Problems in the organization of care for patients with adult congenital heart disease

Problèmes dans l’organisation des soins de la cardiopathie congénitale

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Summary  The prevalence of congenital heart disease among adults in Europe, or in any country in Europe, is not known. This is due to a lack of agreement on the incidence of congenital heart disease, with estimations varying from four per 1000 births to 50 per 1000 births, and it is not known how many patients with congenital heart disease have died. Based on several studies that estimated and calculated the number of adult patients with congenital heart disease, the number of patients should be much higher than the number of patients that are actually seen in specialized centres throughout Europe. This implies that either a large proportion of adult patients with congenital heart disease do not receive appropriate medical care, or that the calculations and estimations are grossly wrong. A combination of the two is also possible. A substantial expansion of the number and size of specialized centres for adult congenital heart disease is advocated, but since setting up (and running) a service for this disease is a costly affair, and because uncertainty remains about the actual number of patients needing specialized care, this has been difficult to realize in most European countries in the past few years.

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Congenital heart disease is the most common of all congenital defects. In terms of organization of care, the emphasis has always been on the impact of congenital heart disease in the first years of life: in the era before cardiac surgery, congenital heart disease was the most important cause of infant death and only a few of those born with complex congenital heart disease survived until adulthood. Since the introduction of cardiac surgery by Lillehei in 1953, a gradual improvement in survival has been achieved over the subsequent decades. This has led to a situation where the number of patients with congenital heart disease who reach adult age has gradually increased, and with the current level of healthcare it is expected that over 90% of babies born in developed countries with congenital heart disease will now survive until adulthood.

Most patients who survive into adulthood after cardiac surgery in childhood, are not 'cured' for life; almost all have residual abnormalities [1]. The need for continuation of specialized care after their childhood years has been emphasized since the early 1980s [2]. The existing healthcare networks for adult patients with cardiac problems — the regular cardiology services — are not equipped for this task, because of lack of training and exposure and therefore skills in this specific field [3]. Awareness of this issue has led to the establishment of programmes for specialized adult congenital heart disease (ACHD) care throughout Europe. Some programmes started in the 1980s, many in the 1990s, and the number of centres is still increasing. In addition, existing centres tend to grow, many of them rapidly [4].

In a recent survey, Moons et al. [5] identified 70 centres in Europe that could be labelled as centres for ACHD. Altogether, these 70 centres had some 130,000 adult patients in their care. The authors stated that this was only a fraction of the entire population of patients with ACHD, that this population is heavily under serviced in terms of available care at an adequate level and that many more centres — or much larger units — would be needed.

Establishing and running a unit for specialized ACHD care is, however, a costly affair; as these chronically ill patients with considerable morbidity [6], need a relatively large amount of "doctor time" and claim a fairly large proportion of health-care resources [7]. If we want to convince healthcare planners and boards (or directors of hospitals) of the necessity of investing in such a costly service, we need to answer a few basic questions. How many patients in total are involved? How many of these patients have complex ACHD, how many have ACHD of moderate severity and how many have mild ACHD? Which defects really require specialized tertiary referral ACHD care and which can be dealt with in regional hospitals by cardiologists? How many patients need no special cardiac follow-up at all? These are simple and fair questions, but are difficult to answer.

We do not know the incidence of congenital heart disease (i.e., how many patients are born per year with congenital heart disease in a specific population or country), because reports on this topic vary enormously from four per 1000 live births to 50 per 1000 live births.

We also do not know the prevalence of congenital heart disease (i.e., how many patients are alive with congenital heart disease in a population or country), because we do not know the starting point (the incidence) or how many patients have died. We are not aware of any country in which there is a population-based registry that is solid and detailed enough to answer these questions.

What should be considered as complex congenital heart disease? There is no uniformity in the definition that has been used in the various published studies, task force reports and position papers. For example, tetralogy of Fallot and atrioventricular septal defects — both fairly large diagnosis groups — are defects that are classified as severe or complex by some and as moderately severe by others. Neither is there consensus about which patient group actually needs highly specialized, tertiary referral care. There is not much discussion about really complex congenital heart disease (it is accepted that patients should be seen in a specialized centre) and there is also a shared belief that truly simple lesions do not need specialized care. But what is the best option for the group of patients with moderately complex congenital heart disease? Regional care provided by the regular cardiology services near the patient, tertiary referral specialized care, or both options as shared care? And who should do a catheter-based intervention or a surgical proce-
dure in a simple defect detected in adulthood? Atrial septal defect closure and aortic coarctation are the most typical examples. Should they all be referred to a tertiary referral centre?

