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## Clinical and Genetic Evaluation of 23 Children with Infantile-onset Epileptic Encephalopathy

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***Background:*** Infantile epileptic encephalopathies (IEE) are a group of conditions in which cognitive, sensory, and/or motor functions deteriorate as a consequence of epileptic activities, which consist of frequent seizures and/or major interictal paroxysmal activity. There are various causes of IEE and they may occur at any age.
***Methods:*** We reviewed patients in the Department of Paediatrics and Adolescent Medicine of the University of Hong Kong, Queen Mary Hospital and Duchess of Kent Children's Hospital with the clinical diagnosis of IEE of unknown aetiology over a 10-year period (2003-2012). Five genes (*ARX, CDKL5, KCNQ2, SCN1A,* and *STXBP1*) were screened using sequencing.
***Results:*** A total of 23 patients were identified and their electroclinical features were studied. Of the 23 patients, 10 (43.5%) had epileptic spasm as the presenting seizure type. Throughout the clinical course, patients were characterised by frequent seizures that were multiform and pharmaco-resistant. The commonest subsequent seizure type was generalised tonic/clonic/tonic-clonic seizure (17 out of 23, 73.9%). All of the patients had developmental delay of various degrees. Movement disorder in terms of dystonia was the most common associated clinical feature (10 out of 23, 43.5%). Five genes (*ARX, CDKL5, KCNQ2, SCN1A,* and *STXBP1*) were screened in 20 of our patients. We identified three patients with *STXBP1* mutations, two patients with *SCN1A* mutations, and one patient with *KCNQ2* mutation. The overall detection rate was 30% (6/20). Two out of three patients with Dravet phenotype were screened positive for *SCN1A* mutation. The only patient with typical Ohtahara phenotype was screened positive for *STXBP1* mutation.
***Conclusion:*** This study highlighted the clinical characteristics of IEE and studied the yield of mutational screening of five selected genes in this group of patients. Dravet syndrome and Ohtahara syndrome have characteristic phenotypes. *SCN1A* and *STXBP1* mutational analysis should be performed in children with classic presentations of the above-named conditions respectively.