PROGRAMME

1 June Monday

08.30 Opening address Beckman Lars President, Umeå University
Jack Lundström Chief Physician, Skellefteå Hospital
Allan Boström Kommunträd, Skellefteå
Lars Steen President of the Meeting

09.00-9.30 Honorary lecture
Contribution by Japanese scientists to FAP study. Shukuro Araki

09.30-10.00 Plenary lecture 1
A transcription cascade of molecular events regulate the expression of transthyretin and other genes expressed mainly in rodent hepatocytes. Kleanthis Xantopoulous

10.00-10.20 Coffee break

10.20-10.50 Plenary lecture 2
Evolution and ontogeny of transthyretin B. Southwell

10.50-11.50 Session 1 Molecular biology of FAP
Chairman K. Xantopoulous
Co-chairman C. Thylen

0.1 Developmental expression of transthyretin mRNA J. Herbert

0.2 Cultured rat retinal pigment epithelium secretes transthyretin J. Herbert

0.3 The mammalian pineal is a functional site for the retinolbinding protein R.L. Martone

0.4 Genetically modified cells producing human TTR (TTR Met 30): A potential tool for the study of amyloidogenesis M.J. Bonifacio

11.50-12.50 Lunch

12.50-14.35 Poster session 1 (3 min presentation, 2 min discussion)
Chairmen J. Hamilton/
D. Jacobson/F. Salvi
12.50-13.30  The TTR molecule
Chairman J. Hamilton

Screening of transthyretin associated with amyloid neuropathy with FD6 monoclonal antibody
(P.M.P. Costa).P1

Screening of TTR variants in the Portuguese population by HIEF
(I.L. Alves).P2

Hydrophilicity and circular dichroism analyses of FAP TTR
(J. Skare).P3

Secondary structure alteration of pure variant transthyretins (Met 30)
associated with type 1 Familial amyloidotic polyneuropathy from homozygote
gene carriers
(S. Ikegawa).P4

Interactions of transthyretin with metals --1:TTR binds divalent cations
which influence its conformation and solubility.
(R.L. Martone).P5

Two transthyretin mutations associated with euthyroid hyperthyroxinemia.
(S. Izumoto).P6

Transthyretin synthesis in primary choroid plexus neoplasms.
(J. Herbert).P7

Preliminary conformational studies of lle-122 mutant transthyretin.
(A.M. Damas).P8

13.30-14.15  New TTR mutations
Chairman D. Jacobson

New amino acid mutations (Ala30, Asn84, Leu33) at previously established
FAP-associated TTR sites.
(L.A. Jones).P9

Transthyretin Pro 36 associated with familial amyloidotic polyneuropathy in
an Ashkenazic Jewish kindred.
(D.R. Jacobson).P10

A novel transthyretin (TTR) mutation (Arg-47) associated with familial
amyloidotic polyneuropathy FAP.
(T. Murakami).P11

Gly47Ala: A new transthyretin gene mutation in hereditary amyloidosis TTR-
related.
(A. Ferlini).P12
New TTR mutations cont.

Transthyretin Pro 55, a variant associated with early-onset, aggressive, diffuse amyloidosis including neurologic, cardiac, and gastrointestinal involvement.
(D.R. Jacobson). P13

Familial amyloidotic polyneuropathy presenting with carpal tunnel syndrome and a new transthyretin mutation: Asn 70.
(S. Izumo). P14

A new transthyretin variant (His-69) associated with vitreous amyloid in an FAP family.
(M. Skinner). P15

A new transthyretin variant (Ala-71) and FAP in a French family.
(M.D. Benson). P16

A new transthyretin (TTR) variant (TTR Gln 89) in a Sicilian kindred
hereditary amyloidosis.
(M.R. Almeida). P17

14.15-14.45 TTR mutations and clinical symptoms, prevalence
Chairman F. Salvi

FAP type I in black patients - report on two Brazilian families.
(M. Waddington Cruz). P18

