EUROGLYCANET

Project No. LSHM-CT2005-512131

Congenital Disorders of Glycosylation:
a European network for the advancement
of research, diagnosis and treatment of a
growing group of rare disorders.

Integrating and strengthening the European Research Area

Priority 1:	Life sciences, genomics and biotechnology for health
Identifier:	FP6-2003-Lifescihealth-I
Instrument:	**Coordination Action**

- Annex F1 -
PUBLISHABLE FINAL ACTIVITY REPORT

Coordinator	Prof. Gert Matthijs
Institute	Katholieke Universiteit Leuven
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Revision:	

ftp://ftp.cordis.europa.eu/pub/lifescihealth/docs/project_rep_guid_lifesci.pdf

1. Project execution

The Congenital Disorders of Glycosylation (CDG) are a group of inborn errors of metabolism. Most of these genetic defects result in severe disease, mental retardation and physical handicap. The main objective of EUROGLYCANET was to promote (early) diagnosis by offering the diagnostic tools for screening as well as for expert analysis and by raising awareness. At the same time, EUROGLYCANET has tried to integrate research activities in the field, and work towards the development of therapies for CDG and related disorders.

The network has installed a 'carousel system' to organize and follow a sample flow through the different laboratories. This included the establishment of the database, accessible through Internet, and the provision of tools for (early) and expert diagnosis. Also, to raise awareness about this group of rare diseases, EUROGLYCANET provided information to the public and to physicians and other professionals, and training by offering courses to expert clinicians and researchers in the field. The network organized 4 successful annual meetings, for clinical and basic scientists.

Importantly, EUROGLYCANET has installed referral centres in many different European countries. These are important for the recruitment of samples and the assistance to local physicians and families, faced with the diagnosis of these diseases.

The different laboratories involved in the diagnostic carousel have been able to introduce novel methods, like the isoelectric focussing of ApoCIII for the identification of O-glycan defects, and new technologies like SELDI-TOF-MS and MALDI-MS for the structural analysis of N-glycans, as well as mutation analysis for a plethora of novel genes. Overall, more than 20.000 patients with unexplained (metabolic) diseases have been screened for a glycosylation defect. Several new types of CDG have been discovered. From this viewpoint, it is certain that the network's activities have contributed both to the detection and diagnosis of CDG patients, and to the awareness and knowledge about these diseases among the medical community. However, due to the complex nature of the glycosylation pathways, pinpointing the genetic defect in all patients is not trivial. For the same reason, progress towards the development of therapies for CDG is slow.

After 4 years, more than 200 unsolved cases have been transferred to the research laboratories for further investigations. They represent a rich source for the identification of novel defects, and for the elucidation of basic mechanisms of glycosylation. The network is committed to solving this cases as soon as possible, for the sake of the patients and their families.

Description of the individual partners' contributions

Most partners in the network are involved in the diagnostic testing of CDG-case. All national and peripheral centres recruit patient samples, and forward the material and the information to the core laboratories, from where the samples are then dispatched to the different research laboratories that offer expert testing.

The group in Leuven is coordinating the network, managing the central database, and has a diagnostic and research activity for some of the diseases.

The group in Paris is also involved in the core activity. In addition, this group has organised a very successful international meeting in 2007.

The group in Nijmegen is the third core centre, with a large diagnostic and a strong training activity. The group is responsible for the interactions with the new national referral centres, and has also organized the external QA scheme.

The groups in Zurich have a task in the expert diagnosis, and in the coordination of the (pure) research activities and is responsible for the scientific program of the meetings.

The group in Barcelona offers one of the largest clinical recruitment activities. The group is also in charge of the work package on awareness, and supervises different other centres.

The London group recruits samples from all over UK, and is involved in expert diagnostics, and in training activities for primary care physicians.

The groups in Brussels, Heidelberg, one of the groups in Paris (formally Villejuif) and one of the groups in Catania are all involved in the expert diagnosis of unsolved CDG cases. They all have a research activity into CDG as well.

The groups in Athens, Catania, Porto, Warsaw, Copenhagen, Prague, Sofia, Jerusalem, Haifa and Zagreb all act as national referral centres for the recruitment of patient samples. They offer first-line diagnostic services and have direct interactions with the local clinicians. They all have a task in raising awareness about the disease in their countries.

The group in Madrid has a history of CDG testing, and has become a partner in the network in the past few years. The group offers first-line testing and molecular analysis for the more common types of the disease.

Orphan Holding and Orphan Europe Academy (Orphan s.a.r.l.) are involved in the research towards therapies for CDG and in the organisation of the FOCUS course on glycosylation, respectively.

Climb is the patients and parents organization, that focuses on the training of primary care physician and non-specialists in CDG, and on information for the families.

Certus is the IT company that has developed the software for the database.

2. Dissemination and use

The network has generated international scientific publications as well as brochures for patients and families.

The website, which was developed during the project, will be maintained.

The database will be maintained after the granting period, and the group is preparing for another international congress in 2010. The external QA scheme for CDG screening, which was initiated in the context of the network, has been adopted by ERNDIM, and will be offered annually from now on.

3. Participants

Partic. No.	Participant name	Participant short name	*Country*
01	Katholieke Universiteit Leuven	K.U.Leuven	B
02	Assistance Publique-Hôpitaux de Paris, Paris 2a/ Hôpital Bichat 2b/ Hôpital Necker	AP-HP	F
03	Radboud Universiteit Nijmegen	UMCN	NL
04	University of Zurich	UZH	CH
05	Hospital Sant Joan de Déu, Barcelona	HSJD/IBC	E
06	University College, Institute of Child Health, London	UCL	UK
08	Eidgenössische Hochschule, Zurich	ETHZ	CH
09	Christian de Duve Institute of Cellular Pathology, Brussels	ICP	B
10	INSERM U504, Villejuif	INSERM	F
11	Institute of Child Health, Athens	ICH	GR
12	University of Catania	UNICT	I
13	University of Porto	IGMJM	P
14	Children's Memorial Health Institute, Warsaw	CMHI / CZD	PL
15	University Hospital of Copenhagen	RH	DK
18	Charles University, Prague	CU	CR
19	Medical University of Sofia	MBALMBAL "Alexandrovska"	BU
20	Universidad Autonoma de Madrid	CEDEM- UAM	E
22	Rambam Medical Center, Haifa	Rambam MC	IL
23	Department of Paediatrics, Zagreb	MS-UNIZA	CR
25	Institute of Chemistry and Technology of Polymers, Catania	CNR	I
26	Orphan Europe Holding s.a., Paris	Orphan Europe H	F
27	CLIMB, Crewe	CLIMB	UK
28	Universitätskinderklinik, Abt. 1, Heidelberg	UK-HD	D
29	Orphan Europe s.a.r.l., Paris	Orphan Europe S	F
30	Certus Technology Assoc. Ltd., Exeter	Certus	UK
31	Hadassah Medical Center, Jerusalem	MC Jerusalem	IL

Coordinator contact details

Coordinator	: Prof. Gert Matthijs
Address	: K.U.Leuven, Campus Gasthuisberg Center for Human Genetics Herestraat 49, O&N1, POBox 602 3000 Leuven Belgium
Tel	: +32-16-346070
Fax	: +32-16-346060
E-mail	: gert.matthijs@med.kuleuven.be

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Project public website : www.euroglycanet.org