Review article

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Genetic literacy series: genetic epilepsy with febrile seizures plus

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Epileptic Disorders

Genetic Literacy Series:
Genetic Epilepsy with Febrile Seizures Plus (GEFS+)

- Genetic epilepsy with febrile seizures plus (GEFS+) is a familial epilepsy syndrome in which affected individuals within a family typically have a variety of epilepsy phenotypes.

- The most common GEFS+ phenotype is febrile seizures (FS) followed by febrile seizures plus (FS+).

- FS+ is defined as either or both of the following:
  A. Febrile tonic-clonic seizures that begin before 3 months and/or continue after 6 years
  B. Occurrence of both febrile and afebrile generalized tonic-clonic seizures.

- The GEFS+ spectrum comprises a heterogeneous group of generalized and focal epilepsy phenotypes including the developmental and epileptic encephalopathies of epilepsy with myoclonic-atonic seizures and Dravet syndrome.

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- Mutations in *SCN1A* are the most common identified genetic cause of GEFS+, accounting for approximately 19% of families.

- Whether clinical molecular genetic testing in patients with FS+ or other GEFS+ phenotypes is indicated is currently under debate.
  - If genetic testing is offered, *SCN1A* sequencing should be included, and this may be most efficiently performed as part of a panel or whole-exome sequencing.

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