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Phenotypic spectrum and long-term outcome of children with genetic early-infantile-onset developmental and epileptic encephalopathy

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- In our study, We described the clinical features and long-term outcome of genetic causes of early infantile onset DEEs.
- The clinical manifestations of early infantile onset DEEs are variable, including dyskinesia. Early infantile onset DEE-BS usually responds poorly to ASMs therapy.
- We found that the ion channel gene mutations are the most common mutated genes.

- In WS, we detected *SCN3A*, *SCN2A*, *SCN8A*, *CACNA1H*, *DEPDC5*, *MECP2*, *DYNC1H1*, *CDKL5*, *ALG11*, *CCDC88C*, *GABAA1*, *IL1RAPL1*, *RNASEH2B*, *SLC19A3*, *STXBP1*, *RARS2*, *COL4A2* mutations. In addition to common gene mutations, we reported rare possible pathogenic genes: *CCDC88C*, *IL1RAPL1*, *RNASEH2B* and *COL4A2* in WS.
- We detected rare possible pathogenic genes: *SETBP1*, *DPYD*, *CSNK2B* and *H3F3A* in non-syndromic genetic causes of early infantile onset DEEs.
- Although genetic causes of early infantile onset DEEs responded poorly to ASMs treatment, we found that some genetic mutations related early infantile onset DEEs received good effects on specific ASMs.