

MINUTES OF THE 11TH ANNUAL GENERAL ASSEMBLY Cologne – october 16th – 17th 2015

Report : Françoise COURTOIS, Barbara BUTZECK, Jean-Daniel KAHN, December 30th, 2015

Agenda: **Appendix 1**

In Attendance (at the AGM, October 17th): ABELE Maria (Hungary), BRISSOT Pierre (France), BUTZECK Barbara (Germany), COUESLANT Béatrice (France), COURTOIS Françoise (France), DELEU SNYERS Anne-Sophie (Belgium), DELWART Bernard (Belgium), DON Howard (UK), EVANS Robert (UK), EVANS Patricia (UK), FINZEL Stephanie (Germany), GIRELLI Domenico (Italy), HEAD David (UK), KAHN Jean-Daniel (France), LAMBEETS Geert (Belgium), Mc GRATH Ann (Ireland), MARRIS Ben (Australia), MULLETT Margaret (Ireland), PINEAU Brigitte (France), PORTO Graça (Portugal), PÖTZ Michael (Germany), SANCHEZ Mayka (Spain), SANTOS Paolo (Brasil), SORRILL Robert (Italy), TEIXEIRA Emerencia (Portugal), TOSKA Ketil (Norway), VANCLOOSTER Annick (Belgium).

In Attendance (at the Scientific Day, October 16th): ARENS Ursula (Germany), BLUMENROEDER Stephan (Germany), CAPPLIS Anna (Ireland), COLCLOUGH Fiona (Ireland), DEGUELDERE Gisela (Germany), POTTHOF Dietrich (Germany), FOLLEA Gilles (France), FRITSCH Renate (Germany), GAMMEL Yvonne (Ireland), GATHOF Birgit (Germany), GRUHN Kerstin (Germany), GRUENBERG Heinz (Germany), GRUNDWALD Bernd & Hilke (Germany), GUGGENBUHL Pascal (France), JOBLING Majella (Germany), KADAR Janos (Germany), KAISER Jutta (Germany), KIELY Patrick (UK), KLUMANN T.W (Germany), KREIS Annemie (Germany), KREUELS Til (Germany), NIEDNER Hartmut (Germany), NIEDERAU Claus (Germany), NOACK Silvia (Germany), OBERLIESSEN Luise (Germany), RUDDOCK Hazel (Ireland), SHEMELD Steven (Germany), SCHLUETER-BRUST Klaus (Germany), SCHMECK Bernd (Germany), VAN DER WAART Menno (Netherlands).

Apologies : ALTES Albert (Spain), BARDOU-JACQUET Edouard (France), DE MAEGHT Stéphane (Belgium), DISTANTE Sonia (Norway), HORN SCHLEGEL Gerda (Switzerland), LOREAL Olivier (France), MILMAN Nils (Denmark), RIALLAND Jean (France), RYAN John (Ireland), STAKVIK Dag Erling (Norway), ZOLLER Heinz (Austria).

I . Scientific and Medical Session

Barbara Butzeck welcomes warmly the attendees from Europe, Australia and Brazil. She pointed out, that she is looking forward to interesting scientific presentations and a lively discussion between patients, clinicians, scientists and nurses-all “players” in the care of HH patients. She thanks Dr. Schlueter-Brust for his hospitality to hold the meeting in the old chapel of the St.Franziskus-Hospital. She introduced Prof. Claus Niederau, co-founder of HVD and expert in HH as moderator.

1. **Recent news in HH – Report from Biolron Hangzhou China September 2015 : Pr Pierre Brissot**
 - a. Genetic abnormalities have been identified, which favor in hemochromatosis the development of iron overload or of cirrhosis. If confirmed, these results should, in the future, lead to useful prognostic markers.
 - b. It has been shown that iron enters the heart by a different way than used for the liver and the pancreas. The exact nature of this pathway remains, however, to be determined.

