

A large, semi-transparent watermark of the Rotary logo is centered in the background. It features a yellow gear with a blue band across the top containing the word "ROTARY" in white capital letters. The gear has a central hub and several spokes.

# The Rotary Fellowship on Molecular Epidemiology of Parkinson's disease

Results and Perspectives after 3 year

**Maurizio Facheris, MD, MSc**

Institute of Genetic Medicine, EURAC Bolzano

Department of Neurology, Central Hospital Bolzano

2001

- Residency in Neurology (Milano)
- Interest in Degenerative (Alzheimer's) and Movement Disorders (Parkinson's)



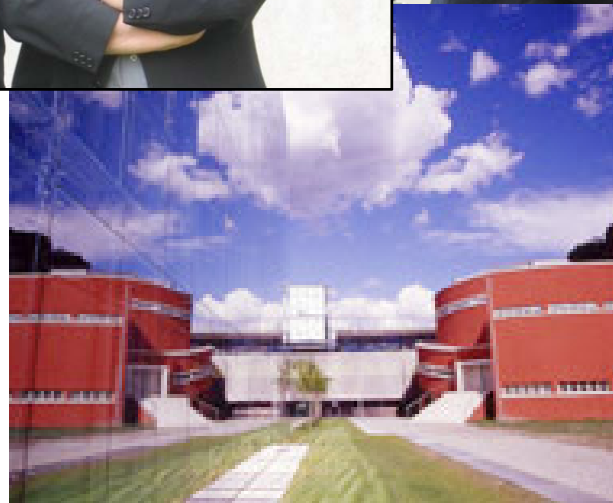
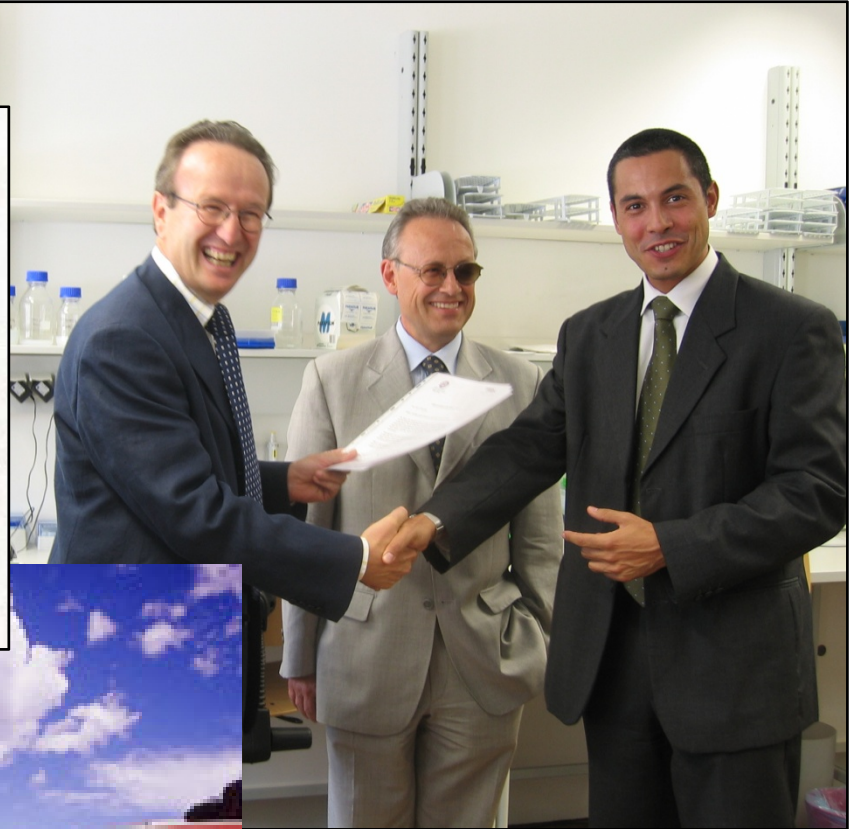
2004

- First year fellowship Mayo Clinic (USA) – genetics of PD
- Contact with Dr. Peter Pramstaller – raising of the EURIGEM



2005

- Rotary-Brixen Fellowship → EURIGEM → Dr. Facheris  
€30,000, 1.5 years in USA, upon return: 3-year requirement in South Tyrol



2007

- Graduation (Master in Clinical Research)
- Beginning of work at EURIGEM and PD Outpatient Clinic – Hospital Bolzano



# “Quick” Timeline

2001

- Residency in Neurology (Milano)
- Interest in Degenerative (Alzheimer’s) and Movement Disorders (Parkinson’s)

2004

- First year fellowship Mayo Clinic (USA) – genetics of PD
- Contact with Dr. Peter Pramstaller – raising of the EURIGEM

