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Neonatal seizures associated with different pathologic KCNQ2 mutations

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Statement of the Problem:
KCNQ2 mutations have been found in patients with familial benign neonatal seizures, myokymia or early onset epileptic encephalopathy. The diagnosis of neonatal seizures could be difficult and challenging in some cases. KCNQ2 encodes the potassium channel subunit Kv7.2 and its mutations have been shown to cause benign familial neonatal seizures (BFNS) with a favourable prognosis but are also associated with neonatal onset seizures and poor outcomes, such as early onset epileptic encephalopathy (EOEE).

Patients: We present a summary of our experience with neonatal seizures associated with KCNQ2 mutations. In all cases a full diagnostic evaluation including MRI was performed before the DNA analysis. The first patient has a positive family history, early onset tonic neonatal seizures, clinical signs of encephalopathy. The following mutation was identified: c638G>A,p. Arg213Gin, exon 4, KCNQ2, seizure free on carbamazepine treatment The second patient presented with early tonic seizures, tonic eye deviations, burst-suppression EEG pattern, MRI –hypoplasia of corpus callosum, clinical evidence of severe encephalopathy, KCNQ2 mutation associated with EIEE was identified: G18416>A,pGly281Arg, exon 6. The third clinical case is a term baby presented with early seizures –generalized tonic and facial myoclonic, KCNQ2 mutation c.637C>TpArg213T, exon 4, successful treatment with carbamazepine, but severe developmental delay. Our fourth patient was with a positive family history, early tonic and clonic seizures, no evidence of encephalopathy, KCNQ2 mutation – partial deletion of exon 17, known as a cause of benign familial neonatal seizures. This infant is seizure free and the neurodevelopment is completely normal.

Conclusion:
KCNQ2 mutations could be associated with variable clinical presentation, the genetic testing should be performed in all neonates with unknown seizure cause after complete diagnostic evaluation.

Biography:
Ralitza Gueorgieva has her expertise as neonatologist in NICU, University Pediatric Hospital in Sofia. The main scientific interests are focused on the fields of neonatal intensive care and neonatal neurology. The team of the department has a great experience managing neonates born with perinatal asphyxia and hypoxic-ischemic encephalopathy, premature infants with different perinatal brain lesions. Another important issue in our clinical and scientific work is the treatment and follow up of infants with neonatal seizures.

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