

## MINUTES OF THE 10<sup>TH</sup> ANNUAL GENERAL ASSEMBLY Verona – September 14<sup>th</sup> 2014

Report : Françoise COURTOIS, Barbara BUTZECK, Jean-Daniel KAHN, October 10th, 2014

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### Agenda: *Appendix 1*

**In Attendance:** BRISOT Pierre (France), BUTZECK Barbara (Germany), COURTOIS Françoise (France), DELWART Bernard (Belgium), DISTANTE Sonia (Norway), EVANS Robert (UK), HORN SCHLEGEL Gerda (Switzerland), KAHN Jean-Daniel (France), LOREAL Olivier (France), Mc GRATH Ann (Ireland), MILMAN Nils (Denmark), MULLETT Margaret (Ireland), PINEAU Brigitte (France), PORTO Graça (Portugal), SANCHEZ Mayka (Spain), SORRILL Robert (Italy), STAKVIK Dag Erling (Norway), TEIXEIRA Emerencia (Portugal), TOSKA Ketil (Norway), VANCLOOSTER Annick (Belgium), VARKONYI Judith (Hungary).

**Apologies :** ALTES Albert (Spain), BARDOU-JACQUET Edouard (France), DE MAEGHT Stéphane (Belgium), FERNAU Janet (UK), RIALLAND Jean (France), RYAN John (Ireland), ZOLLER Heinz (Austria).

**Invited :** BADAR Sadaf (Italy) CREMASCO Chiara (Italy), GIRELLI Domenico (Italy), GRENI Federico (Italy), HORN Reinhard (Switzerland), LUSCIETI Sara (Spain), PIPERNO Alberto (Italy)

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President of EFAPH, **Barbara Butzeck** and Italian HH-Association President **Robert Sorril** opened the 10th Annual General Meeting.

Barbara Butzeck welcomed all the participants in the Palazzo della Guardia. The new members from Flanders, represented by Annick Vanclooster, the new president of HBA and Bernard DELWART, both Belgian and Nils Milman from the Danish Interest group Haemochromatosis were warmly welcomed.

Barbara thanked Domenico Girelli and the Ladies from COGEST for their outstanding work organizing the whole congress. She expressed her appreciation for the generous contribution given by the Associazione Emocromatosi, Monza to facilitate the EFAPH meeting.

Robert Sorril addressed the meeting and announced the handover of three awards to young scientists for their extraordinary work on hemochromatosis.

The third award was given to Annick VANCLOOSTER, Leuven-Belgium for her poster: Key-interventions derived from three evidence based guidelines, for treatment and follow-up of patients with HFE haemochromatosis.

The second award was presented to Federico GRENI, Monza- Italy for his poster: Proprotein convertase 7 (PCSK7) rs236918 variant is associated with increased risk of liver fibrosis in Italian patients with HFE-related hemochromatosis.

The first price was donated to Sadaf BADAR for her impressive oral presentation, titled: Targeted Next Generation Sequencing of the Five Hemochromatosis Genes in Italian Patients with iron overload and non-diagnostic first level genetic test: a pilot study.

### **I. Recent news on Haemochromatosis from the EIC : Pr Pierre Brissot (*Appendix 5*)**

Firstly, Pierre Brissot summarized the information on factors modifying the expression of HFE homozygosity after recognition that only 30 % male and 1% female persons with homozygous C282Y mutation got severe Iron overload: The phenotypic variability is modified by low CD8-T-Lymphocytes, elevated S100a9 calgranulin due to research of Graca Porto's group.



Pierre Brissot explained the role of CREBH (Pietrangelo's group) and chemokine CCL2 (Muckenthaler's lab), two proteins which led to iron overload and HH-symptoms in knockout mice. Regarding the visceral target Pierre Brissot explicated the co-factors for liver fibrosis which are high alcohol consumption, fatty liver, the genetic factors NRF2 and PCSK 7 (Graca Porto's and Alberto Piperno's working group).