- c. An Australian epidemiological study has defined, for the first time, the respective prevalence of HFE versus non HFE hemochromatosis (HC). The « classical » form of HFE-HC (C2828Y/C282Y or type 1 HC), clinically expressed, would be present in approximately 1/1000. As to the non HFE HC, type 2 HC (related to hemojuvelin gene mutations) is estimated at 1/5000 000), type 3 HC (involving mutations of transferrin receptor 2 gene) at 1/6000 000, and type 4 HC (« ferroportin disease ») at 1/1000, therefore a prevalence close to that of type 1 HC (probably due to the fact that the ferroportin disease has a dominant mode of transmission).
- d. From the diagnostic viewpoint, the interest of high throughput techniques (NGS = next generation sequencing) for determining mutations has been presented. This technology provides simultaneously a large number of mutations and represents a very powerful new diagnostic approach. However, a significant limitation is related to the fact that, an increasing number of novel mutations being identified, the key point is to know whether they are not deleterious. It is also essential that the indication of such a methodology is guided by a very rigorous clinical analysis.
- e. Experimental studies are still on the way for evaluating the therapeutic interest of mini-hepcidins in order to counteract hepcidin deficiency observed in hemochromatosis and in various hematological conditions.

2. Blood donation and HH:

- a. ***Dr Gilles Follea: A European point of view:*** Dr Gilles Follea, formerly EFS (French Blood Establishment) Executive and EBA (European Blood Alliance), presented an overview of the pros and cons of Blood Donation: policies and recent evolutions in blood use, main findings and reservations and ethical principles for donors and patients. Suitability of blood from HH patients, possible reservations bacterial and viral residual risk, erythrocytes dysmorphism. A general discussion has arisen with possible follow-up (Pat Evans, Gilles Follea,...). *The whole presentation is available on www.efaph.eu*
- b. ***Pr Birgit Gathof: Experiences with the “Cologne model”***
Pr. Birgit Gathof, Director of the Bloodbank at University Cologne describes her experiences with Blood donation and HH. According to a Votum of the Ministry of Health, Germany from May 2015 HH-patients can donate blood if their Ferritin is in normal ranges. At University Cologne all blood donors get a check on their Ferritin. According to this activity the Blood bank recognized carriers for the homozygote C282Y-Mutation in 1:1000 cases. This is less than expected, probably due to a negative pre-selection of persons with high liver enzymes. HH-Patients can have phlebotomies at University Cologne on special times and the frequencies, they need. If the blood is used for transfusion, “normal” donors get a reimbursement; HH-patients in contrary will get an analysis of their blood with all iron parameters. In conclusion there is an advantage for both sides: the blood bank gets blood and the patients get treatment, analysis and finally persons at risk are identified.
- c. ***Mayka Sanchez: The Cataluna approach in Spain***
Dr Mayka Sanchez sums up the process : further to a discussion between Dr Altès (AEH president), Dr Contreras (Head of Blood and Tissue Bank) and Mrs Gloria Renom (member of Cataluna Parliament) in November , 2014, a motion is adopted by the Cataluna Parliament in April, 2015, requesting the Spanish Parliament (Madrid, June 2015) to modify the National Royal Decree on Blood Donation.
- d. ***EmerenciaTeixeira: EFAPH-Survey on blood donation:***
Emerencia presented the preliminary results of the survey. In general the response from the members of the national associations has been more than satisfactory. It varies from country to country and will require further analysis. Emerencia will present the final results at the next AGM.

e. Round table: harmonization in EU (Pr Graça Porto)

Following all the presentations, a round table discussion was initiated where the speakers were posed questions by the audience regarding the issues raised in their presentations. The debate was very lively, with patients manifesting their worries and concerns about the lack of harmonization amongst different countries, regions or centers and the experts manifesting the difficulties in practice, to reach such a harmonization. The issue of the blood safety was also discussed, some participants having highly criticized the presented published evidence of anomalies in HH red blood cells. It was concluded that an action must be taken to either reject that study or to conduct new reliable studies on the biology of red blood cells in HH.

