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Webpage, information and involvement of SMS and patients organizations

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Sofia City Hotel, Sofia, Bulgaria

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cost
EUROPEAN COOPERATION IN SCIENCE AND TECHNOLOGY

Weakness/Comments:

- CVs of all members of the Euromene Cost Action;
- News or information regarding events/initiative on CFS/ME in each EU country;
- Use of the social media (Facebook, Twetter) for sharing our activites;
- Involvement of the patients associations for the organization of events/meeting with Euromene experts/scientific members

SMS involvment

Magi Lab group srl, located in Rovereto in Northen-Est of Italy interested in rare disorders and with experties in genetic diagnostic/research with labs applying advanced technologies (i.e. Nextgen) (http://www.magi-group.eu)

MAGI GROUP

A group of 30 health professionals (medical doctors, biologists, researchers) active since 2006 on rare and genetic diseases.

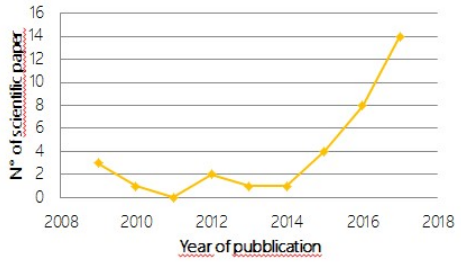
AIM to achieve a molecular diagnosis for patients affected by rare and genetic diseases.

http://www.magi-group.eu/

What does MAGI do?

Diagnostic

Research



12 published papers in 2017

Formation of medical/biological students and Divulgative speech

Support to developing countries

Myalgic Encephalomyelitis/Chronic Fatigue Syndrome

- The disease mechanisms are unknown.
- Several lines of evidence support the possibility that ME/CFS development may involve a **heritable component**, although environmental factors, viral illnesses, stressful life events or traumas may also be implicated .
- It has been reported that increased risks of ME/CFS among first degree relatives (Albright *et al.* 2011*)*
- Studies on twins suggest that there is a higher rate of fatigue concordance in monozygotic twins when compared with dizygotic twins (Buchwald *et al.* 2001)
- Genes associated are not known yet.
- Symptoms such as myalgia and exercise intolerance could be associated with dysfunctions in genes involved in metabolic pathways (myoadenylate deaminase (AMPD1), carnitine palmitoyltransferase II (CPT2), and muscle glycogen phosphorylase (PYGM)).

Genetic evaluation of AMPD1, CPT2, and PGYM metabolic enzymes in patients with chronic fatigue syndrome

P.E. Maltese[1], L. Venturini[2], E. Poplavskaya[2], M. Bertelli[1], S. Cecchin[1], M. Granato[1], S.Y. Nikulina[3], A. Salmina[3], N. Aksyutina[3], E. Capelli[4], G. Ricevuti[2] and L. Lorusso[5]

MAGI GROUP involvement in EUROMENE - European Network on Myalgic Encephalomyelitis/Chronic Fatigue Syndrome

Genetic testing

- Exclude differential diagnosis of ME/CFS by analysing gene associated to disease with overlapping phenotype.
- Perform Whole exome sequencing on family with familial form of ME/CSF with pediatric onset negative to variant in genes associated to diseases with overlapping phenotype.