FIRST EUROPEAN CONGRESS on Hereditary ATTR amyloidosis
IMPROVING CLINICAL MANAGEMENT
November 2 and 3, 2015

Co-organizers David Adams (Paris, France) and Teresa Coelho (Porto, Portugal)

More information
www.ec-attr.com

PROGRAM

Institut Pasteur - 25-28 rue du Docteur Roux - 75015 Paris - France
Hereditary ATTR amyloidosis are rare diseases due to transthyretin gene mutations. They present usually as Familial Amyloid Polyneuropathy (FAP) or Cardiomyopathy (FAC). This is an irreversible and lethal debilitating systemic degenerative disease involving also kidneys and eyes. FAP have been described in northern Portugal and Sweden 60 years ago in endemic areas and sporadically in several European countries 25 years ago with the help of genetics. Considerable therapeutical progress have been made concerning the anti-amyloid therapy for FAP by liver transplant in Sweden 25 years ago, oral medications with TTR tetramer stabilizers, leading to marketing approval in Europe three years ago to slow the progression of the neuropathy at an early stage. Currently, clinical trials are ongoing to evaluate TTR stabilizers in FAC, TTR gene silencing strategy in FAP and FAC. Treatment of visceral amyloid and symptomatic treatments are also major to improve the quality of life: digestive and sexual disorders and pain. Recently, a therapeutic education program has been developed. A long way still to go to improve and standardize diagnosis and care of HATTR at the European level. That requires first a structuring at a national level, as has been done in France 10 years ago with a certified national reference center for FAP. Its’ aim is to improve the diagnosis, therapy and genetic counseling in connection with a national network. An European network regrouping national reference centers has been developed since 2012. The European Congress for HATTR will allow the meeting of the specialists of all European countries and the sharing of experience. The stakes are major: to further improve the early diagnosis of sporadic cases and genetic carriers, to review anti-amyloid treatments and clinical trials, to improve genetic counseling. This is an opportunity to bring together patients associations. At less than 2 hours from the European capitals, Paris hosts the first conference. We hope to welcome you numerous to this major event.
Monday, November 2

8:45 – 9:00
Welcome to the congress

9:00 – 9:30
TTR amyloidosis: a scientific journey since Andrade
(M. Saraiva)

9:30 – 10:30
Epidemiology and phenotypes in ATTR amyloidosis

Oral communications:
DISCOVERY: A Study Examining the Prevalence of Transthyretin Mutations in Subjects Suspected of Having Cardiac Amyloidosis.
(Olakunle Akinboboye)

Unravelling the epidemiology of late-onset and asymptomatic carriers of FAP ATTR V30M in a Portuguese population.
(Carolina Lemos)

TTR-FAP: A single-center experience in Sicily, an Italian endemic area.
(Anna Mazzeo)

Clinical, Epidemiological, Genetic, and Electrophysiological Characteristics of Transthyretin Familial Amyloid Polyneuropathies in Israel.
(Menachem Sadeh)

Epidemiology of Familial Amyloid Polyneuropathy in Bulgaria.
(Stayko Sarafov)

The hidden story behind gender differences in familial amyloid polyneuropathy (FAP) ATTRV30M.
(Diana Santos)

Familial Amyloidotic Polyneuropathy in Crete, Greece
(Minas Tzagournissakis)

The phenotypical expression of an European inherited TTR amyloidosis in Brazil.
(Márcia Waddington Cruz)

Wild-type transthyretin amyloidosis in female patients.
(Arnt V. Kristen)

10:30 – 11:00 Break
11:00 – 11:45
Neuropathic Phenotypes and natural history of FAP (D. Adams)
Cardiologic Phenotypes and natural history of FAC (C. Rapezzi)
Emerging CNS involvement in FAP-TTR long survival patients (L. Maia)

12:00 – 13:30 Lunch + Industry sponsored symposia (Pfizer)

13:30 – 14:15
Diagnosis
Do we need to demonstrate amyloid in tissue ? Pros and Cons
(P. Westermark, L. Magy, M. Melo Pires)

14:15 – 15:00
Minimal assessment of index cases
The point of view of the neurologist, cardiologist, ophthalmologist
(M. Slama, E. Hund, N. Ferreira)

