### Partners:

- Siemens AG, Erlangen, Germany
- Lynkeus SRL, Rome, Italy
- I.R.C.C.S. Giannina Gaslini, Genoa, Italy
- University College London Great Ormond Street Children's Hospital, London, UK
- Assistance Publique Hopitaux de Paris, Hôpital Necker, Paris, France
- European Organisation for Nuclear Research (CERN), Geneva, Switzerland
- Maat G Knowledge, Toledo, Spain
- University of the West of England, Bristol, UK
- University of Athens, Athens, Greece
- Università degli Studi di Genova, Genoa, Italy
- National Institute for Research in Computer Science and Control (INRIA), Sophia Antipolis, France
- European Genetics Foundation, Bologna, Italy
- Aktsiaselts ASPER BIOTECH, Tartu, Estonia
- Gerolamo Gaslini Foundation, Genoa, Italy

## www.health-e-child.org

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### Project co-ordinator: Siemens AG, Germany Dr. Jörg Freund Tel: +49 (9131) 84-3389 Fax: +49 (9131) 84133389 E mail: joerg.freund@siemens.com URL: http://www.Health-e-Child.org

Project Management & Dissemination Lynkeus Srl, Italy Tel: +39 (06) 844 08 01 emf@lynkeus.com sattanino@lynkeus.com a.trezzani@lynkeus.com



# An integrated platform for European paediatrics based on a Grid-enabled network of leading clinical centres

### **Objectives of the project**

• To gain a comprehensive view of a child's health by vertically integrating biomedical data, information, and knowledge, that spans the entire spectrum from genetic to clinical to epidemiological.

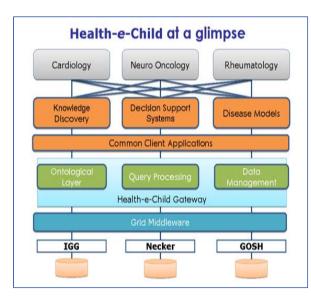
• To develop a biomedical information platform, supported by sophisticated and robust search, optimisation, and matching techniques for heterogeneous information, empowered by the Grid.

• To build enabling tools and services on top of the Health-e-Child platform, that will lead to innovative and better healthcare solutions in Europe.



Fashioned around three paediatric diseases with at least partly unknown causes, classification and/or treatment outcomes - heart diseases (right ventricular overload [RVO], cardiomyopathies), inflammatory diseases (juvenile idiopathic arthritis [JIA]), and brain tumours (gliomas), Health-e-Child is building the enabling tools and services that improve the quality of care and reduce its cost by increasing efficiency, through:

Integrated disease models, Database-guided decision support systems, Cross modality information fusion and data mining for knowledge discovery.



Key to the Health-e-Child system is the establishment of multi-site, vertical, and longitudinal integration of biomedical data, information and knowledge delivered via a Grid-based platform, supported by robust tools for search, optimisation and matching processes. The core of Health-e-Child revolves around its efforts dedicated to meeting the challenges entailed in biomedical information analysis.

Central to these are Health-e-Child's research activities centring on the tasks of integrated disease modelling, decision support and knowledge discovery for the advancement of personalised medicine.

The following are a few telling examples of Health-e-Child's ongoing research activity in each of the three tasks.

#### **Disease Modelling in Cardiology**

Health-e-Child is currently addressing the main tasks of developing learning 3D+t segmentations of the right ventricle, and electromechanical and physiological models of RVO + CM. The project's research goals concentrate on:

• identifying significant parameters for subtypes of cardiomyopaties that could lead to indications for additional genetic tests,

• adapting generic models to clinical data to extract patient-specific high-level discriminative features for decision support and knowledge discovery,

validating new measurements for diagnosis.

• indicate the capacity of drugs to stop/slow down disease evolution (automatic suggestion of drug prescriptions)

• analysing the correlation between genomic, proteomic, clinical and image data, with images mapped to (few) quantitative parameters and establishing a candidate gene set (responsible for bone remodelling) for study.

### Knowledge Discovery in Brain Tumours

Priority research goals of Health-e-Child in this area address actions to develop applications to:

 $\bullet$  verify the diagnosis/categorization of low-grade gliomas

• correlate clinical, imaging, and genomic data

• correlate prognosis with tumour origin site

 $\bullet$  define prognosis (e.g., correlations with spectroscopy)

- suggest treatment strategies
- predict outcome
- provide more precise classification of diseases

• detect correlations between age and outcome and between genetics and outcome

elaborate meta-analyses of published findings.

### **Decision Support in Cardiology**

The project is currently developing tools that will enable:

- the monitoring of right ventricular overload and decision support based on similar cases (similarity search based on specified features and association rules extraction for the given case)
- the prediction of whether atrial septal defect (ASD) will close by itself or will become larger, thereby precluding trans-catheterisation

• the prediction of "high-risk" ASD patients in whom erosion/rupture after trans-catheterisation is likely.

#### **Knowledge Discovery in Rheumatology**

Applied to juvenile idiopathic arthritis (JIA), Health*e*-Child focuses on the specific research goals of

- identifying gene variant combinations (haplotypes) correlated with particular diseases (bones/joints erosion)
- comparing the presence of different proteins in fluid at different stages of the disease to discover behaviour of cells close to fluid
- improving the current classification of JIA subtypes, and identifying homogeneous groups of clinical features
- elaborating explicit criteria for the early prediction of disease outcome/evolution
- developing image-based methods which rapidly indicate the capacity of drugs to stop/slow down disease evolution (automatic suggestion of drug prescriptions)

• analysing the correlation between genomic, proteomic, clinical and image data, with images mapped to (few) quantitative parameters and establishing a candidate gene set (responsible for bone remodelling) for study.