3. Transferrin-Saturation: Management in HH- EFAPH promotes clinical, practical recommendation : (Pr. Pierre Brissot)

Although the concentration of plasma ferritin remains the major indicator of iron load stores in HC, TS (which reflects iron circulating in the blood) presents a number of interests :

1. Diagnostic interest. Increased transferrin saturation is the earliest biochemical abnormality in HC and represents, therefore, the first line diagnostic indicator of the disease. Two successive determinations are, however, necessary due to the variability of this parameter.
2. Prognostic interest. When TS level is over 75% (normal <45%), the presence in the blood of a new iron form, potentially toxic and possibly clinically significant, named non-transferrin bound iron, is possible.
3. Therapeutic interest. Whereas TS has no interest during the induction phase of venesections (because its levels remain very high until the very end of the treatment), it may be useful to monitor this parameter, for example twice a year, during maintenance therapy, in order to ensure that, despite normal satisfactory ferritin levels (around 50 µg/L), TS is not over 75%. In practice, it is useful to keep in mind the rule of the « 50 » : 50 for ferritin, fifty for transferrin saturation...

4. HH-Arthropathy:

- a. Dr Patrick Kiely: An overview and the UK experience:** Dr P.Kiely (St Georges Hospital London, UK) provides an overview on the disease as well as the UK experience, comparing the statistics of his St Georges Clinic (62 patients, Gr1) with the survey carried out by the Haemochromatosis Society UK (470 patients, Gr2). The details are available on www.efaph.eu . Noteworthy is the elapsed time between the first symptoms and the diagnoses: 47% > 5 years and 31% between 1 to 5 years. Interestingly some differences can be noted between the groups: ankle + hip , Gr 1 twice more than Gr2 and wrist Gr2 52% vs 34% Gr 1. Dr Kiely pointed also out that venesection was not efficient with respect to pain (for 86,4% patients venesection has no effect on pain and, in some cases even increases it). In conclusion, the major concern of the patients is the late diagnose.

b. Dr Stephanie Finzel: Typical features in (HRpQ) Computed Tomography

Dr.St. Finzel, Rheumatologist at University Erlangen, Germany, presented a special technique of Computed Tomography, called HR-pQCT . She showed typical features in high-resolution imaging of finger joints in Haemochromatosis, compared to findings in Psoriasis and Rheumatoid Arthritis. In her summary she pointed out the main results in differential diagnosis of inflammatory and degenerative joint diseases: 1. Erosive lesions, bone cysts and osteophytes show distinctly differentiable patterns of involvement: - PSA: wide-spread, corona-like osteophytes; presumably involvement of capsule and tendon insertions; HH: cartilage-bone interface and joint margin. 2.

Bone cysts are rare in mainly inflammatory diseases. 3. Presumably there exist different mechanisms of bone turnover in RA, PSA and HH.

- c. **Dr Pascal Guggenbuhl: Anakinra in HH-Arthropathy:** Pr P. Guggenbuhl (Pontchaillou hospital, Rennes France), reminds that one of the major patients complains are joint pains and that the currently available treatments are not satisfactory. Anakinra appears to be the most promising direction. The THERA project (clinical trial) has been started with 2 teams (Paris and Rennes). The primary objective is to measure the efficiency improving joint pain at day 15; secondarily to appreciate the improvement on other clinical signs more generally the quality of life. The criterias for patients inclusion or no-inclusion are restrictives and are detailed in Appendix **(Appendix 3)**. It is considered that on a phase III trial basis that Anakinra is not justifiable if the rate of success is below 40%. Currently 7 patients are included in the trial. It is envisaged to extend it in some EU countries covered by EFAPH.
- d. **Pr. Klaus Schlueter- Brust: Hip and knee replacement in HH-Arthropathy** (St. Franziskus-Hospital, Cologne). Pr Schlueter-Brust has commented a video-documentary showing the details of the operatory technics put into effect for hip and knee replacement. This has brought a very concrete vision of these processes.
- e. **Dr Barbara Butzeck: Patients' experiences and perspectives.**
In a short but very authentic presentation Barbara describes the problems of HH-Patients with HH-Arthropathy as a chronic situation with pain and stiffness. She especially focus on handicapped / inability of hand and foot function which could cause problems in personal hygiene and daily activities, loose of social contacts in terms of hobbies and communication aswell as untimely retirement. She concluded that it is essential to start a working group with experts in rheumatology and HH-patients. Pr P.Guggenbuhl, France , Dr St.Finzel, Germany and Dr P. Kiely, UK agreed to start this project with EFAPH.
- f. **Rare forms of HH – European Reference Network: Pr. Domenico Girelli,** (University Verona, Italy). Pr. D.Girelli has given an excellent and pedagogical overview on this theme. His presentation is available on EFAPH's website (www.efaph.eu).