15:00 – 15:30 Break

15:30 – 15:50
New tools to diagnose and follow FAC patients :
Biomarkers (G. Merlini)

15:50 – 16:10
Imaging Amyloid
(Ph. Hawkins)

16:10 – 17:10
New tools for diagnosis
Oral communications :

DPD Scintigraphy for diagnosis of amyloidosis in 1191 patients– a single centre experience
(David Hutt)

Quantitative MR-neurographic parameters can determine and specify nerve injury in amyloid related polyneuropathy
(Jennifer Kollmer)

Positron Emission Tomography (PET) utilizing Pittsburgh compound B (PIB) detects amyloid heart deposits in hereditary transthyretin amyloidosis (ATTR)
(Björn Pilebro)
Ocular Manifestations of transthyretin-related familial amyloid polyneuropathy
(Antoine Rousseau)

Analysis of Disease Progression in Patients with Transthyretin Cardiac Amyloidosis
(Julian D. Gilmore)

Vasculopathy in transthyretin Val30Met familial amyloid polyneuropathy
(Haruki Koike)

Multi-modality imaging in cardiac ATTR amyloidosis: agreement between echocardiography, MRI and DPD-scintigraphy
(Ludivine Eliahou)

ATTR Amyloidosis: development of cardiac symptoms during 6 years of follow up in different ATTR-variants
(Sebastiaan Klaassen)

Misdiagnoses of transthyretin amyloidosis: a clinical and electrodiagnostic study
(Andrea Cortese)

Neurophysiological pitfalls in TTR-FAP Val30Met
(Isabel Conceição)

17:10 – 17:25
Highlights of the day

17:25 – 17:55
Representatives of patients' associations with amyloidosis

18:00 – 19:00
POSTER SESSION I

19:30
Congress dinner
Tuesday, November 3

8:00-9:00
POSTER SESSION II

8:30 – 9:00
Overall care of ATTR amyloidosis
(L. Obici)

9:00 – 9:30
TTR-FAP : Liver Transplantation vs oral medication, how and when. pros and cons
(T. Coelho, BG. Ericzon)

9:30 – 10:00
Ongoing clinical trials for ATTR amyloidosis
When deciding to enroll a TTR-FAP in clinical trial? (J. Buades)
Overview on clinical trials for ATTR amyloidosis (J. Gillmore)

10:00 – 11:00
Disease modifying therapy
Oral communications:

Familial Amyloid Polyneuropathy treatment with Tafamidis – evaluation of one- and two-year treatment in Porto, Portugal (Teresa Coelho)

Phase 2 Open-Label Extention (OLE) Study of Patisiran, an Investigational siRNA Agent for Familial Amyloidotic Polyneuropathy (FAP) (David Adams)

Diflunisal therapy for cardiac ATTR amyloidosis: a longitudinal, prospective, single centre study (Candida C. Quarta)

Phase 2, Open-Label Extension (OLE) Study of Revusiran, an Investigational RNAi Therapeutic for the Treatment of Patients with Transthyretin Cardiac Amyloidosis (Julian D. Gillmore)

The ISIS-TTRRx-CS2 phase 3 study in patients with familial amyloid polyneuropathy: Baseline results of the first 100 patients for the NIS, NIS+7 and mNIS+7 using different methods of scoring: identification of consistencies and key differences (Rito Bergemann)

Diflunisal in late-onset FAP patients with moderate to severe neuropathy (Laura Obici)
Tuesday, November 3

11:00 – 11:30 Break

11:30 – 11:45
The place of heart transplantation in hereditary ATTR amyloidosis
(S. Varnous)

11:45 - 12:00
The landscape of treatment of chronic kidney disease in hereditary ATTR amyloidosis
(L. Lobato)

12:00 – 12:30 Lunch

12:30 – 13:30 Sponsored Symposia 2 (Alnylam)

13:30 – 14:00
New therapeutic perspectives – amyloid removal
(M. Pepys)

14:00 – 14:30 - Moderator : A. Benachi
Genetic Counseling and assisted medical reproduction
Guidelines for genetic counselling in ATTR amyloidosis (J. Sequeiros)
Preimplantatory diagnosis in TTR-FAP in Portugal (F. Carvalho)

