

CJD-spørgeskema

Navn: _____

CPR nr.: _____

Fødeland: _____

Permanent opholdsland ved sygdomsdebut: _____

Tidspunkt for sygdomsdebut: _____ (måned, år)

1. Diagnose

Hvilken sygdom drejer det sig om? (se appendix 2, side 7)

- 1. sporadisk Creutzfeldt-Jakob Disease (CJD)
- 2. familiær CJD
- 3. iatrogen CJD
- 4. ny variant CJD
- 5. Gerstmann-Sträussler-Scheinker (GSS)
- 6. Fatal familiær insomni (FFI)
- 7. ikke prionsygdom

Hvis det er CJD, hvordan er sygdommen da klassificeret i forhold til de diagnostiske kriterier for CJD (se appendix 2, side 7)?

- 1. sikker
- 2. sandsynlig
- 3. mulig

Hvis det ikke er en prionsygdom, hvilken diagnose har patienten da fået?

Er patienten i live d.d.? 0. nej, dødsdato: _____ (dag, måned, år)
 1. ja

Hvis nej, er der foretaget sektion? 0. nej
 1. ja, **bedes en kopi af sektionsbeskrivelsen vedlagt**

2. Klinisk præsentation*

Klinisk præsentation ved **sygdomsdebut** (se appendix 1, side 5, for definitioner):

- 0. Hurtigt progredierende demens
- 1. Heidenhain
- 2. Kun psykiatriske debutsymptomer
- 3. Langsomt progredierende demens
- 4. Kun cerebellare debutsymptomer
- 5. Kun extrapyramidale debutsymptomer
- 6. Slagtilfælde lign. debutsymptomer
- 7. Kun sensoriske debutsymptomer
- 8. Andet
- 9. Information mangler

Hvilke af de typiske symptomer for CJD var til stede **i forløbet** efter indtræden af CJD?

- 1. Hurtigt progredierende demens
- 2. Langsomt udviklet demens
- 3. Myoklonier
- 4. Visuelle problemer
- 5. Cerebellare problemer
- 6. Akinetisk mutisme
- 7. Andre pyramidale symptomer
- 8. Andre extrapyramidale symptomer
- 9. Psykiatriske symptomer
- 10. Vedholdende smerte (direkte smerte + dårlig følelse)
- 11. Andre, specificer:

***En beskrivelse af sygdomsforløbet, evt. i form af kopi af epikrise bedes vedlagt**

3. Laboratorieundersøgelser

Er der fundet **14-3-3 protein** i spinalvæske fra patienten?

- 0. nej, ikke påvist
 - 1. ja, påvist
 - 2. tvivlsomt resultat
 - 3. ufortolkeligt af tekniske årsager
 - 8. undersøgelsen ikke udført
-

Er der fundet **Neuronspecifik Enolase (NSE)** i spinalvæske fra patienten (referenceværdi <35)?

- 0. nej, NSE <35
- 1. ja, NSE \geq 35
- 2. tvivlsomt resultat
- 3. ufortolkeligt af tekniske årsager
- 8. undersøgelsen ikke udført

Er der fundet **tau protein** i spinalvæske fra patienten?

- 0. nej, ikke påvist
- 1. ja, påvist
- 2. tvivlsomt resultat
- 3. ufortolkeligt af tekniske årsager
- 8. undersøgelsen ikke udført

Er der fundet **p-tau protein** i spinalvæske fra patienten?

- 0. nej, ikke påvist
- 1. ja, påvist
- 2. tvivlsomt resultat
- 3. ufortolkeligt af tekniske årsager
- 8. undersøgelsen ikke udført

Er der fundet sygdomsspecifik **PRNP-mutation**?

- 0. nej, ingen mutationer
- 1. ja, påvist
- 8. resultat afventes
- 9. undersøgelse ikke udført

Hvis der er fundet **PRNP polymorphisme i codon 129**, giv en beskrivelse:

- 0. Met₁₂₉/Met₁₂₉ (Metionin, homozygot)
- 1. Met₁₂₉/Val₁₂₉ (Metionin, Valin)
- 2. Val₁₂₉/Val₁₂₉ (Valin, homozygot)
- 8. resultat afventes
- 9. undersøgelsen ikke udført

Hvis der er fundet **PRNP polymorphisme i en anden codon**, giv en beskrivelse:

4. EEG

Har patienten et EEG*, der støtter diagnosen CJD (se appendix 3, side 9)?

