

## A rare epileptic syndrome of ring chromosome 20 with non-convulsive status epilepticus - First two cases reported from Thailand

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**Objective:** To study the electro-clinical features of non-convulsive status epilepticus (NCSE) and positive ring chromosome 20 in Thai patients.

**Methods:** Two Thai females, age 25 and 37, who suffered from frequent complex partial status epilepticus were studied. Both have also had generalized tonic clonic and complex partial seizure of varying frequencies since adolescence. Their intellectual functions were normal. 24-hour video/EEG telemetry was recorded during the NCSE. Intravenous antiepileptic drugs (AED) were infused and clinical response observed. The patients were followed up for 22 and 26 months and responses to oral AED were determined. Standard chromosome study using Geimsa - banding technique was done.

**Results:** During the NCSE, the patients' consciousness fluctuated between overt unresponsiveness and normal awareness. Mild tonic seizures and eyelid myoclonia were sometimes noted in case I. The onset and ending of each episode were ill-defined except by bedside tests. The duration varied between 20 seconds to 5 minutes. The ictal EEG consisted of long-lasting generalized rhythmic 3-5 Hz slow or sharp waves with rare spikes, seldom interrupted by 8 Hz background. Despite the continuous discharges, the patients had subtle clinical seizures that were easily aroused by verbal stimuli. The NCSE lasted for several days. Intravenous phenytoin and valproic acid could temporarily normalize the ictal EEG. However, there was no temporal correlation between the infused AED and consciousness or rhythmic EEG episodes. Chromosome study showed ring chromosome 20 in 20% of female karyotype in both patients [46,XX,r(20)(p13q13)/46,XX] but were negative in 4 healthy siblings. No lesion could contribute to NCSE in their MRI. Oral AED decreased more than 75% of the overt complex partial seizure episodes in both cases during follow-up period. However, they had no effects on the natural history of NCSE. The patients' daily activities were minimally affected by the ongoing discharges detected on periodic EEG recordings.

**Discussion and Conclusion:** Ring chromosome 20 with NCSE is a rare but distinctive epileptic syndrome.<sup>1</sup> High percentages of cells with r(20) were found to be associated with young age of epilepsy onset or early death, dysmorphism, mental retardation, behavioral disorder, and intellectual dysfunction. MRI were normal in most studies, suggesting unknown structural substrate. Recent findings of [18F]fluoro-L-Dopa PET has shown bilaterally decreased striatal dopamine uptake.<sup>2</sup> This was postulated to be responsible for the NCSE by impairing the mechanism to interrupt seizures.

This is the first case report of ring chromosome 20 with NCSE in Thailand. This rare epileptic syndrome should be added to the list of known "genetic" epilepsies in this region. Our cases have similar electro-clinical features as other reports but have a more benign and less pharmacologically resistant course, probably from the low rate of r(20) mosaicism. Assessing the severity of this disorder based on clinical seizures as well as EEG is crucial.

### References

1. Inoue Y, Fujiwara T, Matsuda K, *et al.* Ring chromosome 20 and nonconvulsive status epilepticus, a new epileptic syndrome. *Brain* 1997; 120: 939-53.
2. Biraben A, Semah F, Ribeiro M-J, *et al.* PET evidence for a role of the basal ganglia in patients with ring chromosome 20 epilepsy. *Neurology* 2004; 63:73-7.