14:30 - 14:45
Which assessment for the carriers ?
The point of view of the neurologist
(I. Conceicao)

14:45 – 15:00
The point of view of the cardiologist
(A. Kristen)

15:00 - 15:30
Oral communications :

Quality of life in ATTR amyloidosis
(Thirusha Lane)

Psychopathological dimensions in familial amyloid polyneuropathy patients
(Alice Lopes)

Transthyretin familial amyloid polyneuropathy impact on health-related quality of life.
(Inês Mónica)
Tuesday, November 3

15:30 – 16:00  Break

16:00 – 16:15  Symptomatic therapy in ATTR amyloidosis
Pain killers in TTR-FAP
(N. Attal)

16:15 – 16:30  Management of digestive disorders
(O. Suhr)

16:30 - 16:45  Therapeutic education program
(M. Theaudin)

16:45 – 17:00  Highlights of the day

17:00  Conclusions and next steps
Poster sessions (Monday)

PL1 Familial amyloidotic polyneuropathy associated with the transthyretin CYS 114 gene in a Russian pair of monozygotic twins. Igor Srokov et al. Moscow, Russia; Utrecht, Netherlands.

PL2 Hereditary transthyretin amyloidosis in Slovenia Janez ZIDAR Ljubljana, Slovenia

PL3 FAP in India: A first genetically proven case. Daniel et al. London, United Kingdom; le Kremlin Bicetre, France

PL4 Epidemiology of transthyretin familial amyloid polyneuropathy in Portugal Mónica Inês et al. Lisboa, Portugal

PL5 Glu89Gln transthyretin-related amyloidosis in Italy and Bulgaria: does geographic area influence phenotype beyond the shared mutation? Christian Gagliardi et al. Bologna, Italy Sofia, Bulgaria

PL6 Val50Ala variant of familial amyloid neuropathy – a rare case in the Czech Republic Tomas Pika et al. Olomouc, Prague, Brno, Czech Republic.

PL7 Transthyretin-related Familial Amyloid Polyneuropathy (TTR-FAP) caused by a very rare, de novo mutation in a Polish patient. Marta Lipowska et al. Warsaw, Poland

PL8 Hereditary ATTR amyloidosis in Hungary: His88Arg and Phe33Leu mutations in three patients Zsuzsanna Arányi et al. Budapest, Hungary

PL9 Teachings from the French database of TTR familial amyloidotic polyneuropathy (TTR-FAP): large genetic and phenotypic heterogeneity, usefulness of TTR gene testing. David Adams et al. Le Kremlin Bicêtre, France

PL10 Clinical, Epidemiological, Genetic, and Electrophysiological Characteristics of Transthyretin Familial Amyloid Polyneuropathies in Israel Menachem Sadeh et al. Holon, Ramat Gan, Jerusalem, Israel

PL11 Epidemiology of Familial Amyloid Polyneuropathy in Bulgaria Stayko Sarafov et al. Sofia, Bulgaria

PL12 Familial Amyloidotic Polyneuropathy in Crete, Greece Minas Tzagournissakis et al. Heraklion, Greece

PL13 The hidden story behind gender differences in familial amyloid polyneuropathy (FAP) ATTRV30M Diana SANTOS et al. Porto, Portugal

PL14 The phenotypical expression of an European inherited TTR amyloidosis in Brazil. Márcia Waddington Cruz et al. Rio de Janeiro, Brazil

PL15 Wild-type transthyretin amyloidosis in female patients Arnt V. Kristen et al. Heidelberg, Germany

PL16 Five novel TTR variants: associated phenotypes and structural consequences. Dorota Rowczenio et al. London, UK

PL17 Genotypic and Phenotypic Presentation of Glu89Gln mutation in Turkey Hacer Durmus et al. Istanbul, Turkey

PL18 Identification of a new variant of TTR involved in familial amyloid cardiomyopathy (FAC) in Brazil: from the patient to the protein Priscila Ferreira et al. Rio de Janeiro, Brazil

PL19 Transthyretin familial amyloid polyneuropathy (TTR-FAP) in Mallorca: A comparison between late- and early-onset disease Manuel Raya-Cruz et al. Palma de Mallorca, Spain