- 0. nej
- 1. ja
- 8. EEG ikke udført

***Kopi af EEG beskrivelse bedes vedlagt**

5. MR-scanning

Fund ved MR-scanning* (se appendix 4, side 10):

- 0. normal MR
- 1. abnormal MR
- 9. undersøgelsen ikke udført

*** Kopi af MR beskrivelse bedes vedlagt**

De følgende 4 spørgsmål skal kun besvares, hvis der er udført MR-scanning!

Viser MR-scanning hyperintense signaler i caudatus og putamen?

- 0. nej
- 1. ja

Viser MR-scanning hyperintense signaler i posterior thalamus, større end andre områder?

- 0. nej
- 1. ja

Viser MR-scanning atrofi?

- 0. nej
- 1. ja

Viser MR-scanning andre uspecifikke abnormaliteter? (f.eks. iskæmiske, aldersrelaterede forandringer)

- 0. nej
- 1. ja

Dette er slut på spørgeskemaet. Tak for hjælpen.

Dato: _____

Navn: _____

Hospital, afdeling: _____

Telefonnummer: _____

E-mail: _____

Skemaet bedes sendt til:

Epidemiologisk afd., Statens Serum Institut, Ørestads Boulevard 5, 2300 København S

Appendix 1:

Definitions of clinical presentations at first appearance of disease

This is an attempt to classify the different modes of presentation.

It may not be possible to accurately classify a particular case. If the case has good data but does not clearly fit one of the specified categories, then the code 8 "other" value should be used. If there are insufficient data to categorise the case, then code 9 "missing" should be used.

Code 0: Rapidly Progressive Dementia

The majority of cases will probably be in this category. Precise presenting symptoms will vary from case to case. The picture is one of an encephalopathic illness with dementia and diverse other neurological features, progressing rapidly over weeks to a few months, with no individual cognitive or physical deficit being present alone for more than two weeks.

Code 1: Heidenhain's

These cases present with impairment of visual acuity and/or field, progressing onto cortical blindness, without other significant clinical deficit for the first two weeks of illness.

Visual symptoms might include visual loss, visual inattention, visual illusions and visual hallucinations. It is essential that the symptoms progress to cortical blindness. Cases with other onsets that progress to include cortical blindness are NOT included in this category.

Code 2: Pure Psychiatric Onset

These cases present with psychiatric symptoms, such as depression, anxiety, paranoia, and delusions, without the presence of other features for a period of at least four weeks.

Non-specific malaise or apathy do not count unless accompanied by some of the above symptoms. Visual or auditory hallucinations alone do not count, but may accompany the above features.

It may be difficult to distinguish between the early features of dementia and a more specifically psychiatric onset. Behavioural change straightforwardly due to a developing dementia is not included in this category. The essential characteristic of this presentation is that the patients present with a disturbance that suggests a psychiatric disturbance rather than an obvious dementia and specifically neurological features are absent.

Code 3: Slowly Progressive Dementia

These cases present with a slowly progressive dementia, developing over months to years, without any other significant neurological features for the first six months. It is important to distinguish this from Rapidly Progressive Dementia (as defined in Code 0, above).

Code 4: Pure Cerebellar Onset

Presentation is with a progressive cerebellar syndrome without other significant features, for at least two weeks.

Code 5: Extrapyrarnidal Onset

Presentation is with an extrapyramidal syndrome involving Parkinsonian features with or without chorea, athetosis or dystonia, but without other significant features for at least two weeks.

Code 6: Stroke-Like Onset

Presentation is abrupt enough for a diagnosis of stroke to be entertained in the initial stages.

Code 7: Sensory Symptoms at Onset

Presentation with somato-sensory symptoms alone for at least two weeks. Such symptoms might include parasthesia, dysaesthesia, numbness, specifically neuro-genic pain etc, but would not include vague, non-specific aches and pains. This category does not include presentation with special sensory symptoms (i.e. visual, auditory, olfactory, gustatory).

