

## String of Pearls and congenital heart disease

In the Netherlands, about 1400 children a year are born with a congenital heart defect. Thanks to the advances in cardiovascular surgery over the past few decades, most of these patients now reach adulthood. Consequently, there are now more than 25,000 Dutch adults living with a congenital heart condition and this number is increasing by about 5 per cent a year. Given that approximately 25,000 children also have such a condition, the country's total patient population is estimated at about 50,000. CONCOR, the national registry and DNA bank of adult patients with congenital heart disease and one of the PSI "pearls", co-ordinates the tracking of those aged 18 and over for the purposes of scientific research.

The term "congenital heart defect" covers a variety of conditions. The four most significant are:

- Ventricular septal defect: holes in the wall separating the two ventricles of the heart;
- Atrial septal defect: holes in the wall separating the two upper chambers of the heart;
- Aortic coarctation: narrowing of the aorta;
- Tetralogy of Fallot: narrowing of the pulmonary valve and a hole in the wall separating the two ventricles of the heart.

This diversity makes it difficult for individual hospitals to generate sufficient data for scientific research from their own patient groups alone. Because CONCOR brings together the information on a national scale, a greater research population is available and so it is easier to produce meaningful results.

When CONCOR was launched in 2002, about 8000 adult patients from the eight Dutch teaching hospitals were included in the registry. In 2004 it also began registering those at other hospitals. It now holds data on more than 11,000 people from 103 Dutch hospitals. DNA has been isolated from almost half of these subjects.

### Scientific value

Adults living with a congenital heart defect represent a new and relatively young patient population, about whom comparatively little is known. What is now clear,

though, is that a large proportion of them develop complications in the longer term. Scientific research is therefore needed in order to improve our understanding of the various conditions concerned. Moreover, as more and more of the genes involved in the development of the heart are identified, so scientific interest in the genetic causes of congenital defects is increasing. Since the majority of the patients are now of reproductive age, it can be very important for them to know whether their condition has a hereditary component. However, successful genetic research requires a large population to study.

By joining forces with the String of Pearls Initiative (PSI), CONCOR is now able to make use of its infrastructure. This covers those patients who are being treated at the Dutch teaching hospitals. The data registered by CONCOR has to be updated on a regular basis, which is something the people, the methods and the technology available to PSI can facilitate. That benefits the quality of the data and hence the reliability of the results derived from it. Conversely, participation by CONCOR reinforces PSI's position as a bulwark of research – and not just in the Netherlands, but at the European level as well.

Cardiologists and other medical specialists from all the participating institutions can make use of information from the database. To gain access, however, they must submit an application which is assessed by a medical ethics committee.

### About the pearl

As CONCOR was already doing, the congenital heart disease "pearl" collects information about patients aged 18 or over with a condition of this kind. Some, but not all, have already undergone surgery. Patients are requested to take part through the consultant responsible for their treatment, usually a cardiologist, and must give prior written consent for their clinical data – type of condition, details of any operations carried out and complications suffered, for instance – to be included in the database. They are also asked to give two samples of blood, from which their DNA is isolated and stored. During the regular updates, information about significant complications and any other changes are added to the records of patients already registered in the database.

Quality checks are also carried out on a regular basis. These involve CONCOR research nurses reviewing a representative sample of the registered patients to confirm that the data held about them and their DNA samples have been recorded correctly.

The partnership between CONCOR and PSI was instigated by the Interuniversity Cardiology Institute of the Netherlands (ICIN), an institute of the Royal Netherlands Academy of Arts and Sciences (KNAW) representing the cardiology departments of all the Dutch teaching hospitals.



## **The String of Pearls Initiative**

The String of Pearls Initiative is the result of a unique partnership between the eight Dutch university medical centres (teaching hospitals). Founded in 2007 by NFU, the Dutch Federation of University Medical Centres, the initiative gathers clinical data and biomaterials from all the participating institutions so that together, they can promote the ad-

vancement of science, improve patient treatment and encourage the development of new products, as well as strengthening the economic position of biomedical research in the Netherlands. Initially, the project is focusing upon nine groups of medical conditions, its so-called "pearls". In the future, its activities may be expanded to include

others. For more information, you can contact the String of Pearls Initiative at [info@string-of-pearls.org](mailto:info@string-of-pearls.org).