PL20 CLINICAL AND LABORATORY TEST IN PATIENTS WITH FAMILIAL AMYLOID POLYNEUROPATHY: DIFFERENCES BETWEEN SYMPTOMATIC PATIENTS AND ASYMPOMATIC CARRIERS Manuel Raya-Cruz et al. Palma de Mallorca, Spain
**Poster sessions (Monday)**

**PL21** Does the course of Val122Ile differ from SSA, or is selection bias a factor? Hallie Geller et al
Boston, MA, USA

**PL22** Characterization of conformation-specific, human-derived monoclonal antibodies against TTR aggregates with potential for diagnostic and therapeutic use Aubin MichalonSchlieren, Switzerland; Umea Sweden, Porto Portugal.

**PL23** MALDI spectrometry for salivary samples analysis: a new tool for TTR amyloidosis diagnosis. Julie SEGUIER Marseille, France

**PL24** Diagnostic value of fat aspirates for amyloidosis in 950 patients J.A. GILBERTSON et al
London, UK

**PL25** [18F]FDDNP performed better than [18F] Florbetapir to distinguish transthyretin cardiac amyloidosis (TTR-CA) patients from healthy controls: An ex vivo study Anne-Claire Dupont. Tours, Paris France

**PL26** Disphosphonates cardiac uptake in familial amyloid neuropathy: Comparison between DPD and HMDP Hamza Regaieg et al Paris, Clamart, France

**PL27** Comparison of MIBG and Diphosphonate scintigraphy in cardiac involvement of aTTR-FAP Renata Chequer (1), et al. Paris, Clamart, France

**PL28** Cardiac extracellular volume quantified with T1 mapping techniques reflects degree of cardiac and neurological involvement in Hereditary Transthyretin Amyloidosis Esther González-López et al. Majadahonda, Huelva Palma de Mallorca, Madrid Spain

**PL29** Multi-modality imaging in cardiac ATTR amyloidosis: agreement between echocardiography, MRI and DPD-scintigraphy Ludvine Elihou et al. et al.Clamart, Paris, France

**PL30** Quantitative comparison between amyloid deposition detected by 99mTc-diphosphonate imaging and myocardial deformation evaluated by strain echocardiography in transthyretin related cardiac amyloidosis. Gianluca Di Bella et al. Messina, Italy

**PL31** Posterior longitudinal strain by speckle tracking echocardiography, marker of cardiac amyloidosis? Julian TERNACLE et al. Créteil, France

**PL32** Usefulness of 99mTc-HMDP scintigraphy for the etiologic diagnosis and prognosis of cardiac amyloidosis Arnault Galat et al. Créteil, France

**PL33** MR-Neurography of the sural nerve in patients with hereditary amyloidosis: Jennifer Kollmer et al Heidelberg, Germany

**PL34** Axon reflex-mediated vasodilation is reduced in proportion to disease severity in familial amyloid polyneuropathy Thierry Kuntzer. Lausanne, Switzerland

**PL35** The diagnostic accuracy of Sudoscan in TTR-FAP Jose Castro et al. Lisboa, Portugal

**PL36** Coexistence of degenerative aortic stenosis and wild type transthyretin-related cardiac amyloidosis: a potentially dangerous association that can be non-invasively identified Simone Longhi et al. Bologna, Italy

**PL37** Spotting senile systemic amyloidosis: why we miss it. Marianna FONTANA et al. London, UK

**PL38** Ocular Manifestations of transthyretin-related familial amyloid polyneuropathy Antoine Rousseau et al. Le Kremlin-Bicêtre, France

**PL39** Red-flag symptom clusters in transthyretin familial amyloid polyneuropathy (TTR-FAP) Isabel Conceicao et al. Lisbon Portugal
Poster sessions (Tuesday)

PM1  Analysis of Disease Progression in Patients with Transthyretin Cardiac Amyloidosis Julian D. GILMORE et al. London, UK, New York, Cambridge, USA

PM2  ATTR Amyloidosis: development of cardiac symptoms during 6 years of follow up in different ATTR-variants Sebastiaan Klaassen et al. Groningen, The Netherlands

