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Insufficient Medical Primary Health Care of Adults with Congenital Heart Defects associated with Genetic Disorders

Neidenbach R.(1), Diebold I. (5), Pieper L. (2), Oberhoffer R. (1,3), Schelling J. (4), Sanftenberg L. (4), Nagdyman N (1), Klein H.-G. (6), Seidel H. (1,6), Ewert P. (1), Kaemmerer H (1)
(1) Department of Paediatric Cardiology and Congenital Heart Defects, German Heart Centre Munich, Technical University Munich, Germany;
(2) Department of Behavioural Epidemiology, Technical University of Dresden, Germany;
(3) Department of Preventive paediatrics, Department of Sport and Health Sciences, Technical University of Munich, Germany;
(4) Institute for General Practice, University Hospital of the Ludwig-Maximilians-University Munich, Germany;
(5) Medizinisch Genetisches Zentrum Munich, Germany;
(6) Zentrum für Humangenetik und Laboratoriumsdiagnostik, MVZ Martinried, Germany;
(7) Institut für Humangenetik, Technical University of Munich, Germany

Introduction:
Due to recent medical advances, approximately 280,000 adults with congenital heart defects (ACHD) are currently living in Germany. Although most CHD occur sporadically, a causative genetic component is very likely, including chromosomal, monogenic, multifactorial inheritance disorders or environmental factors.
Most ACHD need experienced medical advice. Their care concept not take into account cardiological issues, and also a genetic counseling by a clinical geneticist experienced in CHD.Recent German data show, that more than 200.000 ACHD are not treated by certified ACHD-specialists. As many general physicians do not have specific knowledge concerning CHD, genetic disorders and related comorbidities, particularly patients with rare hereditary diseases are at risk to be not advised or treated sufficiently.
Aim of the study is to demonstrate “real life” primary care of ACHD and with hereditary diseases / genetic disorders.

Methods: A cross-sectional study, questionnaire-based survey including data of ACHD and general practitioners (GP).

Results: Included were 954 ACHD (51.7% female, mean age 36.0 ± 12.4 years) and 369 general practitioners (GP). GP treat patients with any type of CHD, including rare hereditary diseases such as Marfan syndrome (n=95; 25.7%), Ehlers-Danlos-Syndrome (n=28; 7.6%), Turner syndrome (n=74; 20.1%) or Morbus Fabry (n=27; 7.3%). In case of cardiac or non-cardiac problems and questions, more than 70% of the surveyed patients contact their GP primarily. From the patient's perspective, up to 50 % seek for disease oriented advice, which is often unmet. GP indicate in more than 70% of the cases to be poorly informed about ACHD specific physicians and centres where also a genetic counselling takes place.

Conclusions: ACHD need optimized, comprehensive medical care and modern, scientifically based care concepts, particularly if a genetic disorder coexists.
Although GP’s have to guide ACHD to receive adequate diagnosis, treatment and preventive measures, GP’s are insufficiently informed about the existing ACHD facilities.
In the future, ACHD-experts and -centers must become more visible and cooperate stronger with GPs to provide adequate management treatment and therapy concepts, including genetic aspects.