Molecular diagnosis of the transthyretin Met111 mutation associated to familial amyloid cardiomyopathy in Denmark.
(B-Y. Nordvåg). P19

Asymptomatic carriers in the Majorcan focus of FAP I.
(M. Munar-Qués). P20

A prevalence study of FAP in two areas of Portugal: Povoa/Vila do Conde and Unhais-da-Serra/Seia/Covilhã.
(A. Sousa). P21

A prevalence study of FAP in Northern Sweden.
(A. Sousa). P22

Some patterns of age-of-onset variation in Swedish FAP families.
(U. Drugge). P23
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| 14.45-15.15 | **Plenary lecture 3**  
Analogies with transmissible dementias (CJD, GSS) suggest that transthyretin amyloidoses of FAP and other amyloidoses may also be transmissible and infectious.  
D. C. Gajdusek |
| 15.15-15.55 | **Session 2 The TTR molecule**  
Chairman T. Pettersson  
Co-chairman E. Lundgren |
| 05     | Crystal structure of recombinant Met30 transthyretin suggests a molecular basis for amyloidogenesis in FAP. (20 min.)  
C.J. Terry |
| 06     | Comparison of the high-resolution x-ray crystal structures of normal transthyretin and two variants, Met30 and Thr109.  
J.A. Hamilton |
| 15.55-16.15 | Coffee break |
J. Murrell |
| 08     | Transthyretin interacts with globin to form protein complexes with hemedependent solubility.  
R.L. Martone |
| 16.45-17.15 | **Plenary lecture 4**  
Variant proteins and hereditary amyloidosis.  
M.D. Benson |
| 17.15-18.00 | A new mutant transthyretin (Arg10) associated with familial amyloid polyneuropathy.  
T. Uemichi |
| 09     | A compound heterozygotic individual for two transthyretin variants presenting increased thyroxine binding capacity.  
I.L. Alves |
| 10     | A transthyretinicemic mice in studies of TTR, contribution to thyroxine homostasis.  
M.J.M. Saraiva |
2 June Tuesday

08.30-09.00  Plenary lecture 5  
Non-pathogenic TTR variants in the molecular pathology of the TTR related disorders. M.J.M. Saraiva

09.00-10.00  Session 3 TTR mutations and clinical syndromes  
Chairman P. Costa  
Co-chairman G. Holmgren

012  Familial amyloidosis at the Boston University amyloid research center. M. Skinner

013  Haplotype analysis of the TTR gene in blacks with and without the TTR Ile 122 gene. D.R. Jacobson

014  A review of 107 cases in Portuguese FAP population in whom both parents were disease-free. T. Coelho

015  Homozygous transthyretin His58 associated with unusually aggressive familial amyloidotic polyneuropathy. D.R. Jacobson

10.00-10.20  Coffee break

10.20-11.30

016  A double mutant TTR allele (Ser 6, Ile33) in an Israeli kindred ("SKO") with familial amyloid polyneuropathy. D.R. Jacobson

017  Molecular detection of transthyretin Met111 affection among members of a Danish family with familial amyloid cardiomyopathy, using DNA from paraffin embedded tissues. B-Y. Nordvåg

018  Hereditary amyloidosis transthyretin related in Italy. A. Forlini

019  A strikingly benign evolution of FAP in an individual found to be a compound heterozygote for two TTR mutations: TTR Met50 and TTR Met119. T. Coelho

020  The frequency of TTR Met30 carriers in northern Sweden studied with the FD6 monoclonal antibody. G. Holmgren

11.30-12.30  Lunch
12.30-15.00 Poster session II
Chairmen: S. Araki, M.D. Benson, A. Cohen, S. Kiuru

12.30-13.00 Non-TTR associated FAP
Chairman: S. Kiuru

Apolipoprotein A1 (APOAI) mutation Arg26 causing hereditary non-neuropathic autosomal dominant systemic amyloidosis.
(D.M. Vigushin) P24

