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Creation of a single-center DNA-bank of congenital heart disease with focus on cardiac phenotype

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Background: The creation of DNA-banks is the necessary basic requirement for developing genetic research in Pediatric Cardiology. Precise cardiac phenotyping is mandatory in order to exploit the samples. The main future objective of such programs will be to test different candidate genes derived from animal and human research in different groups of CHD.
Methods: We started a single center DNA-bank called CARREG (CARdiacREGulation genes) of patients' blood samples with congenital heart disease. This DNA-bank is not restricted to a specific research project. Initially this program was limited to CHD of the outflow tract (OFT) and then extended to all types of CHD after informed consent of patient and legal representative. Samples are also taken from consenting parents of an affected patient. Congenital heart disease of each participant is fully phenotyped according to clinical exam, echocardiography, imaging and surgery protocol. Each patient is grouped in one of the 10 major groups according to the recently published "Anatomic and clinical classification of congenital heart defect" classification (ACC-CHD). Once grouped, each CHD is described precisely according to the International Pediatric Cardiac Congenital Code (IPCCC) with the number of necessary codes.
Results: Starting from April 2009 1243 blood samples have been collected and stored in a dedicated space of the institution's DNA-bank. 698 are patients' blood samples. 545 blood samples come from parents or relatives. According to ACC-CHD there are 7 samples in group 1, 6 samples in group 2, 13 samples in group 3, 16 samples in group 4, 18 samples in group 5, 30 samples in group 6, 14 samples in group 7, 551 samples in group 8, 39 samples in group 9, 4 samples in group 10.
Practical outcomes : In order to investigate genetic foundations of CHD, DNA-banks have to contain a representative number of all possible CHD. The initial restriction to patients with OFT disease explains the present predominance of group 8 in our gene bank. To make international comparison and data exchange easier, cardiac phenotyping in CARREG relies on IPCCC and the newly developed ACC-CHD. In the future this program will be extended to a national level.