With the major issues of incidence and prevalence largely unresolved, and no consensus about the number and category of patients that should be seen in a tertiary referral, specialized ACHD centre, it is very difficult to make plans to create adequate healthcare for these patients. This probably explains the current situation in Europe regarding ACHD care, as described by Moons et al. [5].

There have been some excellent attempts in recent years to tackle this issue. Although based mainly on estimations and not on actual measurements, and coming mostly from the USA, these studies might be very helpful in trying to understand the scope of the problem in Europe.

What is the problem with assessment of congenital heart disease incidence?

The landmark study on this topic was published by Hoffman and Kaplan in 2002 [8]. They reviewed 62 studies published after 1955 on the incidence of congenital heart disease, and found that it varied from four to 50 per 1000 live births. This huge variation requires explanation, and Hoffman and Kaplan provided that. In short, large, population-based studies will represent the number of live births reliably but will rarely be able to use sophisticated screening tools for a very high number of patients, so will probably fail to detect all cases of congenital heart disease. This will lead to an underestimation of the real incidence.

Smaller studies, applying sophisticated screening tools (e.g., repeated echo studies throughout the first months of life), will probably detect all cases of congenital heart disease, but the population in which they are applied is often small and therefore the outcome might not be representative of the entire population; owing to selection bias, these studies tend to overestimate the incidence of congenital heart disease.

The use of echocardiography is also important. Early studies, before the mid 1980s, did not have echocardiography as a diagnostic tool and will have missed many small defects. However, even in later studies that applied echocardiography, the differences between studies remained large. It appears that the huge variation was explained almost exclusively by varying percentages of mild and trivial lesions being detected.

The incidence of moderate-to-severe congenital heart disease was similar in most studies and appeared to be relatively stable for many years and in many different countries, largely irrespective of the use of echocardiography: approximately six per 1000 live births, of which three per 1000 were severe and three per 1000 were moderate.

Based on the meta-analysis, we may conclude that the incidence of congenital heart disease that is relevant to the discussion about the need for specialized ACHD care is at least three per 1000 live births with regard to complex congenital heart disease, and possibly as many as six per 1000 live births if moderately severe congenital heart disease is included.

In a paper from the UK published in 2001, Wren and O’Sullivan [9] reviewed all confirmed cardiovascular malformations diagnosed between 1985 and 1999 in children born in 1985 to 1994 in one health region (Newcastle, England). The observed incidence of congenital heart disease in infancy was 5.2 per 1000 live births. It was very interesting that of all the patients diagnosed with congenital heart disease who had a life expectancy greater than 16 years of age, 35% were diagnosed after the first year of life. This indicates that basing the incidence of congenital heart disease on screening in the first year of life, even in a relatively recent era with the benefit of echocardiography, leads to a substantial underestimation of the number of patients that is relevant for the discussion about care for ACHD. Based on the complexity of the disease (and using a slightly different definition to Hoffman and Kaplan) they predicted that two per 1000 live births would need specialized care for congenital heart disease beyond childhood. This is substantially lower than the estimation of Hoffman and Kaplan, partly explained by lower detection of congenital heart disease (despite a substantial number of patients detected after the first year of life) and partly by the definition of which patient category needs specialized care throughout life.

Two diagnoses complicate the discussion about the incidence of congenital heart disease: bicuspid aortic valve (BAV; 13 per 1000 live births) [10] and congenital mitral regurgitation/stenosis, including mitral valve prolapse (40–50 per 1000 live births) [11]. These lesions are congenital and could therefore rightfully be included in reports about incidence. In particular, BAV has a fairly high likelihood of needing surgical intervention during adult life. If these lesions were included in the calculation of need for specialized ACHD care, the numbers (and the costs attached to these programmes) would be greatly inflated. It is our opinion that because patients with BAV and congenital mitral stenosis/regurgitation, in considerable numbers, have been taken care of for so long by general adult cardiologists and surgeons (who have the necessary expertise because the lesions occur so frequently), they do not fit into the category of ACHD patients for whom specialized care should be arranged.