Role of apolipoprotein AII in patients with familial amyloidotic polyneuropathy (FAP): analysis of plasma lipid particles and apolipoprotein AII immunoreactivity for amyloid deposit.
(Y. Tanaka) P25

Homozygosity for Finnish type familial amyloidosis is associated with severe nephropathy.
(C.P.J. Maury) P26

Familial amyloidotic polyneuropathy type IV: gene analysis of the first Japanese family.
(Y. Sunada) P27

Clinical finding in 30 patients with familial amyloidosis of the Finnish type (FAF).
(S. Kiuru) P28

All affected individuals with familial amyloidosis of the Finnish type carry G 654-A mutation of Gelsolin.
(T. Paunio) P29

13.00-13.25 TTR-amyloid fibril protein
Chairman: M.D. Benson

Senile (Apo-AII) and secondary (AA) amyloidosis in mice during chronic toxicity studies.
(M. Meijers) P30

Subcutaneous adipose tissue for purification of ATTR.
(B. Johansson) P31

Purification and characterization of ATTR from the myocardium of a deceased Danish patient with familial amyloid cardiomyopathy.
(L.F. Hermansen) P32

Hereditary amyloidosis transthyretin (TTR) related to the vitreous body: Clinical and molecular characterization in two Italian families.
(F. Salvii) P33

Amyloid related proteins in the plasma of patients with familial amyloidotic polyneuropathy.
(Y. Ando) P34
13.25-15.00 Clinical aspects: symptoms, assays, treatment
Chairmen S. Araki and A. Cohen

Pupillary analysis in familial amyloidotic polyneuropathy hereditary non-
(FAP) type I.
(Y. Ando) P35

Ocular manifestations in FAP.
(O. Sandgren) P36

Initial motor dominant polyneuropathy in patients with familial
polyneuropathy (FAP) type I.
(Y. Ando) P37

Reduced number of gastrointestinal endocrine cells in familial amyloidotic
polyneuropathy - a preliminary report.
(M. El-Sahly) P38

Autonomic disorders in the Majorcan focus of familial amyloidotic
polyneuropathy type I: a study by standards tests and power spectral analysis
of heart rate.
(J.F. Forteza) P39

Echocardiographic and doppler features of familial amyloidotic
polyneuropathy type I in the Majorcan focus.
(J.F. Forteza) P40

Myocardial involvement and ventricular function in the Majorcan focus of
familial amyloidotic polyneuropathy type I: a study by myocardial
scintigraphy and radionuclide angiography.
(J.F. Forteza) P41

Familial amyloidosis (AF): a study of 52 north American-born patients seen
during a 30-year period.
(R.A. Kyle) P42

Comparative assessment of autonomic function by spectral analysis of heart
rate, Valsalva manoeuvre, standing deep breathing and head-up tilting.
(M. Carvalho) P43

Early carpal tunnel syndrome in FAP Met30.
(T. Coelho) P44.

Study of skin and sural nerve biopsies in FAP Met 30 asymptomatic carriers.
(T. Coelho) P45

Clinical study of nephropathy in TTR Met 30 familial amyloidotic
polyneuropathy.
(L. Lobato) P46

Late-onset FAP with the TTR Met 30 mutation in France.
(G. Grateau) P47

Impact of the presymptomatic test on the reproductive behaviour of
Portuguese young adults at risk for FAP: a preliminary study.
(T. Coelho) P48
Clinical aspects. cont.

