HUNTINGTON'S DISEASE

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ETHIOLOGY

- Autosomal dominant disease
- >36 CAG triplet repeats in huntington gene on chromosome 4
- Mutated proteins aggregating in nucleus caudatus causing symptoms of diminished brain functions
- Anticipation : increased numbers of CAG repeats in subsequent generations (->earlier onset of symptoms)

EPIDEMIOLOGY

- Worldwide prevalence 5-10 cases per 100.000 people
- Similar risk for male and female
- Peak incidence : ~ 40 years of age

SYMPTOMS – INITIAL STAGE

- Movement dysfunction (chorea, oculomotor disorders..)
- Hyperreflexia
- Sensory deficits
- Autonomic deficits (hyperhidriosis, urinary incontinence)

SYMPTOMS – ADVANCED STAGE

- Movement dysfunction (hypokinetic motor symptoms, akinetic mutism, motor impersistence, dysarthria and dysphagia)
- Dementia
- Depression (suicidal thoughts)
- Aggression and psychosis
- Apathy
- Cachexia (due to dysphagia and high energy consumption)

DIAGNOSIS

- Patient history
- Genetic testing (also for prediction of disease in embryo)

TREATMENT

- Ongoing physiotherapy, ergotherapy, logotherapy and psychotherapy (if needed)
- Chorea movement (tetrabenazine, clozapine, amantadine)
- Psychosis (atypical neuroleptics - clozapine)
- Depression (SSRIs – citalopram)

PROGNOSIS

- Progressive disease non curable
- Mean duration of illness : app. 20 years
- Cause of death : aspiration pneumonia, respiratory insufficiency, suicide

THANK YOU FOR YOUR ATTENTION