We come to the conclusion that for the discussion about healthcare planning for ACHD and the number of patients that need specialized ACHD care, an incidence of at least two per 1000 live births should be used (Wren and O’Sullivan) and possibly up to six per 1000 live births (Hoffman and Kaplan: combined complex and moderately severe cases).

What is the problem with assessment of congenital heart disease prevalence in Europe?

There are no reliable data on the prevalence of congenital heart disease in Europe. According to the definition, prevalence is the total number of patients with congenital heart disease at a certain moment or in a defined period: all patients born with congenital heart disease minus those who have died. The total number of live births per year has been registered quite reliably in most countries for many decades. If one assumes an incidence of complex and moderate-to-
Table 1 Estimated number of patients with adult congenital heart disease in the USA and in six Western European countries.

<table>
<thead>
<tr>
<th>Country</th>
<th>Population a</th>
<th>No. of identified ACHD centres</th>
<th>Severity of congenital heart disease</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Severe</td>
</tr>
<tr>
<td>USA</td>
<td>280,000,000</td>
<td>4</td>
<td>117,000</td>
</tr>
<tr>
<td>France</td>
<td>64,000,000</td>
<td>5</td>
<td>26,676</td>
</tr>
<tr>
<td>Germany</td>
<td>82,000,000</td>
<td></td>
<td>34,164</td>
</tr>
<tr>
<td>Italy</td>
<td>58,000,000</td>
<td>5</td>
<td>23,400</td>
</tr>
<tr>
<td>Belgium</td>
<td>10,000,000</td>
<td>4</td>
<td>4178</td>
</tr>
<tr>
<td>The Netherlands</td>
<td>16,000,000</td>
<td>7</td>
<td>6669</td>
</tr>
<tr>
<td>United Kingdom</td>
<td>61,000,000</td>
<td>12</td>
<td>25,536</td>
</tr>
</tbody>
</table>

ACHD: adult congenital heart disease.

a Population of USA in 2000; population of European countries in July 2009.

severe congenital heart disease to be between two and six per 1000 live births (see above), the number of patients who have been born with congenital heart disease in the past 60 years, for example, can be calculated. However, we have no means of knowing how many patients have died. We are not aware of any nationwide registry that has run for such a prolonged time, with the required high accuracy and specificity that could supply the necessary data.

The paucity of data on this topic has hindered the development of ACHD all over the world. Two groups, one from the USA and one from Canada, did excellent work in this area and shone some light on what was, until recently, a dark field.

In 2007, Marelli et al. [12] reported on measured prevalence of congenital heart disease in Quebec. This group were able to do that because every individual in Quebec obtains a unique Medicare number, and all diagnoses and health-care services rendered are recorded systematically until death. All patients who came into contact with the health-care system between 1983 and 2000 were entered into a congenital heart disease database. In the year 2000, they reported a prevalence of congenital heart disease for the entire population of 5.78 per 1000; for children, the prevalence was 11.89 per 1000 and for adults it was 4.09 per 1000. The prevalence of severe congenital heart disease was 1.45 per 1000 children and 0.38 per 1000 adults. A substantial proportion of the severe lesions were not followed regularly in a centre with specific ACHD expertise [13].

Severe lesions were defined as tetralogy of Fallot (and similar abnormalities, such as pulmonary atresia with ventricular septal defect and truncus arteriosus), atroventricular septal defect, transposition of the great arteries in all possible forms and univentricular hearts. This is another definition of severe or complex congenital heart disease that differs from that of Hoffman and Kaplan, but is unarguably a list of severe malformations, with many residual abnormalities and high morbidity, for which lifelong specialized ACHD care is indicated. All other congenital heart disease lesions (that did not fit into this “severe” category) combined had a prevalence of 10.44 per 1000 children and 3.71 per 1000 adults.

If one assumes that the socioeconomic situation in Quebec is largely similar to that of Western European countries (currently and historically over the past 50 years), it might be possible to extrapolate some of these data to calculate patient numbers in Europe. For example, in France, with 64 million inhabitants, this would amount to 24,300 patients with complex congenital heart disease and 237,440 patients with other forms of congenital heart disease. For the Netherlands, with 16 million inhabitants, it would amount to 6080 patients with severe congenital heart disease and 59,360 patients with other forms of the disease. These calculated numbers of patients differ so much from what is known from clinical practice in The Netherlands, that one has to accept that there is either an enormous under-service for adult congenital heart disease in The Netherlands, despite its high-level, highly-organized health-care system, or that the methodology of the Quebec study has too many flaws to allow these calculations and extrapolation to the Dutch situation.

