Patient journey KCNQ2-encephalopathy

EpiCARE

1 ….first symptoms…. 2 ….diagnosis…. 3 ….treatment….. 4 …follow up… 5 ….transition…

1. FIRST SYMPTOMS

Timeline: from the 1st day of life up to 1 year

Clinical signs / Symptoms
- In most cases neonatal seizures occur in the first days of life.
- In some cases, the seizures present after 1 month and within the first year of life.
- Epilepsy may be absent; in few cases, despite no signs of seizures, an abnormal eeg accompanies delays in development and or behavioral development.

Identify patient needs
- Parents need to be correctly informed about how different forms of seizures need to be managed.
- Parents need to have basic information on epilepsy and epileptic seizures
- Parents need to be informed their child could have a severe cognitive disability
- Parents need to know how to access early rehabilitation

Ideal results/ Support
- Parents concerns are taken seriously and are given reassurance.
- Families are given instructions on how to manage an epileptic attack should it recur; description of emergency medication and when to go to hospital
- Rehabilitation plan

2. DIAGNOSIS

Timeline: from 2 months up to 6 years (normally at 2 years)

Clinical signs / Symptoms
- Developmental progress differs from child to child, this disease effects the entire lifespan of the person
- Even in the first few months overall development is delayed; people affected by a KCNQ2 encephalopathy rarely become independent
- The child has several different types of epileptic seizures: focal or generalized seizures, tonic-clonic, myoclonic, spasms, with or without a trigger factor
- Sometimes seizures are related to fever, sometimes they last longer, above all in the first years of life status epilepticus may recur. In most cases the seizures disappear within the first 3-5 years of life; however, severe/medium/light psychomotor retardation can remain
- Comorbidities can be present such as language impairment, motor disturbances, behavioral disorders, orthopedic issues, visual problems, digestive difficulties, difficulty swallowing and autism.

Identify patient needs
- Parents need an adequate explanation of the diagnosis and relative prognosis with psychological support
- Parents need to be offered genetic testing
- Parents should understand that development is different from child to child
- Parents have to be sure of the diagnosis ("benign" form of KCNQ2-related epilepsies *versus* encephalopathy)
- Parents need to have indications of how to face up to/treat epileptic seizures and what other non-pharmacological therapies their child might need
- Parents should be informed if in their countries there are clinical studies on KCNG2 and if their child can participate.
- They must know how to keep their child safe (detect seizures at night, fever management...).
- Parents need to know what social assistance is available from the government.

Ideal results/ Support
- Genetic consultancy, explanation of causes and the possibility of reappearance.
- Professional support is given to face up to the diagnosis and the family is directed to the parent's support group and/or the association.
- Parents receive clear instructions, emergency protocol, explanation of risks and how to minimize them.
- Parents receive clear information about possible clinical studies in which their child can participate with in-depth information on risk and benefits.
- It is important for the family to know how important education and rehabilitation are for the development of the child and should be closely monitored
- The family is given a document summarizing the social benefits available and offered for the type of disease according to residency.

3. TREATMENT

Timeline: lifelong

Clinical signs / Symptoms
- Epileptic encephalopathy does not respond to medication, particularly during the first years of life. In children, seizures resolve at around 3-5 years of age, in other cases seizures persist. Treatments should aim at controlling seizure activity, mainly during infancy, and reducing side effects.
- Considerable attention should be paid to triggering factors and seizures prevention.

Identify patient needs
- Parents need counselling and professionals' help.
- Parents need information on prescribed medications, side effects, on side-effect monitoring in the long term
- Parents also need advice on how to handle triggering factors, on the condition, on the multiple issues of severe motor and cognitive impairment.
- Parents need medical help /advice on non-epileptic symptoms.
- Advice on pre-school/school/aid.
- Access to clinical trials for new treatment opportunities.

Ideal results/ Support
- A regular consultancy with health care professionals is offered.
- Up-to-date information is available for parents any time.
- Parents are informed on medication side effects and on follow-up blood test timing.
- Non-epileptic symptoms are effectively treated.
- Parents are offered support to find a school / daily assistance and care.
- Healthcare centers involve patients in research activity.
- Over the years physiatrists help families to choose devices, whose timely delivery is guaranteed by involved orthopedic units.

4. FOLLOW UP

Timeline: 2-16 years

Clinical signs/Symptoms
- Other problems like intellectual and motor disability, behavioral, orthopedic and intestinal issues may occur

Identify patient needs
- Parents need advice and evidence-based information on additional symptoms
- Parents need emotional support
- Evidence-based therapies (psychomotricity, speech therapy, postural re-education, behavioral therapy)

Ideal results/ Support
- Monitoring above mentioned issues and if possible, offering any treatment;
- Developing standards for the quality of adults' life.
- Availability of home and/or institutional care at the highest level.
- Defining a rehabilitation program (psychomotricity, speech therapy, postural re-education, behavioral therapy)

5. TRANSITION

Timeline: 16 years and up

Clinical signs / Symptoms
- Transition to adulthood
- Usually, seizures do not occur any longer. However problems relating to cognitive and motor development increase.
- Generally, patients with severe cognitive outcome show autistic behavior or are diagnosed with autism.
- Comorbidity features increase.
- In several healthcare centers the lack of cooperation between pediatricians and services oriented to adults may result in poor support for patients and their families.

Identify patient needs
- Parents need counselling and support in the transition to adulthood
- Treating adult patients, considering the severe motor impairment affecting young patients
- Monitoring development, new treatment opportunities, behavior and neuropsychological situation.

Ideal results / Support
- A transition process from pediatricians to GPs should be introduced
- Setting up of a rehabilitation plan for maintenance
- Occupational therapy / day-care centers / residential centers
- Dealing with the increase of different problems such as motor disability, swallowing issues, behavior, social and cognitive impairment.
- Community-based group homes for young adults, in view of a long-term adulthood program "during us" and perspective "after us" stages

European Reference Network

Co-funded by the European Union