Familial amyloidotic polynuropathy: electromyographic studies in 8 Italian families.
(P. Montagna) P49

(L. Wikström) P50

Pacemaker implantation in patients with familial amyloidotic polyneuropathy.
(L. Maciel) P51

Treatment of diarrhea in familial amyloidotic polyneuropathy (FAP) with octreotide.
(M.L. Sales Luís) P52

Plasma catecholamines in familial amyloidotic polyneuropathy of the Portuguese type.
(I. Ducla Soares) P53

15.00-15.30 
Plenary lecture 6
Molecular biology of gelsolin-related amyloidosis. C.P. Maury

15.30-15.50 
Coffee break

15.50-16.50 
Session 4 Non TTR associated FAP
Chairman C.P. Maury
Co-chairman O. Sandgren

021 Insertions, deletions, and polymorphisms in transmissible amyloidotic spongiform encephalopathy. L.G. Goldfarb

022 Familial spongiform encephalopathy and familial amyloidotic polyneuropathy: kindred diseases in different kindreds. P. Brown

023 In vitro creation of amyloid fibrils from mutant Asn187 C.P.J. Maury Gelsolin peptides.

024 Apolipoprotein A1 mutation Arg60 causes hereditary autosomal dominant non-neuropathic systemic amyloidosis. M.B. Pepys

16.50-17.20 
Plenary lecture 7
Age-associated FAP/SSA. P. Westermark

17.20-17.50 
Session 5 Age associated FAP/amyloidosis.
Chairman P. Westermark

025 The recognition of senile cardiac amyloidosis antemortem. R.A. Kyle

026 Antigenic mapping of TTR-derived amyloid and ATTR. Å. Gustavsson
3 June Wednesday

08.00-08.30 Plenary lecture 8
TTR amyloid fibril protein
M. Skinner

08.30-10.00 Session 6 TTR amyloid fibril protein.
Chairman M. Saraiva
Co-chairman P. Westermark

027 Is fibril amyloid-enhancing factor in experimental
AA Amyloidosis a scrapie-like nucleating infectious
protein?
D.C. Gajdusek

028 Partial denaturation of transthyretin is sufficient
for amyloid fibril formation in vitro.(30 min)
J.W. Kelly

029 Myelin P2 protein is specifically associated with
vitreous amyloid from patients with familial
amyloidotic polyneuropathy (TTR Ser84).
R.L. Martone

030 Interaction of transthyretin with metals--II:
Metal chelation solubilizes transthyretin amyloid.
J. Herbert

031 Transthyretin amyloid fibril formation - production
of mutated and truncated transthyretin variants for
model studies.
C. Thylén

10.00-10.20 Coffee break

10.20-10.50 Plenary lecture 9
Diagnostic imaging and quantifications using radio-
labelled serum amyloid P-component.
M. Fepys

10.50-11.50 Session 7 FAP-clinical aspects
Chairman R. Kyle
Co-chairman O. Suhr

032 Scintigraphy with 123 I-serum amyloid P component
in familial amyloid polyneuropathy.
P.N. Hawkins

033 Changes in bone metabolism and serum Ca levels in
patients with familial amyloidotic polyneuropathy (FAP).
Y. Tanaka

034 Clinical finding in six TTR mutation of Italian familial
amyloidosis (TTR Val 30 Met, TTR Ala 36 Pro, TTR Gly
47 Ala, TTR Thr 49 Ala, TTR Phe 64 Leu).
F. Salvi

035 Cardiovascular autonomic dysfunction in primary (AL)
and five subtypes of familial (ATTR) amyloidosis.
S. Ikekawa

11.50-12.50 Lunch
12.50-13.20  **Plenary lecture 10**  
Liver transplantation-state of the art lecture.  
B.G. Ericzon

13.20-14.00  **Session 8 FAP-trends in treatment**  
Chairman M. Pepys  
Co-chairman L. Steen

036  Secondary hypoplastic anemia in familial amyloidotic polyneuropathy (FAP) and trial of erythropoictin for treatment.  
K. Asahara

037  Stationary course during sixteen months after liver transplantation in familial amyloidotic polyneuropathy.  
O. Andersen

038  Familial amyloidotic polyneuropathy type I. First liver transplantation in Spain.  
F. Lopez-Andreu

039  Liver transplantation in four Swedish FAP met30 patients.  
G. Holmgren

14.00-14.30  General discussion.

14.30-15.00  Closing remarks.