II. Annual General Assembly

President of EFAPH, **Barbara Butzeck** opened the 11th Annual General Meeting.

1. **EFAPH statutes: Jean-Daniel Kahn (JDK).**

JDK reminds the Assembly of the Executive Committee's (Bureau) concern to provide structurally the Federation with adequate continuity in terms of governance. It is therefore proposed (art.7, para.1) that the "outgoing" President be statutorily Vice-President and member of the Board.

JDK has taken this opportunity to work with (volunteer) Lawyer Sylvie de Vendeuil for clarifying the Statutes' presentation and improving the compliance with the French Law on Associations (Loi de 1901), with no changes to the other Statutes' basic provisions. The revised Statutes have been circulated to the members prior to the AGM.

JDK recapitulates for the Assembly the highlights of the Statutes:

1- The members (National Associations and individuals) must be up-to-date with their dues to participate in the debates.

2- The National Associations members appoint one representative to exerce their right of vote (5 votes each, whereas 1 vote for each individual member, art. 9, para.4).

3- The Board (Conseil d'Administration) is comprised of 3 up to 11 members, elected for 2 years by the General Assembly. The Executive Committee (Bureau) is designated by the Board.

4- The members of the Board and of the Executive Committee are not remunerated. They are reimbursed of their expenses upon justification (art.8, para. 4).

5- English is the working language of the Federation. The Associations members may translate the documents from EFAPH into their own language. In any case a translation into French will be provided to be made available to the French Authorities (art. 6, para. 2).

Statutes V11, see Appendix 4

The statutes are unanimously approved .

2. EFAPH Activity Report 2012-2013 : Dr Françoise Courtois, General Secretary

FC summarized the activities of the Executive Committee during the 12 month period between the Verona AGM (September, 2014) and (October, 2015): 1 Board meeting, 3 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. EFAPH has been represented at 5 international manifestations.

- Of particular interest are the EURORDIS membership meetings (Madrid, May 2014 and the 2nd European Reference Networks (ERNs) for Rare Diseases Conference, Lisbon October 8-9). Pr. Graça Porto joined FC at the Lisbon Conference and confirmed the strategic importance for EFAPH to be involved in a large Grouping of Haematological RD and to induce all EFAPH national members for obtaining the support of their government to designate National Reference Centers. Also, JDK represented EFAPH at the EPF General Assembly, at which the European Empowerment Campaign was launched, with participation to further workings groups. Please refer to Appendix 2 for more details.

- Regarding fund raising, the partnership agreement with Etablissement Français du Sang (EFS) has been reconducted. Another grant was obtained from EURORDIS as a contribution to the organization of the Cologne AGM. The 4th and last edition of the Golf ProAM at Ciccé-Blossac took place (October, 2014 in Rennes). The generous contribution of Associazione per lo Studio dell' Emochromatosi to the Verona AGM should also be mentioned.

- In the field of communication, the cooperation with YLB Conseil (Communication consultant) has continued and HEMO News N°3 has been issued. The general discussion evidenced the need to create a Redaction Committee as well as the necessity to cross-communicate the national newsletters.

The success of Erwan Leroux in the "Route du Rhum" transatlantic race (November 2014) has been highlighted by his acceptance to be EFAPH's ambassador, carrying the new logo.

