Palliative management of 14 patients with Creutzfeldt – Jakob Disease (CJD)

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CJD: “Proteins behaving badly”

- ‘TRANSMISSIBLE SPONGIFORM ENCEPHALOPATHY’
- RARE - one in a million, 35 cases per yr in Aus
- FATAL
- RAPID DEGENERATION – “Dementia on fast forward”
- AUTOPSY for confirmation
- TENACIOUS
- HUMANS & ANIMALS
Types of CJD

• **Sporadic** - (85 - 90%) just happens!
  - 50 - 70 year age group, survival 3 - 6 mths

• **Genetic** - (10 - 15%), autosomal dominant
  - 30 - 60 year age group, survival 2 - 5 yrs

• **Acquired** - (1%)
  - **Iatrogenic** - explanted materials, surgical instruments
  - **Variant** - BSE
Got it! Mad Cow Disease, right?

“CJD not tested for, very unlikely in setting of being a vegetarian”
Why were we interested to look at patients with a diagnosis of presumed CJD and their families?

• 14 patients from 2011-2014
• Limited published information
• Health services – room to improve
• Issues faced by patients and families
What issues are faced by patient and families?

- Complex, rapidly escalating symptoms
- No cure
- Unfairly stigmatised
- Sporadic vs Genetic
- Media
Retrospective audit of 14 patients with presumed CJD

Demographics
10 Male
4 Female

Age
Median age 67 years
Range 54 - 58 years

Comorbidities
Nil significance

Characteristics
Diverse ethnicities & occupation
How long was the patients’ journey?

Data obtained for 10 patients

- < 1 month: 10%
- 1 - 2 years: 10%
- 2 - 3 years: 10%
- 1 - 3 months: 40%
- 3 - 6 months: 20%
- 6 - 12 months: 10%
- 1 - 2 years: 10%
35 DAY ROLLER COASTER RIDE

- D1 PSYCH - 'bizarre behaviour’, ?Psychosis, CT Brain NAD, Failure to respond to Mx, ?organic

- D15 GEN MED - 1:1, FNC, hallucinating, dysphagic, Fall, incontinent, fluctuating GCS, VCAT, Code Grey, ?Encephalitis, MRI Brain NAD, LP

- D23 NEURO - “CJD Likely”- aphasic, rigid, terror stricken, myoclonic jerks. For EEG

- D26 - Family meeting - informed likely CJD, decision for placement.
• D29 - s/b Palliative Care - rigid, non verbal, ?PCU

• D30 - Stopped eating, CTCAP & rpt MRI Brain - “Probable CJD”, active Mx cont

• D31 - Family request pall. approach, fluctuating GCS, MET Call, seizures

• D33 - s/b PCT - ‘too unwell to transfer’

• D34 - PALLIATIVE CARE UNIT - arrives late pm unresponsive

• D35 - 0500 RIP, daughter present
How did these patients present?

The presenting symptoms were very diverse

- Gait
- Cognition
- Personality
- Restlessness
- Speech
## Investigations performed

<table>
<thead>
<tr>
<th>INVESTIGATIONS</th>
<th>% OF PTS</th>
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<tbody>
<tr>
<td>MRI</td>
<td>93</td>
</tr>
<tr>
<td>LUMBAR PUNCTURE</td>
<td>86</td>
</tr>
<tr>
<td>CT BRAIN, SYPHILIS</td>
<td>57</td>
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<tr>
<td>EEG</td>
<td>64</td>
</tr>
<tr>
<td>HSV</td>
<td>43</td>
</tr>
<tr>
<td>AUTOIMMUNE, THYROID</td>
<td>36</td>
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<tr>
<td>HIV</td>
<td>36</td>
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<tr>
<td>PARANEOPLASTIC, HEAVY METAL</td>
<td>21</td>
</tr>
<tr>
<td>HEPATITIS, PET SCAN</td>
<td>14</td>
</tr>
<tr>
<td>OTHER</td>
<td>7</td>
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</tbody>
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## Differential Diagnoses

<table>
<thead>
<tr>
<th>DIFFERENTIAL Dx</th>
<th>% OF PTS</th>
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<tbody>
<tr>
<td>DEMENTIA</td>
<td>50</td>
</tr>
<tr>
<td>ENCEPHALITIS, SYPHILIS</td>
<td>43</td>
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<tr>
<td>HSV</td>
<td>35</td>
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<tr>
<td>HIV</td>
<td>28</td>
</tr>
<tr>
<td>HEAVY METAL</td>
<td>28</td>
</tr>
<tr>
<td>STATUS EPILEPTICUS, PARANEOPLASTIC SYNDROME, AUTO IMMUNE</td>
<td>21</td>
</tr>
<tr>
<td>ACUTE BRAIN SYNDROME, HEPATITIS</td>
<td>14</td>
</tr>
<tr>
<td>HYDROCEPHALUS, CVA, MALIGNANCY, DRUG RELATED, FE DEF, CMV, CO2 NARCOSIS, MASTOIDITIS, WERNICKE’S SYNDROME, CEREBRAL ATAXIA</td>
<td>7</td>
</tr>
</tbody>
</table>
What symptoms developed as people became sicker?

- **GAIT**: 12%
- **INCONTINENCE**: 10%
- **RESTLESS/IMPULSIVE**: 9%
- **SPEECH**: 9%
- **SLEEP DISTURBANCE**: 9%
- **PERSONALITY CHANGES**: 8%
- **VISUAL DISTURBANCES**: 8%
- **DELIRIUM**: 8%
- **DYSPHAGIA**: 8%
- **COGNITIVE DECLINE**: 8%
- **SEIZURES**: 8%
- **STARTLE REFLEX**: 8%
- **CONSTIPATION**: 8%
- **URINARY RETENTION**: 8%

- **93%** gait disturbance
- **86%** incontinence
- **80%** abnormal speech, abnormal movements, sleep disturbance, personality changes
What pharmacological management did patients need?

- Opioids
- Benzodiazepines
- Antipsychotics
- Aperients
- Anticholinergics

There is little information in the medical literature to guide care.
What non pharmacological interventions were documented?
Were patients referred to the CJD support group network?

Only documented in 4 cases

Room for improvement!
There is a wide variation in symptoms experienced at presentation and throughout disease progression.

The trajectory of disease is often more rapid than we normally see in other diseases.

Documentation of interventions and their effectiveness will assist timely management.

Psychosocial support for the family is paramount.
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image courtesy of Dr Chris O'Donnell, Radiopaedia.org, rID: 16320