Regarding diagnosis PB cited a presentation of Dorine Swinkels' group showing the toxic effects of NTBI (Non-transferrin-bound iron) and especially LPI (Labile plasma Iron) in levels of transferrin-saturation above 75 %. This fact may indicate that in future the transferrin-saturation is not only of interest in diagnosis but also treatment. Further studies are on the way.

A poster from Schönengel/Fischer revealed in MRI studies on patients with primary and secondary iron overload that the highest iron concentration in the pancreas is in the tail. Together with a fatty liver the risk for diabetes is higher.

A poster from Sonia Distante showed that lymphocytes from HH-Patients provide a suitable model for studying the effect of systemic to mitochondrial iron metabolism before and after venesection.

PB mentioned the oral presentation of Ela Nemeth, LA how the lack of hepcidin could be balanced: a) synthesis of mini-hepcidin, b) stimulation of hepcidin by inhibitor of matriptase – 2.

At the end PB discussed the question whether HFE knock out individuals have a physical advantage in youth by greater muscle mass.

## **II. EFAPH Activity Report 2012-2013 : Dr Françoise Courtois, General Secretary (Appendix 2)**

- Françoise Courtois (FC) summarized the activities of the Executive Committee during the 16 month period between the London AGM (April, 2013) and (September 2014): 1 board meeting, 5 Executive Committee meetings and 18 work sessions. She mentioned the main topics, details as per attachment.

- FC then reviewed the strong implication of EFAPH with Health organizations partners (EURORDIS, the European Hematology Association - EHA, European Patients Forum - EPF, Health First Europe - HFE. This keeps EFAPH up-to-date on the main trends and orientations in European Healthcare.

- Of particular interest are the EURORDIS membership meetings (Dubrovnik, June 2013 and Berlin May, 2014) aiming at developing European Reference Networks (ERNs) for Rare Diseases. In Berlin, the AGM was complemented by the 7<sup>th</sup> EUCERD meeting, dealing with ERNs and the implication of the European Commission. In June 2014 (Brussels), FC and Olivier Loreal (OL) attended the ERN awareness Conference organized by the EU Comm. and confirming the political implication. Please refer to Appendix 2 for more details.

- Regarding fund raising, FC reminded that a 3 years agreement has been signed with Etablissement Français du Sang (EFS) with FFAMH and EFAPH for promoting blood donation in France and Europe. Another grant was obtained from EURORDIS as a contribution to the organization of the AGM. The 3<sup>rd</sup> edition of the Golf ProAM at Ciccé-Blossac took place in October, 2013 in Rennes and the next one is planned in 2014. A new possibility is being explored with eFolia (a foundation of Celtic sponsors).

- In the field of communication, the cooperation with YLB Conseil (Communication consultant) has continued and HEMO News N°2 has issued.

- EFAPH's new logo has been unanimously approved (**Appendix 3**).

- EFAPH's membership was increased by the Danish Interest Group representing by Nils Milman and by the Belgian Flemish speaking Association (HVV) from Flanders represented by Annick Vanclooster.

- The support to National Associations and Groups has been continued: Switzerland, Austria, Nordic Countries, Belgian and Italy (new leaflet).

- Amongst the achievements FC conducted during 2 years a Pilot Project for sensibilizing GPs in a French department (Yvelines). (*Please refer Appendix N°6*)

- EFAPH produced also one scientific publications (Emerencia Teixeira, Portugal) and 3 posters (London, Berlin, Verona) and is contributing to 2 clinical thesis on "Early diagnosis and GPs" – University Paris-Diderot (Paris VII), General Medicine Unit (2013/2015).

- EFAPH's website has been transferred to Portugal and Emerencia Teixeira is now the webmaster.



- New projects for 2015: - A new application for the ERN has been filed by Olivier Loreal (RGIOD, rare European iron overload Network). - The test of clinical research about arthropathies (Pascal Guggenbuhl) has been started in France with a view to extending it to some EFAPH's members. - A survey on blood donation in the EU (Graça Porto, Emerencia Teixeira) has been initiated using a specialized online software; it is planned to develop some supporting documents in Europe for blood donation.