2005

- Rotary-Brixen Fellowship → EURIGEM → Dr. Facheris  
€30,000, 1.5 years in USA, upon return: 3-year requirement in South Tyrol

2007

- Graduation (Master in Clinical Research)
- Beginning of work at EURIGEM and PD Outpatient Clinic – Hospital Bolzano

2010

- Completion of the 3-year requirement
- Results and perspectives

# At the Mayo Clinic

- Research: Master in Clinical Research
  - Epidemiology
  - Genetics
  - Statistics
  - Study design
- Clinics: gaining experience in the management of Parkinson's disease and other movement disorders

# Back in South Tyrol

- Research
  - Uric acid and Parkinson's disease
  - Cigarette smoking and Parkinson's disease
- Clinics
  - Helping Dr. Pramstaller / Prof. Schönhuber in finalizing a vision for the PD outpatient clinic
  - GESSPARK
- Epidemiology
  - The CHRIS study

# Uric acid and PD

- Uric acid causes gout, but it is a potent natural antioxidant!
- More than 30 publications which report that high urate concentrations might protect from PD
- Discovered a gene (SLC2A9) related to uric acid metabolism: its polymorphism lower uric acid concentration in blood
- We found that those polymorphisms within SLC2A9 lower the age at onset of PD (maybe independently from the antioxidant activity!)

# Uric acid and PD

- Poster at the 61<sup>o</sup> annual meeting of the American Academy of Neurology, Seattle (USA) April 2009
- Highlighted among the best research proposals of the conference



- Paper submitted – awaiting response

# Smoke and PD

- Epidemiological studies have consistently reported that cigarette smokers have a reduced risk of Parkinson disease. However, some smokers still develop PD
- *CYP2A6* gene responsible for more than 90% of nicotine biotransformation to cotinine
- To study the main and joint effects of cigarette smoking and *CYP2A6* gene variants on the risk of PD

# Smoke and PD

- We found that individuals who smoke are more “protected” from PD if they have the *CYP2A6\*1B* variant
- *CYP2A6\*1B* increases the metabolism of nicotine, increasing levels of cotinine
- Cotinine rather than nicotine may be neuroprotective in PD
- Clinical trials of nicotine in PD should consider pharmacogenetic effects (personalized medicine)

# Smoke and PD

- Selected for platform presentation at the 62<sup>o</sup> annual meeting of the American Academy of Neurology, Toronto (Canada), April 2010
- Selected for highlights as ranking top 5% of the best research contributions



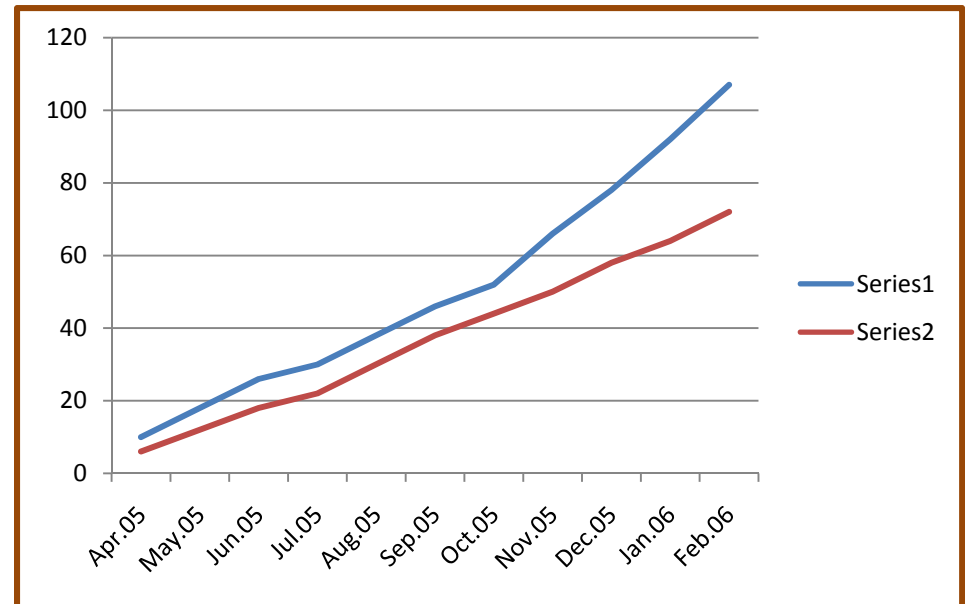
- Paper submitted – awaiting response

# Parkinsonambulanz

- Almost 1500 patients
  - ca. 15 new patients/month
  - ca. 120 controls/month
- 3 doctors (Pramstaller, Facheris, Zanigni)
- 3 nurses from the poliambulatori
- Aiming to a better quality (research, QoL, new expertise – e.g. dysautonomic functions)

