Diagnostic et transmission des maladies à prions

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Quintessence

 Les encéphalopathies spongiformes transmissibles constituent un groupe hétérogème de maladies ayant pour corrélat pathologique commun les prisess.

 En Suisse, la maladie de Creutzfeldt-Jakob sporadique est la forme la plus fréquente; la nouvelle variante n'a encore jamais été observée.

 Les prions sont transmissibles, principalement par les produits sanguins, les transplants tissulaires et le matériel chirurgical.

Le diagnostic de suspicion d'une maladie de Creutzfeldt-Jakob est posé, chox un sujet vivant, à partir du tableau clinique typique d'une part et de l'IEM cérébrale, de l'analyse du liquide céphalo-rachidien et de l'EHG d'autre part.

La déclaration obligatoire des cas cliniques suspects permet de prendre des mesures pour protéger la population et de vérifier leur efficacité. Les principales mesures sont le contrôle et le rappel des produits sanguins ainsi que l'évaluation des techniques de stérification.

Contexte

Les maladies à prions ou encéphalopathies spengiformes transmissibles (HST) sont des maladies provosquées transmissibles (HST) sont des maladies provosquées par des prions (particules problèques infectionses), non curuliles et toujours fatales. Chez l'homme, elles se présentent sous trois formes: familiale, sporadique et acquise.

Les formes familiales sont la maladie de Creutzfeldt-Jakob familiale (DMCJ), l'insommie fatale familiale (DF) et le syndrome de Gerstmann-Sträussler-Scheinker (GSS).

La forme la plus fréquente est la forme sporadique de la maladie de Creutzfeldt-Jakob (sMCJ).

La nouvelle variante (vMCI) et la forme introgène de-MCI (iMCI) comptent parmi les maladies à prions acquises. La transmission des prions se fait de l'hornme ou de l'animal à l'homme dans la (MCI, et de bœufs atteints d'encéphalopathie spengiforme bovine (ESB) à l'homme, par voie intestinale, dans la vMCI. En raison de la transmissifiité, la déclaration des cas eliniques suspects est obligatoire, afin que des mesures de protection de la population puissent être prisés. Le diagmostic précoce est donc important pour la personne touchée et ses proches, mais il présente auxei un intérét général.



Sirmon Jung

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Evolution clinique

MCJ sporadique, introgène et familiale

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Accec 1 à 2 cas par an pour 1 million d'habitants, la sMCJ est la forme la plus fréquente. Elle survient principalement chez les personnes d'un certain âge 065 ans en moyennel et se distingue des autres démences par une évolution rapide télurée: 4,5 à 7 moist. Même si testes les zones cérébrales peurent finalement être sui chies et que par conséquent les symptômes possibles sont très variés, ceux-ci sont plus ou moins fréquents et dépendent du stade de la maladle (voir tab. 1 © 1. Les différences génotypiques sur le codon 129 de la protéine prionique entrainsent une variabilité supplémentaire. La iMCJ thormis les cas de vMCJ et la fMGJ ne se distinguent pas de la sMCJ pour co qui est de la clinique et de l'évolution.

Variante de la MCJ

A la différence de la sMCJ, la vMCJ est une forme nure, qui se manifeste surriout client les jeunes adultes (28 ars en moyenne). L'évolution, un peu plus longue (14 mois), commence typiquement par des symptômes psychiatriques tôlépression, angoisses, apathie, délire, etc.) ou des troubles de la semulatiné (généralement seas forme de dyscathésies douloureuses des extrémités). Le tableau se rapproche de celui de la sMCJ au bout de quelques muis.

Examens complémentaires

Electroencéphalogramme (EEG)

L'EEG confirme le diagnostic de sMCJ quand il mentre des complexes d'ondes périodiques hiphaxiques ou triphasiques [1], bien que cette preuve ne soit pas spécifique [2]. Dans la vMCJ, l'EEG ne présente le plus souvent que des modifications générales non spécifiques.

Liquide céphalo-rachidien

Les paramètres de routine (nombre de cellules, protéines, glucose) sont habituellement normaux. L'élévation des protéines 14-3-3, tau et S100b ainsi que de l'écolase neurone-spécifique est caractéristique de la sMCJ [3, 4], mais elle n'est pas non plus spécifique; elle reflète avant tout la lyse neuronale. Dans un travail actuel sur la valeur des analyses du LCR dans la sMCJ, la sensibilité était de 86% pour la protéine 14-3-3, de 81%, pour la protéine tau et de 65% pour la protéine S100b [4]. En taut que marqueur plus singulier, la protéine 14-3-3 présente donc la sensibilité la plus élevée, sensibili-

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