- Finally FC reminds the attendance that EFAPH celebrates its 10<sup>th</sup> birthday!

### **The activity report has been unanimously approved**

#### **III. Financial Report – Dag Erling Statvik (Appendix 4)**

Dag Erling highlights, for the year 2013, the expenses amounting to 9086, 14 € and the income amounting to 24.191, 70 €. The positive result is 15.105,56 €. Combined with the carryover from 2012 (12.398, 76 €) it amounts to 27.504,32 €. This is due particularly to the importance of the subsidies and gifts amounting to nearly 19.000,00 €. But, the latter do not recur.

The 2014 budget has been developed conservatively and shows a deficit forecasted to – 4.400,00 €. Moreover the Italian Association (Associazione per lo Studio dell' Emocromatosi) has kindly provided for the logistics of the AGM (in the order of 5.000 €).

After the financial report 2013 and the budget 2014 Dag Erling proposed to change the amounts for the membership fees from 2015 on. The purpose is to diminish EFAPH's structural deficit. After a discussion the following figures were approved:

< 100 paying members in national association:	50 €
100 – 300 paying members in national association:	200 €
> 300 paying members in national association:	300 €

### **The Financial report is approved unanimously**

#### **IV. Launch of the European reference network call: which place for haemochromatosis? Dr Olivier Loreal**

A conference organized by the EU takes place in Bruxelles in 2014 June : A call for the launch of European reference Network has been published the 10th March 2014. Provisional schedule suggests that the call will appear in mid-2015. The publication in the EU Official journal delivers the commission delegated decision and the commission implemented decision. Analysis of the two elements indicates criteria and conditions for a network, as well as criteria for membership of a network. These both type of criteria must be carefully examined in order to propose a network having a good shape. It is of particular importance to consider that the network must address highly specialized healthcare for rare and low prevalence complex disease, and that the coordinator of the network must be a practitioner member of the staff. After the technical evaluation of the project, a crucial phase for preselectionned projects, will be the evaluation of each project by the board of representant of member state. To be accepted, a project must obtain the simple majority.

For more details, see [http://ec.europa.eu/health/ern/docs/ern\\_delegateddecision\\_20140310\\_en.pdf](http://ec.europa.eu/health/ern/docs/ern_delegateddecision_20140310_en.pdf) and [http://ec.europa.eu/health/ern/docs/ern\\_implementingdecision\\_20140310\\_en.pdf](http://ec.europa.eu/health/ern/docs/ern_implementingdecision_20140310_en.pdf)

*The discussion highlighted the necessity to designate a Project manager and to include 1 or more Healthcare providers in the team.*

*The possibility to think to Pr Domenico Girelli or to Pr Graça Porto has been mentioned.*



## V. Actual situation of Blood Donation and Haemochromatosis – Pr Graça Porto- Emerencia Teixeira

The question is pending since 2012. In April 2014, the European Council, replying to Pr Porto, differed any action because of the lack of consensus. Reference has been made to the study by the Belgian Red Cross (Vox Sanguinis, 2013), showing large differences in blood donation policies across the EU. However, initiatives took place in 2013 (Belgium) and 2014 (Spain).

What is missing? The patients Voice to know the current practices in different countries/regions from the Patients' viewpoint.

A project is initiated to conduct an online enquiry by a Portuguese team (IBMC), Emerencia Teixeira being in charge. The interlocutors will be the EFAPH members. A questionnaire will be pre-validated in Portugal and confirmed by EFAPH members (November, 2014) and the translation should be available by mid-december. The inquiry will be closed early May, 2015 and the first dissemination of results is planned for September, 2015.

The members are requested to volunteer.

Meanwhile, work could start on pedagogical and promotional documents on blood donation (Pr G.Porto , Dr F.Courtois).

