Alternating Hemiplegia of Childhood - AHC Patient Journey

European Reference Network

EpiCARE

IAHCRC International AHC Research Consortium

AHC can present with any and every neurological symptom. It is like living with many different neurological diseases in one.

First symptom ... ... treatment... ... follow up (childhood & adolescence)...

1. First Symptom (within 18 months)

First symptom is always neurological but non AHC specific. It can be any one or a combination of any of the following paroxysmal manifestations: epileptic seizures, dystonic (painful muscle rigidity) / tonic episodes, episodes of altered awareness, abnormal eye movements, episodes of autonomic dysfunction or other neurological symptoms.
The hallmark of AHC are recurrent episodes of hemiplegia (floppy limbs) alternating in either side of the body, occasionally spreading to the whole body. They usually, but not always, appear later in the course of the disease, in addition to the previous symptoms.
These episodes are usually interrupted by sleep.

Need: **Family** needs information and reassurance during the long and complex diagnostic process. ***Active involved in also of importance. Family reports about the episodes occurring at home should be taken into valuable consideration***.

Ideally: Correct diagnosis as early as possible, preventing the start of ineffective and potentially dangerous treatments. Increased awareness and ability of the family to accept the diagnosis of AHC, its rarity and complexity, and to face the burden of the many neurological co-morbidities through adequate information and support groups.

2. Diagnosis

An early diagnosis can be based on the assessment of the paroxysmal manifestations, possibly confirmed by the genetic test.
Mutations of the *ATP1A3* gene are found in the majority of patients.
In case of no mutation in *ATP1A3*, the clinical diagnosis can be confirmed if the diagnostic criteria is fulfilled and after excluding any other differential diagnoses

Need: Family needs a clinical and genetic counselling about the prognosis and possible evolution of the disease. A multispecialty reference centre with considerable experience in AHC should take in charge the patient, directly or by overseeing a local centre.

Ideally: Family should receive advice about the management of the disease and all its coexisting neurologic issues, paroxysmal and non-paroxysmal as they appear at different ages. A rehabilitation plan (physiotherapy, speech and occupational therapy), must be defined as soon as possible.

3. Treatment

Antiseizure medication is used for patients with confirmed seizures and episodes of status epilepticus. Occasionally if the epilepsy diagnosis is uncertain, antiseizure drugs may be given on balance of risk vs. benefit.
For the other types of paroxysmal manifestations, Flunarizine and other drugs are used, albeit in open label experience only, both as prophylaxis and as acute treatment.

4. Surgery

No surgery is specific for AHC. In case of severe, drug-resistant epilepsy, VNS may be recommended by a reference centre. In case of associated cardiac disturbances, a defibrillator or pacemaker may be needed. Some may require a gastrostomy to support nutrition.

Need: The introduction of any new drug, especially if Flunarizine is not effective, should be discussed in detail and decided in full collaboration with the local neurologist and the AHC reference centre. Family should also be informed about the most known triggers for the AHC-episodes, and about the importance to identify the specific triggers for their child and determine the best ways to avoid them.

Ideally: A comprehensive treatment plan, including strict maintenance of a diary about different types of episodes, and emergency plan is helpful.

5. Follow Up (Childhood & Adolescence)

In addition to the paroxysmal manifestations, other symptoms appear early in childhood: mainly motor, cognitive and sensory deficits. Behaviour problems may also appear in childhood and adolescence. Sudden death is reported, in connection to severe epileptic seizures, prolonged AHC episodes or cardiac abnormalities.

6. Follow Up (Adulthood)

All manifestations, paroxysmal and non, persist to adulthood, with a great variability in their combination, frequency and severity.
The sudden onset of new paroxysmal manifestations, especially seizures, or the sudden worsening of any other symptom, with a consequent regression, may always occur, even in mildest adult patients.

Need: Parents and clinicians need to create a close, peer collaboration with their children's AHC reference neurologists, based on acknowledgment of respective roles and expertise of the disease. Adult patients and their families need to be supported in the transition from the paediatric neurology to the neurology for adults.

Ideally: The multispecialty team should provide regular follow-up visits, including a neurological, a neuropsychological, an ophthalmological and, in certain cases, a cardiological assessment (and other specialities where appropriate). They should also coordinate and support any other involved service: rehabilitation, education, social and home assistance, psychologic support, etc.