- Amongst the achievements the Portuguese team conducted a survey on Blood donation practices in the 12 EU countries, members of EFAPH.

The rollout of the GPs awareness campaigns continued (AHO – French Brittany and Paris area). Further developments expected in some EFAPH members countries.

- EFAPH produced also one poster in Verona September, 2014 (Emerencia Teixeira, Portugal) and is contributing to 2 clinical thesis on "Early diagnosis and GPs" – University Paris-Diderot (Paris VII), General Medicine Unit (2013/2016).

- New projects for 2016: - Involvement in ERN (Graça Porto, Domenico Girelli, Pierre Brissot) – Launch of working group on HH Arthropathy (Barbara Butzeck -Pascal Guggenbuhl) with a view to extending it to some EFAPH's members – A Charity piano concert by artist Yves Henry.

Annual Activity Report 2015 : Appendix 5

The activity report has been unanimously approved

3. EFAPH Financial Report – Jean-Daniel Kahn (*Appendix ???*)

Dag Erling Stadvik, Treasurer, being excused the financial report is presented by JDK.

The total revenues (01/01/2014-31/12/2014) amount to 7 026,21 €, of which:

- membership fees	1 450,00
- collects and grants	5 003,00
- financial income	573,21

The total expenses (same period) amount to 9824,52 €. The main items are:

- travel	5 205,35
- printing/communication material	2 263,76
- mail/telecom	1 201,93

The deficit for year 2014 amounts to 2 798,31 €

JDK points out that most revenues are not of recurring nature, except the membership fees; the increase of the latter voted at the 2014 AGM does not prevent the Federation to be structurally in deficit. The discussion underlines the necessity to secure some permanent funding.

JDK expresses his thanks to (volunteer) Béatrice Coueslant, in charge of EFAPH's book keeping as Assistant to the Treasurer.

Financial Report 2015: Appendix 6

The Financial report is approved unanimously

4. **Scientific Committee: new members and projects (Pr Graça Porto).** GC proposed 2 additional members :
Pr. Pascal Guggenbuhl (Arthropathy expert) and Pr. Domenico Girelli (HH Rare forms and ERN).

Activities of the Scientific Committee focused on two main projects: 1) integration in a future European Reference Network (ERN) on Rare Hematological Disorders, and 2) acceptance of HH subjects as volunteer blood donors. Specific actions included: 1) the participation in the 2nd Conference on European Reference Networks held in Lisbon on the 8-9 October. A joint action was initiated with representatives of the European Hematology Association (EHA) and patients advocacy groups in order to foster the creation of a group of rare hematological diseases. 2) A survey on the general awareness and practices of blood donation by HH subjects was conducted with the participation of all national associations. An update on the evolution and present state of both projects was presented and discussed during the meeting.

5. **European Patient Forum (EPF) –Patient Empowerment Campaign (PEC): Jean-Daniel Kahn**

Jean-Daniel Kahn (JDK) represented EFAPH at the AGM of the European Patients Forum-EPF (May 2015, Brussels), on occasion of which the Campaign has been officially launched. EFAPH is an active member of EPF which is the major “umbrella” patients organization in the EU (over 60 members) and which will pursue in 2016 a high visibility action aiming at convincing the Members of the European Parliament (MEPs) to act upon their respective Governments so that the latter integrate the concept in their structures, procedures and action plans.

The concept, since EFAPH's very beginning, is quite familiar to its National Associations members: the governance and actions of the Federation and of its members have always been entirely patients-centered. It consists in promoting a common understanding so that the patients, individually and collectively, are full partners in the decision making about treatments, health systems, quality of life, thus “sharing the power” with other stakeholders such as health professionals.

The PEC key dates are:

- May-Nov. 2015, workshops,
- Nov. 2015 - Spring 2016, development of a roadmap, a charter and a toolkit,

- Spring 2016, EP event,
- June 2016, exhibition at the EP + lobbying (national and European).