## VI. How to recruit new members by new media? Ketil Toska (*Appendix 6*)

Ketil has presented the use of a social network (Facebook) in Norway resulting in increasing significantly the membership of NHF.

- The starting point was a survey identifying the main sources from which the NHF patients heard first about the Association. It was found that the younger people generally use the Internet, the newspapers readers were 12-13 years older. These 2 channels of communication accounted for in the order of 80% of replies and were of equal importance.
- Highlights were that the youngest people generally use the Internet, the newspapers readers were 12-13 years older. These 2 channels represented in the order of 80% of replies and of equal importance.
- How to recruit new members via social media? The approach has been to open a page on Facebook for discussion between the patients with closed access restricted to invited persons.
- Status for the Nordic Facebook page. As of September 2014 there were 300 participants, mostly Norwegians but also some in the other Scandinavian countries.

It can thus be considered as an efficient and relatively easy way to generate membership.

## VII. Quality of life for HH patients . Annick Vanclooster

First Annick Vanclooster, Treasurer of HVV (new member), reported the start of the Haemochromatose Vereniging Vlaanderen (HVV) in April 2012 after a survey with 250 HH-Patients at the University Hospital Leuven 2011 when the half of the group wish to have a peer group, get information and practical help. In August 2014 HVV had 52 members. Since the start HVV organized 4 meetings with different topics. AV explained that in Belgium HH-Patients can donate blood under "certain conditions" although these conditions were not determined yet. [www.haemochromatose.be](http://www.haemochromatose.be) is the website of HVV.

Then AV presented here PhD project:

The purpose of this study was to develop key-interventions (KI's) to measure and improve the quality of care (QoC) delivered to patients diagnosed with HH.

A RAND-Modified Delphi method was used to develop KI's. The first round consisted of a scoring form to prioritize the recommendations extracted from evidence-based guidelines. The results of the survey were discussed during a consensus meeting followed by a final appraisal of the selected recommendations that resulted in a list of measurable KI's. Initially, a list of 41 key recommendations on screening, diagnosis and treatment/management was extracted from three existing guidelines on HH (European Association for the Study of the Liver EASL, American



Association for the Study of Liver Diseases AALSD and Dutch guideline on HH). Finally, a core set of 24 recommendations resulted in 15 KI's to measure and improve the quality of care delivered to patients with HH. A quantitative retrospective cohort study of 350 HH\_Patients files were done at the University hospital Leuven.

### **VIII. The long way from a local support group to a nationwide patient organisation – Example Switzerland - Gerda Horn**

Gerda Horn reported how the nationwide association “Haemochromatose Schweiz” was founded on February 17th, 2014 in Waengi, canton Thurgau, Switzerland. In Weinfeldern existed a small self support group since 2008 before several attempts to grow up by contacting medical staff, experts and other HH- Patients were successful. The group in Weinfeldern (10 Patients) meets every second month, a second regional self-help group in Zurich (10 Patients), founded in 2013, meets every month. Future actions will focus on gathering intended groups who are already contacted in other German-speaking cities like Basel, Bern, Luzern, Lachen SZ and Chur.

### **IX. News of IAHA - Pr Robert Evans**

Robert Evans, president of IAHA first summarized the short history of the organization, “re-established” in London 2013 with the founder members EFAPH, the Haemochromatosis Societies of Australia, Canada, Brazil and South Africa. Board members are Sara Fernau, UK (Secretary / Treasurer), Ben Marris, Australia (president elect (from 2015).

Rob introduced the objectives of IAHA, the drafted constitution and initial rules and reports about the difficulties to get a registration with the UK Charity Commission since the bank account has not enough funds. Due to a personal changing in responsibilities at “Novartis Iron Health Alliance” the promised grant of \$ 20,000 did not yet materialize. Rob had contacts with Novartis associates during the last 48 h and hopes to succeed.

[www.haemochromatosis.info](http://www.haemochromatosis.info) will be the future web address of IAHA.