PM3  Mass spectrometry analysis of transthyretin (TTR) post-translational modifications (PTMs) in hereditary ATTR: a case-control Spanish experience. Marta Vilà-Rico et al. Barcelona Biscay, Spain

PM4  Cardiac involvement and clinical follow up of patients with hereditary transthyretin related amyloidosis associated with Glu89Gln mutation Mariana GOSPODINOVA et al.Sofia, Bulgaria

PM5  Retinal and choroidal vascular abnormalities in TTR-FAP Antoine Rousseau et al. Le Kremlin-Bicêtre, France

PM6  Patient experience with hereditary and senile systemic amyloidoses: a survey from the Amyloidosis Research Consortium Isabelle Lousada et al. Lincoln Boston New York South San Francisco, United States Pavia, Italy

PM7  Patients with Hereditary ATTR Amyloidosis Experience an Increasing Burden of Illness as the Disease Progresses David Adams et al. Le Kremlin-Bicêtre, France Cambridge, USA Porto, Portugal

PM8  Cardiomyopathy and peripheral polyneuropathy severity in patients with Glu89Gln mutation at the time of diagnosis Mariana Gospodinova et al. Sofia, Bulgaria

PM9  Specific ophthalmologic changes in late onset familial amyloid polyneuropathy (FAP) Portuguese patients Natalia Ferreira et al. Porto Portugal

PM10  Specific ocular changes in TTR MET30-FAP after liver transplantation Natalia Ferreira et al. Porto, Portugal

PM11  What to do when the neuropathy worsens after successful heart and liver transplantation in a Glu89Lys Transthyretin Amyloidosis? Thierry Kuntzer et Francois OchsnerLausanne, Switzerland

PM12  Patient with transthyretin amyloidosis due to domino liver transplantation : 2 years follow up under Tafamidis treatment Arman Cakar et al. Istanbul, Turkey

PM13  Preliminary Assessment of Neuropathy Progression in Patients with Hereditary ATTR Amyloidosis after Orthotopic Liver Transplantation (OLT) David Adams et al. et al. LE KREMLIN-BICETRE, France Palma de Mallorca, Spain, Umea, Sweden Pavia, Italy Porto, Portugal

PM14  Treatment of transthyretin (TTR) amyloid cardiomyopathy with an antisense oligonucleotide inhibitor of TTR synthesis Merrill D. Benson et al. Indianapolis, USA Carlsbad, USA

PM15  A phase 3 clinical trial with ISIS-TTRRx, a 2nd-generation antisense oligonucleotide targeting transthyretin (TTR), for the treatment of TTR amyloid cardiomyopathy Helen Milns et al. Hertfordshire, Middlesex, United Kingdom Carlsbad, United States Philadelphia, United States

PM16  Positive real-world effectiveness of tafamidis for delaying disease progression in transthyretin familial amyloid polyneuropathy Michelle Stewart et al. Groton, New York, San Francisco, USA

PM17  Tafamidis reduces disease progression in patients with transthyretin familial amyloid polyneuropathy: supportive post-hoc analyses of a pivotal trial Denis Keohane et al. New York, Groton, Burlington, Collegeville, United States
Poster sessions (Tuesday)

PM18  Monitoring safety and effectiveness of Tafamidis in transthyretin amyloidosis in Italy: a 3-year longitudinal multicenter study in a non-endemic area  Andrea Cortese et al.  Pavia, Messina, Roma, Verona, Napoli, Milano, Italy

PM19  Early intervention with tafamidis provides long-term benefit in delaying neurological progression in patients with transthyretin familial amyloid polyneuropathy  Marcia Waddington-Cruz et al. Rio de Janeiro, Brazil  New York, Groton, Burlington, USA

PM20  Management of stage 1 TTR FAP: French experience  David Adams et al. Le Kremlin Bicêtre, France

PM21  Diflunisal compassionate use in transthyretin familial amyloidotic polyneuropathy (TTR-FAP): report of the first Spanish experience  Sebastián E Azorín Contesse et al. Barcelona, Spain

PM22  Tafamidis in familial amyloidosis – monitoring the ocular manifestations  Ines Casal et al. Porto, Portugal

PM23  Tissue remodeling after RNAi-mediated knockdown of TTR in a Familial Amyloidotic Polyneuropathy mouse model  Nádia Pereira Gonçalves et al. Porto, Oeiras, Portugal

