophy of basal ganglia,
enlargement of lateral ventricles

Huntington disease
Introduction:

❖Huntington disease (HD) is a progressive brain disorder that causes the breakdown of nerve cells in certain areas of the brain

Definition

- It is also a basal ganglia disease causing a hyperkinetic movement disorder known as chorea.
- Mental abilities generally decline into dementia, depression, apathy, and impulsivity at times.
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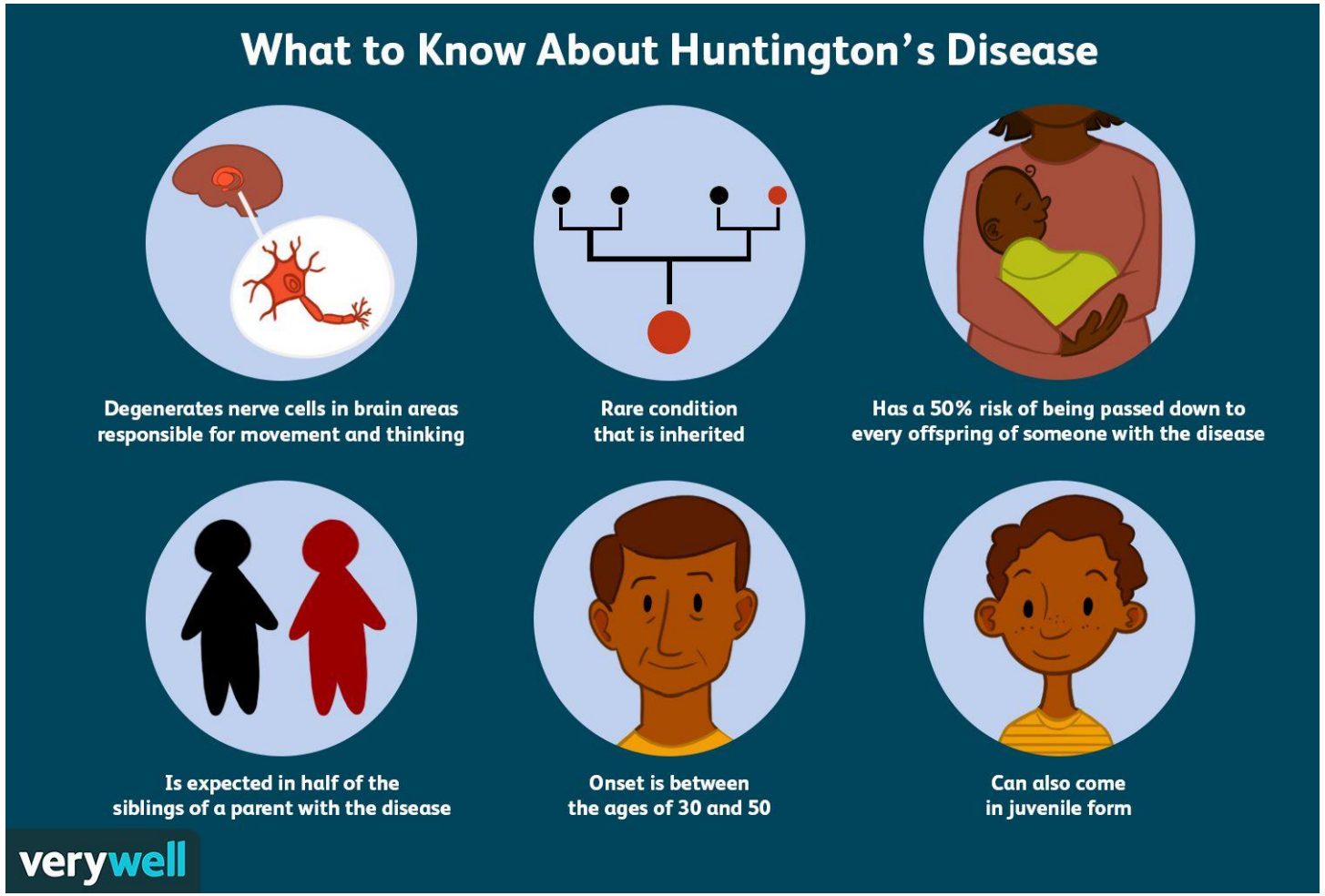
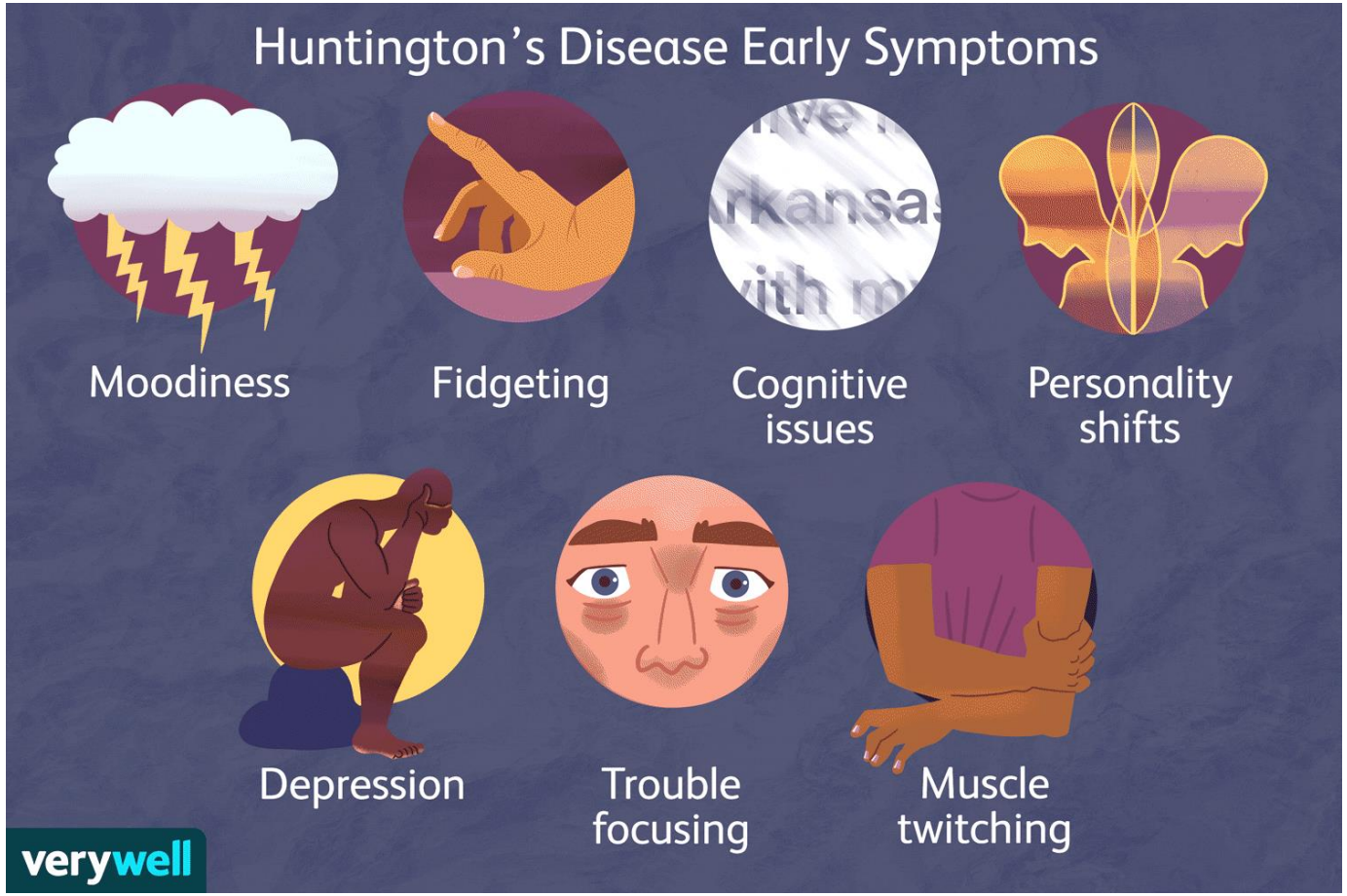
Etiology



