



6th Symposium on atp1a3 in disease AHC Family Organisations

- AHC families: a network of different organisations**
- Families main concerns and strongest wishes**
- How AHC families organisations already help research on AHC**
- Where and how scientists could get logistic and/or financial support**

By Dominique Poncelin (President of French AHC Family organisation)

ATP1A3 symposium- Tokyo 2017
Dominique Poncelin



AHC families: a network of different organisations

- Several AHC family organisations/foundations all over the world in North America / Europe / Asia , and a few families in Australia/Africa/South America, gathering more than 700 AHC patients. The AHCF directory has about 400 families in USA . About 200 European families are registered in various AHC Family organisations in Europe.
- Most of AHC organisations belong to AHCIA (AHC International Alliance – ahcia.org), however each organisation is independant with its own rules and separate budgets.
- Common talks are organised through teleconference every 3 months between AHC patient representatives for updating local activities – fundraising events – research support – new scientific publications
- Same common goals : Family support / scientific research support

ATP1A3 symposium- Tokyo 2017 Dominique

Poncelin



AHC families: main concerns / strongest wishes

Families main concerns

1. Attacks – pain – behaviour - future linked to AHC
2. Social life consequences for patients and all the family
3. Evolution of AHC from Childhood to Adulthood (40%).
4. Life expectancy (43 patients passed away since 1997)

Families strongest wishes:

1. A better quality of life for AHC patients (ie less attacks)
2. A better management (guidelines ?) of severe attacks to prevent from dramatic and non reversible brain damage.
3. Quicker results for on going AHC research projects (1 year does not get same meaning for scientists and families)



How to speed up scientific research on AHC

1. Money (Fundraising)
 2. Human material (DNA, tissues banks, clinical data bases, participation of families to studies)
 3. Advanced technology
 4. Determination from researchers to consider AHC as a priority to get quicker results than it is today
- The future of AHC research will depend on the capacity for scientists and AHC organizations to combine at the same time those 4 criterias



How AHC families organisations could help new projects to start and current projects to speed up

Logistic support

1. Clinical data base – blood bank – videos - photos
2. Participation of families to inquiry - questionnaires
3. Participation of families to clinical studies (treatments)

Financial support:

1. Individual fundraising performed by AHC organisations
2. Call for projects coming from patient's organisations
3. Individual grants allowed to scientist for AHC study
4. Co-funding from AHC organisations for larger projects

On going or past research projects funded by AHC Family organisations during last two years

<p>AHCF / USA www.ahckids.org</p>	<p>**"Molecular Physiology and Pharmacology of ATP1A3 Mutations in AHC" - Vanderbilt and Northwestern (USA) at a cost of \$1, 292,973 (over six years) **Vanderbilt - Antibody Acceleration Project - \$62,237 **Northwestern – Mouse Models – D801Y and G947R - Thomas Holmes \$130,000</p>
<p>AFHA/France www.afha.org</p>	<p>**Triheptanoïne oil pilot clinical trial – Pitié (Fr) : 108,000 € ** AHC/ATP1A3 molecular diagnostic – Lyon France: 6,400 € **Flunarizine effect on AHC (Northwestern – USA) : 40,000€ ** Granted towards a joint international project : 40,000 €</p>
<p>AHCUK / England www.ahcuk.co.uk</p>	<p>**Natural history in ATP1A3-related disease: a deep phenotyping-genotyping project (London) : 50,000£ ** Basic research on drug Rostaforoxin (Leeds): 25,000 £ **Granted towards a joint international project : 5,000 €</p>
<p>AESHA/Spain www.aesha.org</p>	<p>**IBAHC + AHC-MOME-IT: Molecular Mechanisms in AHC (Italy) : 10,000 €</p>
<p>AISEA/Italy www.aisea.onlus.org</p>	<p>**testing candidate molecules against effects of common ATP1A3 mutations : logistic support with I.B.AHC BioBank and Clinical Registry -partnership primary Italian pharma company</p>

Where and How scientists could apply for research support from AHC Family organisations

<p>AHCF / USA www.ahckids.org</p>	<p>Submit grant to Sharon Ciccodicola : sharon@ahckids.org Review by Medical Advisory Board Vote by AHCF Board of Directors Grant amount will be based on funding availability</p>
<p>AFHA/France www.afha.org</p>	<p>Permanent Application Forms available : dominique Poncelin d.poncelin@libertysurf.fr / d.poncelin@icloud.com Scientific evaluation by experts and vote by AFHA board Grant amount depends on evaluation result (up to 50,000 €)</p>
<p>AHCUK / England www.ahcuk.co.uk</p>	<p>Submit grant to Jill Bailey : ahcuk@outlook.com</p>
<p>AESHA/Spain www.aesha.org</p>	<p>Clinical data and biobank and Financial support application : contacts : rafi.munoz@aesha.org and albert@ahcfe.eu Financial capacity for research next 2 years: 50,000 €</p>
<p>AISEA/Italy www.aisea.onlus.org</p>	<p>Clinical data and biobank logistic support : www.ibahc.org Information : info@ibahc.org / franchini@aiseaonlus.org</p>

On behalf of AHC patients representatives, Thank you Dr. Sasaki, scientific committee and symposium committee for hosting and putting together such a wonderful conference.