In 2001, the American College of Cardiology Bethesda Conference Task Force 1 presented in “The Changing Profile of Congenital Heart Disease in Adult Life”, which was their approach to estimating patients with ACHD in the USA [14]. Based on USA census data, the documented birth rates from 1940 to 1989 were averaged, enabling the total number of children born in the USA in this time period to be calculated. The incidence of CHD per 1000 live births was derived from the available literature. As indicated above, the choice of which study to use as the basis for the incidence of congenital heart disease has greatly influenced the eventual outcome. The survival rate for the first year of life and survival until the year 2000 were estimated in a complex but realistic way. The time period was divided into three phases of 20 years (1940—1959, 1960—1979 and 1980—1999) and the diagnoses were divided into three groups: complex, moderate and mild. Different estimated survival rates for the three subgroups in the three time periods led to the outcome: an estimated 787,800 patients with ACHD alive in the USA in the year 2000, when the USA had a population of 280,000,000 people. Of these, 117,000 had complex congenital heart disease, 302,000 had moderate congenital heart disease and 368,000 had mild lesions.

The estimation of numbers was an important mandate for the Bethesda Conference, as it was to provide the basis of programme planning and resource allocation. An extrapola-
tion of these estimations to the Western European situation — acknowledging all shortcomings and limitations of such extrapolations — is shown in Table 1.

It is interesting to see that for the group of patients with complex disease, the measured prevalence in Quebec is similar to the estimated prevalence from the Bethesda Conference. In contrast, the number of moderate and mild lesions in this estimation model is substantially lower: about 50% of the measured prevalence in Quebec.

What can be said about the number of patients with adult congenital heart disease in Europe?

It is questionable whether the estimations and calculations of the number of patients with adult congenital heart disease in the USA and Quebec are valid. Moreover, extrapolating numbers from the USA to arrive at an estimation of the situation of ACHD in Europe can also be disputed. There have been differences in socioeconomic situations in the past 50 years and in accessibility to health-care systems, and cardiac surgery for congenital heart disease was introduced somewhat later in large parts of Europe compared with in the USA. Currently, however, it is all we have, and it can be used to make at least a rough estimation of the situation.

For countries in the European Union, with an estimated population of 491,582,852 [15] in July 2009 (1.75 times the USA population in 2000), the number of patients with complex congenital heart disease would be approximately 180,000–200,000. These patients definitely deserve specialized care.

For moderate congenital heart disease, the figure would be at least 500,000 patients. One can argue whether specialized care for every patient in this category of severity is really necessary, but most will agree that at least part of the care for this patient group (e.g., surgical and catheter-based interventions) should be done in specialized centres [16].

Given that only 130,000 patients received specialized care in one of the 70 ACHD centres that could be traced by Moons et al., it seems obvious that we miss a large proportion of the patients in the health-care system designed to provide this care. However, we do not know whether these patients exist in numbers similar to those in the North American data and estimates. If they do, we do not know who is providing their care. General physicians? Cardiologists? Paediatric cardiologists?

Obviously, we need more data — specifically, European data. In our opinion, we need a European “Bethesda Conference”, and have asked the European Society of Cardiology to support this initiative. The aim of a European Society of Cardiology Task Force for ACHD would be to establish uniformity in definitions and methodologies for the investigation of the scope of the ACHD problem in Europe. An unequivocal classification of severity of congenital heart disease (which diagnosis belongs in which category) and which category of patients should be looked after in which level of care (tertiary referral centre or regular adult cardiology setting) is necessary. These definitions, together with recommendations on how to trace patients and how to calculate estimations, can be handed over to national cardiology societies, who will have to perform the difficult task of actually finding these patients in their own country, probably in cooperation with national working groups in ACHD. Only when armed with data on how large the problem is that we have to deal with, we do have a chance of convincing policymakers to allocate serious resources for ACHD, despite these economically difficult times.

Conflict of interest statement

None.

References