Sensory symptoms may be present along with other symptoms (for example, as part of a Rapidly Progressive Dementia), but this category is for essentially 'pure' sensory presentation.

Code 8: Other

None of the presentations described above is applicable.

Code 9: Missing

There is no clear clinical information available or the information does not allow a definite classification according to the above criteria.

Appendix 2:

Clinical diagnostic criteria

Sporadic CJD (Rotterdam 1998)

Criteria:

- I. Rapidly progressive dementia
- II.
 - A Myoclonus
 - B Visual or cerebellar problems
 - C Pyramidal or extrapyramidal features
 - D Akinetic mutism
- III. Typical EEG

Classification:

Definite Sporadic CJD : (Sikker) Requires neuropathological/immunocytochemical confirmation

Probable Sporadic CJD: (Sandsynlig) I + two of II + III **OR**
possible sporadic CJD + positive 14-3-3

Possible Sporadic CJD : (Mulig) I + two of II + duration < 2 years

Iatrogenic CJD: Progressive cerebellar syndrome in a pituitary hormone recipient **OR** sporadic CJD with a recognised exposure risk, *e.g. dura mater transplant.*

Familial CJD : Definite or probable CJD plus definite or probable CJD in a first-degree relative **OR** neuropsychiatric disorder plus disease-specific PRNP mutation.

Variant CJD (UK, 2000)

Criteria:

- I. **A** Progressive neuropsychiatric disorder
- B** Duration of illness >6 months
- C** Routine investigations do not suggest an alternative diagnosis
- D** No history of potential iatrogenic exposure

- II. **A** Early psychiatric symptoms*
- B** Persistent painful sensory symptoms**
- C** Ataxia
- D** Myoclonus or chorea or dystonia
- E** Dementia

- III. **A** EEG does not show the typical appearance of classical CJD***
 (please enclose EEG recording for review by national expert committee)
- OR** no EEG performed
- B** Posterior thalamic high signal on MRI scan (please enclose MRI scan for review by the national expert committee)

- IV. **A** Positive tonsil biopsy

Classification:

Definite Variant CJD : IA + neuropathological confirmation of vCJD****

Probable Variant CJD: I + 4/5 of II + IIIA + IIIB **OR**
 I + IV A

Possible Variant CJD : I + 4/5 of II + IIIA

* depression, anxiety, apathy, withdrawal, delusions.

** including both frank pain and/or unpleasant dysaesthesia.

*** generalised triphasic periodic complexes at approximately one per second.

**** spongiform change and extensive PrP deposition with florid plaques, throughout the cerebrum and cerebellum.

Appendix 3:

EEG Criteria

WHO consultation on global surveillance, 1998

1. Strictly periodic activity:
 - Variability of intercomplex intervals <500 ms
 - Continuous for at least one 10 sec period
2. Bi- or Tri- phasic morphology of periodic complexes
3. Duration of majority of complexes 100-600ms
4. Periodic complexes may be generalized or lateralized but not regional or asynchronous

Technical:

1. Bipolar montages including the vertex should be used
2. Referential montages including vertex and CZ reference electrodes should be used
3. The ECG should be coregistered
4. External alerting stimuli should be used
5. The whole record should be viewed whenever possible and a 5 minute continuous sequence as a minimum

Appendix 4:

MRI in Sporadic CJD

High signal seen in the putamen and caudate nucleus on FLAIR, T2-weighted and Proton Density-weighted images.

Focal cortical high signal may also be a feature.

Diffusion-weighted images may be helpful.

FINKENSTAEDT M et al "MR Imaging of Creutzfeldt-Jakob Disease"
Radiology 1996; 199:793/798.

MRI in Variant CJD

High signal seen in the posterior thalamus (pulvinar region), dorsomedial thalamus and tectal plate on FLAIR, T2-weighted and Proton Density-weighted images.

Pulvinar sign defined as "Bilateral hyperintensity of the pulvinar nuclei of the thalamus relative to the signal intensity of the anterior putamen".

Diffusion-weighted images may be helpful.

COLLIE D et al MRI of Creutzfeldt-Jakob disease: imaging features and recommended MRI protocol. Clinical Radiology 2001; 56:726-739.