# The clinical and genetic features of generalized epilepsy with febrile seizure plus (GEFS+) in Chinese families

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*Objective:* To study the features of phenotypic variation and clinical genetics in Chinese families with generalized epilepsy with febrile seizure plus (GEFS+).[1,2]

*Methods:* The study subjects were from the 2,296 patients in the outpatient clinic of the Epilepsy Center, Second Affiliated Hospital, Guangzhou Medical College. There were altogether 60 GEFS+ families, based on the following diagnostic criteria: Febrile seizures (FS) from 6 months to 6 years, which persisted beyond 6 years of age, or associated with afebrile seizure, excluding symptomatic epilepsy. The clinical histories and physical examination, EEG, brain CT or MRI were performed on the probands. Clinical information on relatives were obtained by face-to-face or telephone interview.

*Results:* Clinical information of 1,730 members of the 60 families was obtained. Among them were 180 patients with seizures (112 males and 68 females). The mean age of seizure onset was 3.4±1.0 years. The frequency of seizures ranged from once a lifetime to 3 times a day. Most patients had normal neurological development. Mental retardation was seen in 6 patients. Most patients had benign prognosis and 7 patients had refractory epilepsy. There was obvious difference in phenotypes, including 52 patients with FS, 39 with FS+ partial seizure, 22 with FS+ generalized seizure, 2 each with hemiconvulsion-hemiplegia syndrome (HHE) and benign children epilepsy with centro-temporal spikes (BECCT); one each with severe myoclonic epilepsy of infancy (SME), Lennox-Gastaut syndrome, FS+ atonic seizure, myoclonic-astatic seizure and myoclonic absence epilepsy (MAE); and 56 who were not classified. Epileptiform activity was found in 34 out of 57 patients who had EEG examination. In some probands with epileptic syndromes, the relevant discharges were found in their EEG. Sixteen patients had brain CT examination and all were normal. Twenty-six patients had MRI examination, out of which 14 were normal and 12 abnormal. Abnormalities included atrophy in temporal lobe or hippocampus sclerosis (5), minor cerebral white matter density changes (3), cerebellar atrophy (2), callosal agenesis (1) and cerebral ventricle enlargement (1). Genetic analysis was consistent with autosomal dominant inheritance with incomplete penetrance. Bilineal inheritance was found in 7 families. Thus, polygenic inheritance was not excluded.

*Conclusions:* GEFS+ was a common epilepsy syndrome in Chinese. It includes many phenotypes. Inheritance mode may be autosomal dominant or polygenic inheritance.

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## References

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