# GESSPARK

- Genetic Epidemiological South-tyrolean Study on PARKinson disease
- Questionnaires on cognitive status, depression, non-motor symptoms, QoL...
- Blood sample

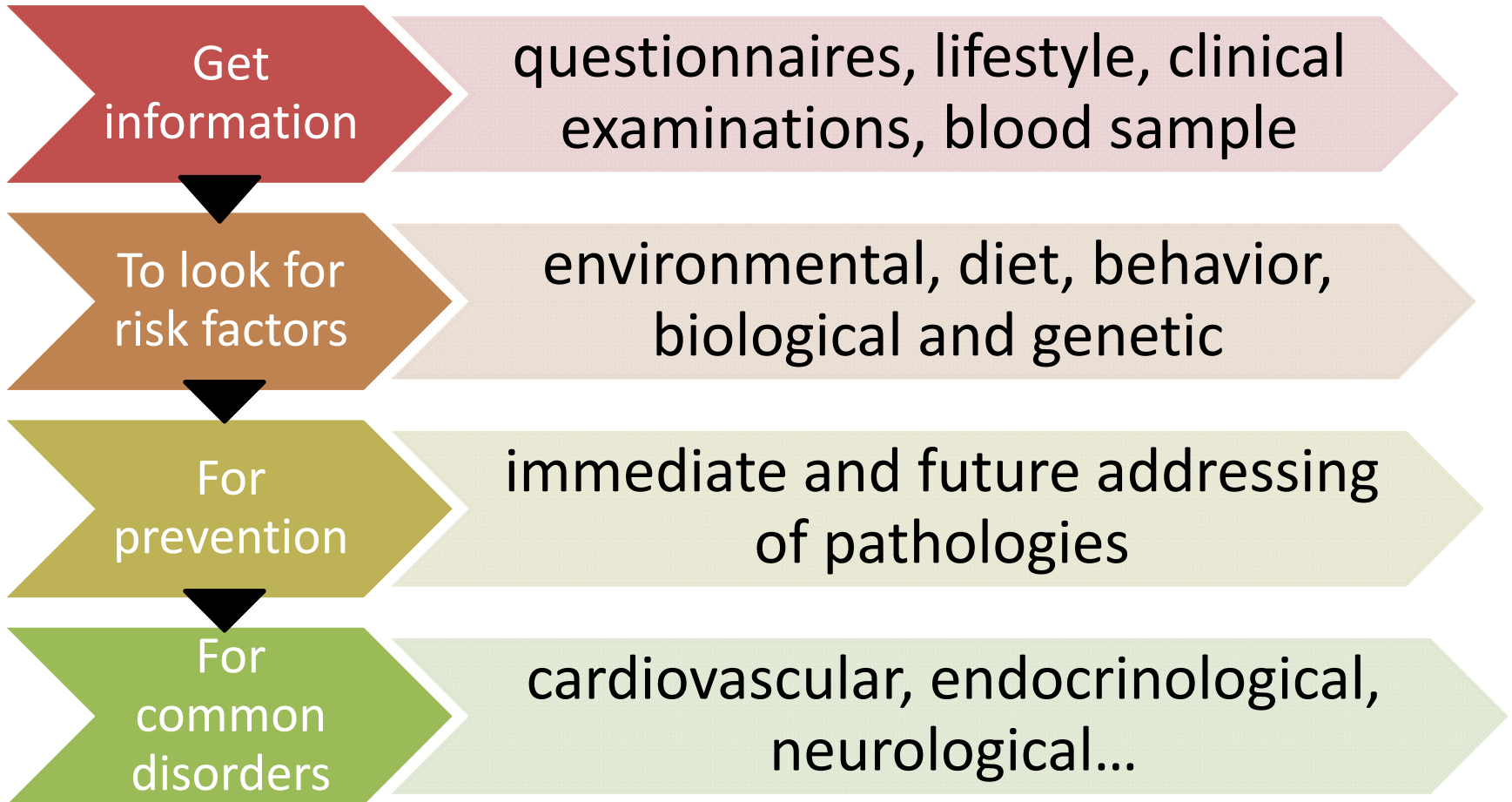


# The CHRIS project



- Cooperative Health Research In South-Tyrol
- Population-based epidemiological study
- Area of start: Vinschgau; Schlanders' Hospital
- Closed cohort. Age >18 (comparable to Helmholtz - Germany)
- Full assessment (whole residents). When response rate of 60% = 14,000 individuals.
- Starting date: 2010 for three years

# The CHRIS project



# Conclusions

- Thank you!
- I am not going anywhere else...