PM24  Neuroprotection of Anakinra on peripheral nerve neurodegeneration in single and combination protocols with TTR siRNA in a transgenic mouse model for human V30M transthyretin  Maria João Saraiva et al. Porto, Portugal

PM25  SOM0226, a repositioned compound for the treatment of TTR amyloidosis  Núria Reig et al. Barcelona, Spain

PM26  The role of complement in ATTR amyloidosis: a new therapeutic avenue?  Elena Panayiotou et al. Nicosia, Cyprus

PM27  Novel conformation-specific monoclonal antibodies against amyloidogenic forms of transthyretin  Jeffrey N. Higaki et al. South San Francisco, United States  Toronto, Canada

PM28  TUDCA as an autophagic modulator of ATTR V30M Amyloidosis  Cristina Teixeira et, Maria João Saraiva. Porto, Portugal

PM29  Comparison and identification of early clinical, biological and echocardiographic prognostic markers in cardiac amyloidosis  Thibaud Damy et al. Créteil, Limoges, France

PM30  Neurophysiological pitfalls in TTR-FAP Val30Met  Isabel Conceição et al. Lisbon, Portugal

PM31  Prevalence, risk factors and correlation with cardiac involvement of carpal tunnel syndrome in amyloidosis  Agnese Milandri et al. Bologna, Italy

PM32  Usefulness of Combining Electrocardiogram and Echocardiography Findings and Brain Natriuretic Peptide in Early Detection of Cardiac Amyloidosis in Subjects with Transthyretin Gene Mutation  Gianluca Di Bella et al. Messina, Italy

PM33  TTR sequencing should be considered ahead of hypertrophic cardiomyopathy in Afro-Americans  Jean Herlé Raphaelen et al. Paris, France

PM34  Misdiagnoses of transthyretin amyloidosis: a clinical and electrodiagnostic study  Andrea CORTESE et al. Pavia, Italy

PM35  Neurophysiological pitfalls in TTR-FAP Val30Met  Isabel Conceição et al. Lisbon, Portugal

PM36  Vasculopathy in transthyretin Val30Met familial amyloid polyneuropathy  Haruki Koike et al. Nagoya, Japan
POSTER SESSIONS (TUESDAY)

PM37 Delayed small bowel octreotide response in patients with hereditary transthyretin amyloidosis Jonas Wixner et al. Umeå, Stockholm, Sweden

PM38 Is 99mTc-diphosphonate uptake the earliest sign of cardiac amyloidosis development in asymptomatic Glu89Gln transthyretin gene mutation carriers? Fabio Minutoli et al. Messina, Italy

PM39 The rehabilitation in the management of Transthyretin Familial Amyloid Polyneuropathy Agnès Morier et al Le Kremlin Bicêtre, France

PM40 LIFE PATHS OF FAMILIAL AMILOIDOTIC POLINEUROPATHY PATIENTS: a descriptive study Alice Lopes et al. Porto, Portugal

PM41 PARENTERAL NUTRITION IMPROVES NUTRITIONAL STATUS, AUTONOMIC SYMPTOMS AND QOL IN PATIENTS WITH TTR-FAP Massimo Russo et al. Messina, Palermo, Italy

PM42 B3461028: A MULTICENTER, INTERNATIONAL, PHASE 3, DOUBLE-BLIND, PLACEBO-CONTROLLED, RANDOMIZED STUDY TO EVALUATE THE EFFICACY, SAFETY AND TOLERABILITY OF DAILY ORAL DOSING OF TAFAMIDIS MEGLUMINE (PF-06291826) 20 MG OR 80 MG IN COMPARISON TO PLACEBO IN SUBJECTS DIAGNOSED WITH TRANSTHYRETIN CARDIOMYOPATHY (TTR-CM). Alison Flynn, Douglas Girgenti, MD, Marla Sultan, MD, MBA, Carolyn Hahn, Robert Moller, PhD, Jennifer Schumacher, PhD, Rodger Kobes, MD, PhD (for the Study Team) Pfizer Inc. Global Innovative Pharmaceutical Business, Collegeville/Groton/New York, USA.