

01 // WHAT'S IN A NAME

KCNQ2 mutations are referred to by all different names; KCNQ2, KCNQ2 Epilepsy, and KCNQ2 Epileptic Encephalopathy. But as experts understand more about this severe form of KCNQ2 mutation, that causes significant developmental impairment in addition to epilepsy, it is now classified as KCNQ2 Developmental and Epileptic Encephalopathy.

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02 // WHAT IS KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY?

KCNQ2 Early Onset Developmental and Epileptic Encephalopathy is a genetic disorder that causes seizures within the first days of life. Seizures most often have prominent tonic-clonic (grand mal) characteristics.

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03 // HOW MANY CASES OF KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY ARE KNOWN?

It is estimated that there are over 700 known cases worldwide and the numbers are growing rapidly. However, KCNQ2 mutations are estimated to occur in once every 26,000 live births, and are not diagnosed due to lack of awareness. At this rate, there 5000 children globally born each year with KCNQ2 epilepsy, meaning there are tens of thousands yet to be diagnosed.

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04 // IS THERE A CURE FOR KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY?

There is no cure for KCNQ2 Developmental and Epileptic Encephalopathy. However, the potassium channel, which is impacted by this mutation, appears to be a good target for new drugs and has the interest of several emerging pharmaceutical companies.

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05 // RESEARCH AND KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY

Research into KCNQ2 may lead to better understanding of other diseases, like autism and epilepsy. Recent studies have shown that KCNQ2 is one of the genes that can cause autism.

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06 // KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY DOES NOT DISCRIMINATE

KCNQ2 Developmental and Epileptic Encephalopathy affects both genders and all races and ethnic groups at an equal rate. Reported cases are from over 33 different countries.

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07 // PEOPLE WITH KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY HAVE A WIDE RANGE OF OUTCOMES: EACH IS UNIQUE

KCNQ2 Developmental and Epileptic Encephalopathy affects everyone differently; some have relatively mild symptoms while others are severely impacted. All people with KCNQ2 Developmental and Epileptic Encephalopathy have some intellectual disabilities including developmental delay, autism or aggression. Most have physical disabilities as well, which can include low muscle tone, inability to walk, eat or breathe without assistance. In some seizures resolve early in life, in others they may continue indefinitely or reappear sporadically. Even the identical variant the mutation in the KCNQ2 gene can lead to very different outcomes, an area that could benefit from further study.

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08 // GENE SEQUENCING HAS IDENTIFIED KCNQ2 MUTATIONS AS THE CAUSE OF SEVERAL BETTER KNOWN CONDITIONS

Gene sequencing (whole genome, whole exome, or seizure gene panels) has identified KCNQ2 mutations in patients previously diagnosed with Ohtahara Syndrome, autism, cerebral palsy (CP), and other symptomatic disorders.

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09 // THE LESS SEVERE FORM OF THE KCNQ2 MUTATION HAS BEEN KNOWN FOR OVER 20 YEARS

KCNQ2 mutations have been understood to cause Benign Familial Neonatal Epilepsy (BFNE), an inherited mutation in the KCNQ2 gene, for more than 20 years. However, KCNQ2 BFNE typically results in seizures in early infancy that dissipate without lasting effect on cognitive function. Understanding which specific KCNQ2 mutations result in the more severe form is an important area of research.

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10 // HOW WAS KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY FIRST IDENTIFIED?

KCNQ2 Developmental and Epileptic Encephalopathy, the more severe form of the KCNQ2 mutation, was first recognized by a research team in 2012 Belgium, led by Doctors Peter De Jonghe and Sarah Weckhuysen (with support from a global research consortium). They noted that a subset of patients with a KCNQ2 mutation had more severe symptoms, and did not inherit the mutation from a parent (a de novo mutation). With growing awareness, and expanding patient population, and research funding, the number of experts studying KCNQ2 developmental and epileptic encephalopathy has grown since that time.

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11 // EARLY LIFE SEIZURES ARE THE HALLMARK OF KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY

KCNQ2 mutations result in dramatic neonatal symptoms, with multiple seizures daily. KCNQ2 mutations are the most common genetic cause of epilepsy in the first week of life.

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12 // MRI FINDINGS APPEAR NORMAL DESPITE SIGNIFICANT COGNITIVE IMPACT

MRI findings appear normal despite longer-term cognitive disabilities in people with KCNQ2 developmental and epileptic encephalopathy. As a result, many hospitals do not initially recognize the symptoms. However, KCNQ2 mutations usually result in an EEG with burst-suppression pattern or multifocal epileptic activity in the period of early infancy.

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13 // AWARENESS IS KEY TO IDENTIFYING ALL AFFECTED AND IMPROVING TREATMENT OPTIONS

The number of people with KCNQ2 Developmental and Epileptic Encephalopathy who remain undiagnosed is significantly higher than the number of those currently diagnosed. The disease is very rare, relatively new, and the symptoms are variable and may appear similar to other diseases. Awareness and identification are key to helping as many patients as possible and developing better treatment options.

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14 // THERE IS NOTHING APPROVED FOR THE TREATMENT OF KCNQ2 DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY OR ITS SYMPTOMS

There are no drugs of any kind approved by the FDA or other international agencies for the treatment of KCNQ2 developmental and epileptic encephalopathy or its symptoms. Patients' seizures are typically treated with combinations of drugs approved for other types of epilepsy, with mixed results. There are no treatments for the cognitive or physical impairment which are the most significant issues for most people with KCNQ2 developmental and epileptic encephalopathy.

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