



Colibrì (COLLECTION of pedIatric Brain Images and COLLECTION of pedIatric Brain Images - www.colibri.net) is a collaboration between 19 Italian scientific hospitals (IRCCS) – see map for locations - coordinated by three large partners: Bambin Gesù in Rome, "E. Medea" - Bosisio Parini in Lecco and San Raffaele Hospital in Milan.



COLIBRI: A NETWORKING PROJECT FOR RARE DISEASES

A disease or disorder is defined as rare in Europe when it affects less than 1 in 2000. While this may seem to indicate a very small phenomenon, there are between 6000 and 7000 known rare diseases, affecting an estimated 30 million people in Europe alone. Indeed, rare diseases as a category are considered a public health priority for which the EU has established a specific policy.¹

What rare diseases have in common is difficulty in finding the right diagnosis and scarce resources for medical research. Each disease has its own specific traits, and with only a handful of cases in any given region, networking is essential to be able to share information on it and to reach critical mass in understanding it and trying to find a treatment.

The Colibrì Project arises in this context. Funded by the Italian Ministry of Health, Colibrì aims to share magnetic resonance images (MRIs) between 19 research hospitals throughout Italy that specialize in the field of pediatric neurological diseases.

In this context, the network and specialized ICT have a strategic importance. We spoke with Dr. Fabio Triulzi, Project Lead of Colibrì.

Words
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Pictures
Dr. Fabio Triulzi,
Project Lead
(opposite); map
shows centres
involved in the
project

1. http://ec.europa.eu/health/rare_diseases/policy/index_en.htm



“Certainly the technological aspect presents a challenge for the medical community. Each hospital is a fortress, because its primary objective is the defense of patient privacy. However medical data sharing is very important, so flexibility is necessary to circulate information between hospitals and colleagues. In relation to that, network and information technologies play a strategic role because they help to combine privacy and sharing. The interconnection of the Scientific Hospitals (IRCCS) via the Italian research and education network GARR since 2005 means a lot, not only technologically but also in terms of attitude towards sharing, because there is an awareness of being part of a network and of being supported by an advanced and reliable e-infrastructure, which makes data sharing safe and easy. For Colibrì, GARR created a virtual private network to provide high security for data travelling over the network, complemented by secure access via IDEM, the Italian identity federation”.

Let's say I'm a doctor who has just performed an MRI and I suspect the patient is suffering from a rare disease. What can I do?

First, I submit the image to the evaluation of a committee to check whether it fulfills a certain set of prerequisites; then - but only after being approved by the committee - I share the image with the other centers. Of course, patient data are encrypted and what I share is just a protocol number.

At this point the other centers can offer suggestions, further analyze some of the tests and evaluate the case in a collegial manner: all this increases the chance of making a correct diagnosis. It may also happen that another center has already made the diagnosis, achieving a reliable genetic typing: in this case the image is “promoted” as informative to enrich the database and increase the critical mass of knowledge on a specific disease.

Is it important to make an early diagnosis regardless of the existence of a cure?

Of course. This is always the case in medicine, one must try to figure out what the problem is regardless of whether there may be a therapy. Unfortunately many rare diseases don't have a specific cure, but a diagnosis is important in terms of family counseling because in most cases we find a disease with a genetic background, hence if I identify the disease and understand how it is transmitted genetically, I can provide important information both to the parents and to the patient.

Furthermore, there are diseases that can significantly improve with even small changes, for example in the metabolic diseases a proper diet can make the difference: clearly if I begin to treat the disease promptly, I have better chances of success.

How do you see the future? Do you plan to expand the project internationally?

Yes, our idea is to extend our collaboration at the international level. We would like to understand what is happening outside Italy because we know that there are similar experiences elsewhere in the world: the key for the success of Colibrì will be in our ability to collaborate with more and more centers worldwide.

The goal of our network is to make sure that if we see, say 40 cases of a rare disease in Italy, estimating that worldwide, with over 7 billion people, there could be perhaps 7000 people suffering from this disease, thanks to an international network we could see 200 cases and this really makes the difference. Being connected to the Italian research network GARR which in turn is interconnected to the GÉANT international research network can be a strong added value, both in terms of technology and networking.