EPF's summary document covering the PEC has been distributed to the attendants (**Appendix 7**). The Assembly recognizes the importance of the action and agrees on the principle of EFAPH's participation. A final decision will be made when more elements are available.

6. **A 2014/2015 informal review of the 13 EFAPH member associations (F.Courtois- B.Butzeck)**

A different approach is used this year : rather than having each member detail the highlights of his Association a thematic consolidation of their contributions is presented by FC and BB.

- Various happenings have taken place for the HH national awareness days. For instance in Spain (Barcelona June 5th, 2015) various media communications (TV interviews, meetings...) took place with the participation of Barbara B. and Pierre B. Hungary , Belgium, Ireland (over 34 venues throughout the country) have also contributed by informations stands.
- For the Rare disease Day (February 29th), Germany and France are noteworthy (festive meeting in Dusseldorf and Orphanet parade in Paris streets).
- On a regional level (cross-border interaction) a sensibilization day (May 18th, 2015) has been organized in Mulhouse jointly by Alsace and Rheinpalz. A Nordic Facebook page has been issued to survey the HH status in Iceland, Denmark, Sweden, Finland and Norway.
- Government lobbying: the Cataluna Parliament petitioned officially the Spanish Parliament for changing the national Blood Donation law; national guidelines for HH treatment have been presented to Norwegian Health Authorities; in Ireland, the Minister of Health Leo Varadkar launched the HH awareness campaign (May 29th).
- Nice initiatives, which could inspire other members, include a pedagogical and attractive Portuguese HH handbook for elementary school students, an Italian phone app of Iron Tracker for HH patients and in France "The Little Iron Man", a cartoon type of silhouette which lends itself to jokes, slogans, ...
- Management structures: the new board in UK is comprised of Howard Don (chairman, replacing Janet Fernau) and David Head (chief executive officer) and in Hungary, Maria Abele succeeds Judith Varkonyi as Chair person.
- GPs awareness: 2 large campaigns in France (Brittany and Paris area under progress) and in Portugal launch of a mapping of HH Clinical Centers to be dispatched to GPs.

7. **Elections to the Board** : As previously reminded by JDK, the Board is renewed as follows.

Barbara Butzeck	11 voices
Françoise Courtois	11 voices
Dag Erling Stakvik	10 voices
Ketil Toska	11
voices	
Mayka Sanchez	11 voices
Robert Evans	10 voices
Ann Mc Grath	10 voices
Maria Abele	9 voices
Graça Porto	11 voices
Robert Sorrill	10 voices
Annick Van Clooster	9 voices

All candidates have been elected. The Board is comprised of 11 members and designated its **Executive Committee**:

President	Barbara Butzeck (Germany)
General Secretary	Françoise Courtois (France)
Treasurer	Dag Erling Stakvik (Norway)
Vice-President	Mayka Sanchez (Spain)
Vice-president	Ketil Toska (Norway)
Sr.Adviser	Jean-Daniel Kahn (France)

Moreover, Pr Pierre Brissot and Mr Jean Rialland are confirmed as Honorary Vice-President.
Please refer to **the Appendix 8: Organization Chart**

Conclusion and Perspectives -

Barbara Butzeck closed the meeting , thanking all participants for their engagement and lively discussion.
She announces the next AGM in April at Innsbruck, Austria in the frame of the EIC April 7th to 10th, 2016 .

Announcements for future meetings:

**The preliminary date for the next EFAPH General Meeting is during the next EIC Meeting, April
7 – 10 , Innsbruck Austria**



Barbara BUTZECK
(EFAPH President)



Françoise COURTOIS
(EFAPH General Secretary)

Appendixes:

1. Agenda
2. Activity Report 2013-2014
3. P Guggenbuhl: Anakinra in HH-Arthropahy
4. EFAPH Statutes (V11)
5. EFAPH Annual Activity report 2015
6. EFAPH Financial report 2015
7. EPF: Patient Empowerment Campaign (PEC)
8. Organization Chart