Rob appreciates Françoise Courtois's brother who designed the IAHA Logo. He reported from his attempt to secure the musician Mike Oldfield as patron.

### **X. A way to sensitize GPs: a regional Observatory in France (Project pilot) Dr F.Courtois (Appendix 7)**

The project originated from the Lunch Debate in the EU Parliament (Brussels 2011) and the position statement from the EU Commission (2012). The main objectives are to develop the GPs' awareness (early diagnosis), to develop “HH Observatories” and to have the basis for a EU “federal project”.

As a first phase, an experiment has been conducted in the French Yvelines (78) Department with the implementation of the Observatory. As a result the main healthcare organizations concerned have been identified. Four indicators have defined to follow from 2012 on the impact of the different actions.

The 2013 campaign to raise GPs awareness resulted in a significant increase in the number of new GPs sending patients for an iron problem, as well as the number of patients per GP. There was also a high proportion (60%) of new patients.

The methodology (Awareness campaign) based on simple, userfriendly and pragmatic tools proved to be efficient. This precedent is used as a basis for a similar project starting now in French Brittany and in Paris/Ile de France region (2015). Portugal has expressed a strong interest.

Françoise emphasized the impact evaluation aspects through the GPs, Biochemical Labs and Genetic Labs.

In fine, the French experience should cover in the order of 20.000 GPs in 2015 and should serve as a platform to export in some countries members of EFAPH. It would be then worthwhile to envisage the setup of a European working group within EFAPH.

### **XI. Conclusion and Perspectives - Barbara Butzeck**



Barbara Butzeck gave a short update on the project Research in HH-Arthropathy:

- A German/Austrian group (Prof. Georg Schett, University Erlangen) works on high resolution CT imaging and found differences between HH-Arthropathy, rheumatoid arthritis and osteoarthritis.
- Prof. Pascal Guggenbuhl just started a study in Rennes, France on the drug "Anakinra", a biological IL-6 receptor antagonist which is well established in polyarthritis.
- BB suggested to use the new tool Survey Monkey (user license available free of charge to EFAPH members until June 2015), to create a questionnaire to ask patients about their experiences with different treatments.
  
- BB reported about the changing of the EFAPH webmaster from Germany to Emerencia Teixeira, Portugal who thankworthy just started with an update which should follow about 3-4 times a year.
- Listening to many presentations on rare forms of HH BB asked the Scientific Board who could be a responsible person for questions regarding rare forms.

#### Announcements for future meetings:

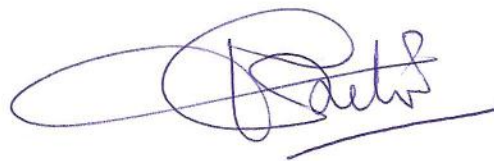
- The European Iron Club will NOT have a meeting in 2015.
- The next EIC meeting will be in Innsbruck, Austria, in April 2016
- "Biolron 2015" will be held in Hangzhou, China, from 6th to 10th September, 2015 (According to the IBIS President HH might not be a big issue there).
- After a short discussion about EFAPH AGM in 2015 it was agreed that it should be in Europe, preferably in October in a location with good accessibility for all participants. Robert Evans proposed to invite members of the IAHA to have a Co-Meeting.

Barbara closed the meeting thanking all participants for their presentations, questions and discussion and gave again sincere thanks to Robert Sorrill from the Italian HH association.

**The preliminary date for the next EFAPH General Meeting is October 2015**  
**in Discussion: Cologne, Germany (on 8<sup>th</sup> Nov. 2014 HVD board meeting will decide)**



**Barbara BUTZECK**  
(EFAPH President)



**Françoise COURTOIS**  
(EFAPH General Secretary)

#### Appendixes:

1. Agenda
2. Activity Report 2013-2014
3. New EFAPH's logo
4. Financial Report 2013-2014
5. Recent News on Haemochromatosis
6. Use of Facebook in Scandinavian countries
7. Pilot Project: GPS awareness and a HH Observatory