- Huntington's disease is caused by an inherited defect in a single gene. Huntington's disease is an autosomal dominant disorder, which means that a person needs only one copy of the defective gene to develop the disorder.
- With the exception of genes on the sex chromosomes, a person inherits two copies of every gene — one copy from each parent. A parent with a defective Huntington gene could pass along the defective copy of the gene or the healthy copy. Each child in the family, therefore, has a 50 percent chance of inheriting the gene that causes the genetic disorder.

Signs and symptoms



Diagnosis

- History collection
- Physical examination
- Neurological Assessment
- Psychiatry Assessment
- CT Scan
- MRI Scan

Treatment

- No treatments can alter the course of Huntington's disease.
- Collaborative goals focus on:
  - - Reducing symptoms
  - - Preventing complications
  - - Providing support and assistance to the patient and significant others
- drugs to treat some symptoms may result in side effects that worsen other symptoms.

- Medications for movement disorders
- Drugs to treat movement disorders include the following:
- Tetrabenazine (Xenazine) is used to suppress the involuntary jerking and writhing movements (chorea) associated with Huntington's disease.
- Antipsychotic drugs, such as haloperidol (Haldol) and chlorpromazine, have a effect of suppressing movements (treat's chorea)
- Other medications that may help suppress chorea include amantadine, levetiracetam (Keppra) and clonazepam (Klonopin).

Treatment cont…

- Thickening agents can be added in food (soups)
- To eat slowly and smaller chips of fluid to prevent choking.
- If eating is impaired through mouth then need to plan for percutaneous endoscopic gastrostomy.
- Physical therapist – non medication management (strengthening , stretching and cardiovascular exercises)
- Breathing and airway clearance technique.
- Walking Aids
- Rehabilitation.

Medications

1. **Tetrabenazine** is a drug for the symptomatic treatment of hyperkinetic movement disorders
2. **Amantadine**, sold under the brand name **Gocovri** among others, is a medication used to treat dyskinesia associated with parkinsonism and influenza caused by type A influenza virus, though its use for the latter is no longer recommended because of widespread drug resistance
3. Antipsychotics
4. Benzodizapines
5. Anti parkinsonism
6. Valporic Acid
7. Eicosapentaenoic acid
8. SSRI

7.

8.

Summary

Genetic counseling benefits these individuals by updating their knowledge, seeking to dispel any unfounded beliefs that they may have, and helping them consider their future options and plans.

The Patient Education Program for Huntington's Disease has been created to help educate family members, caretakers, and those diagnosed with Huntington's disease

Conclusion

- The largest risk is pneumonia, which causes death in one third of those with HD.
- As the ability to synchronize movements deteriorates, difficulty clearing the lungs, and an increased risk of aspirating food or drink both increase the risk of contracting pneumonia.
- The second-greatest risk is heart disease, which causes almost a quarter of fatalities of those with HD.

Assessment

- Define Huntington's Disease.
- List out any 4 causes for Huntington's Disease
- Enumerate the treatment for Huntington's Disease
- Plot out the signs and symptoms of Huntington's Disease

References

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Thank you !