HUNTINGTON’S DISEASE:
A CLINICAL OVERVIEW OF ADVANCED DISEASE SYMPTOMS AND TREATMENT OPTIONS

John O’Sullivan
Associate Professor of Medicine
Royal Brisbane Clinical School, University of Queensland
Senior Visiting Neurologist
Royal Brisbane & Women’s Hospital
St Andrew’s War Memorial Hospital
Wesley Hospital
Huntington’s disease - introduction

• uncommon inherited, degenerative brain disorder

• results in loss of both mental and physical control

• considerable variability in symptoms and progression

• partly treatable but incurable

• requires multidisciplinary input

• often requires institutional care
Huntington’s disease - history

- 1872 described by Dr George Huntington
- 1979 study of >4000 patients in Venezuela
- 1983 HD gene linked to chromosome 4
- 1993 HD gene discovered
How common is HD

• 6-10/100,000 (= 300 – 500 pts in Qld) - uncommon

• “At risk” 1000-1200 in Qld

• New cases 20/yr in Qld
Clinical Features – age of onset

• Typically adult-onset disorder
• average onset 35-44 y (range 2-80 y)
  • Often working, child-bearing age, driving

• <20% have first display symptoms after age 50 y
  • have a slower progression
• <10% cases are juvenile HD onset before age 20 y
  • have a faster progression

• Average survival after onset is 15-18 y (range 5-25 y)
• average age at death 54 y
Clinical Features

I. Motor or movement
II. Behaviour or mood
III. Cognition or memory

Initial / first symptom & progression varies between patients
  - unrelated to age or symptoms in other family members
Clinical Features – early motor

- **Chorea** – unpredictable rapid involuntary jerky movements in different body parts
- **Motor impersistence** – can’t maintain steady muscle contraction
- Poor motor **planning & sequencing**
- Reduced limb movement **speed** and **co-ordination**.
- Abnormal control of eye movement

- “**Jerky, clumsy, slow**”
Clinical Features – late motor

- **Dystonia** – posturing or slow uncoordinated movement
- **Parkinsonism** – slowing of repetitive movements, stiffness
  - More common and earlier in young-onset patients
- **Stereotropies** – repetitive patterned movements
- **Postural instability** - falls
- **Immobility**
- **Incontinence**
- **Akinetic–rigid subtype** earlier in young onset patients
Clinical Features – speech

- Articulation
  - Imprecise consonants, distortions and irregular breakdowns

- Phonation-Respiration
  - Sudden forced inhalation/exhalation, voice stoppages, transient breathiness or strained-harsh (especially if dystonia presentation) voice quality, excess pitch or loudness variation

- Prosody or fluency
  - variable rate, inappropriate silences

- Language deficits
Clinical Features – swallow

- Irregular **upper limb** delivery of food / liquid to mouth
- Poor **head** and **neck** posture and involuntary mvt
- Involuntary **tongue** movement and discoordination
- Absent / inefficient chewing – food retention (**jaw** / **mouth**)  
- Food/liquid spillage out or into **airway**
- Inefficient swallow timing and coordination

- *Prolonged mealtime difficult in residential care*
Clinical Features – nutrition

- Weight loss common
- Poor access to nutritious diet
  - Social / behavioural
- Involuntary movements expend energy
- Impaired swallow limits food options
- Poor motivation to eat
Clinical Features – behavioural

- Irritability, short-tempered, outbursts and aggression
- Sleep disturbance
- Apathy – poor motivation
- Impulsivity, disinhibition
- Anxiety
- Depression, suicide
- Obsessive compulsive behaviour
- Psychosis – paranoia, hallucinations
Clinical Features – cognitive

- Visuospatial
  - Poor visual scanning, perception and judgment

- Executive dysunction:
  - Cognitive planning and sequencing, spatial working memory, cognitive flexibility and shifting set, concentration

- Memory impairment:
  - Slowed learning rates but preserved retention rates

- Language:
  - Word finding deficits, decreased phrase length and complexity, decreased comprehension of complex information
HD – treatment / management

- Involve, listen to, and support the family and carers
- AHDA-Qld invaluable resource
- Utilise formal driving assessment
- Plan early – EPOA, AHD, superannuation, Centrelink
- Family planning, financial planning

- Maintain optimistic but realistic approach to new treatment, trials and research
HD – treatment / management

- Drug therapy
  - Treat symptoms not the disease itself
  - Few symptoms respond to drug therapy
  - Side-effects relatively common
- Address alcohol, recreational drug use
- Rehabilitation limited by cognitive impairment
  - Simple strategies reinforced by family/carer/staff can help
- Explore behavioural options
- Speech & occupational therapy and dietetics helpful
HD – drug therapy

**CHOREA** - Dopamine blocking agents
- Antipsychotics: risperidone, haloperidol, olanzapine –
  - sedation, weight gain – but may help irritability and sleep
- Tetrabenazine
  - sedation, depression

**ANXIETY DEPRESSION IRRITABILITY OCD (APATHY)**
- Serotonin reuptake inhibitors > tricyclic antidepressant
  - Somnolence/insomnia, libido change, agitation
Advanced HD – management

- Accept sleep disturbance / daytime somnolence
- Controlling involuntary movement may result in sedation
- Restraints / protectors may be necessary
- Involuntary movements mistaken for aggression
- Make time for meals
- Rarely utilise PEG feeding
- Maintain routines
- Consider palliative care
HD – family member challenges

• Care for affected in-law(s) – parents / siblings
• Predictive testing process
• Fertility, risk to children, childcare, care for affected children
• Behavioural - irritability, aggression, inflexibility, alcohol
  - apathy, depression, anxiety
• Sleep disturbance – insomnia / daytime somnolence
• Occupational limitations / finances
• Driving / transport
• Reduced social interactions, communication, intimacy
• Residential placement – limited, guilt, advocacy