

# Congenital heart disease is no longer a paediatric specialty

## Adults with congenital heart disease must be managed by a new generation of subspecialist cardiologists, says John Deanfield



(credit Sam Rogers)

Five decades ago children with congenital heart disease were not expected to survive into adulthood but now >90% do. This success has been achieved by medical and surgical breakthroughs, step wise improvements in care and organizational changes. The result is a new subspecialty of adult cardiology, namely adult congenital heart disease (ACHD).

‘Congenital heart disease is no longer a paediatric specialty’, says Prof. John Deanfield (London, UK). ‘Adult cardiologists have to organise services to manage this growing challenging group of patients’.

Medical systems across Europe are at various stages of reorganizing care to address the needs of ACHD patients. The first principle is that adults with congenital heart disease need to be looked after in an adult environment with adult facilities. ‘There should be no such thing as a paediatric cardiology centre’, says Deanfield. ‘Every centre should treat patients with congenital heart disease over their whole life’.

Established paediatric cardiology specialist centres should have firm links with an adult institution to which patients can be seamlessly transferred.

Currently, there are very few physicians, surgeons, and support staff (nurses, technicians, etc) who are experts in ACHD. In most countries, care is being organized in a hub and spoke model with a small number of specialized centres and a network of care in the community. This model of care is designed to ensure that all patients have access to expert care appropriate to their condition. This has been supported by the European Society of Cardiology guidelines on the management of grown up congenital heart disease, first published in 2003 and updated in 2010.

The system then needs to be populated with properly trained people. In most European countries, the majority of experts in congenital heart disease has a background in paediatric cardiology. Adult cardiologists, however, are now also entering the field through formal training programmes that enable them to subspecialize in ACHD as part of their adult training.

Deanfield says it is ‘reasonable for specialists to come through the paediatric cardiology or adult cardiology route, provided they address gaps in their training. Adult cardiologists should have exposure to paediatric cardiology to really understand ACHD. Similarly, paediatric cardiologists must undergo training in adult medicine.

Research in ACHD is facing a number of issues. First is that the spectrum of cases in congenital heart disease is diverse, with relatively small numbers of different diagnoses at a large number of centres. This contrasts to coronary disease, which can be investigated in

adequately powered studies because there are large numbers of patients with a similar condition.

Second is what Deanfield describes as ‘a cultural barrier in paediatrics to undertaking research on patients’. Paediatric cardiologists who look after congenital heart disease patients have, until recently, been less keen on undertaking randomized trials which are standard practice for the evaluation of new treatments in adult cardiology.

Third is that collecting outcomes in ACHD has been more difficult. Large powered studies in adult cardiology use hard endpoints such as death, heart attacks, and strokes. Deaths, however, are relatively uncommon in adults with congenital heart disease and functional outcomes are required. These have been harder to quantify and standardize. Great progress has been made over the last decade in non-invasive imaging of the structure and function of the complex cardiac circulations that are frequently present in adult survivors. MRI, echocardiography and exercise testing can provide serial quantitative endpoints.

Progress in research on ACHD requires collaboration. That will enable scientists to recruit sufficient numbers of patients to conduct adequately powered, properly structured trials of outcomes, and the effects of treatment. Registries have been created in Europe and beyond but enrolment has often been patchy.

Funding is a fourth issue, with much of the research in cardiology driven by industry relationships and the development of new drugs. ‘Industry has not been very involved in ACHD research other than for devices’, says Deanfield. ‘But grant funders including the scientific charities are becoming more interested as the scientific community starts to submit better quality research proposals’.

The most important determinants of outcome in the ACHD population are the development of arrhythmia, ventricular failure, and valve function. Excitingly, catheter-based procedures, including stenting and percutaneous valve implantation, are increasingly used to treat many of these problems that may persist even after surgical repair in childhood. Understanding the late outcomes of congenital heart disease treatment is important, not only for patient care, but also to provide feedback to alter treatments in early childhood. A classic example is transposition of the great arteries which was historically treated with the Mustard or Senning operations. These were successful in childhood but had poor late outcomes. These disappointing late results enabled introduction of the arterial switch operation which creates a more anatomically normal heart and which has been shown through research to generate better long-term outcomes.

'Lifetime care of congenital heart disease must be the new standard', says Deanfield. 'Research in the adult population, who now survive even with complex congenital heart disease, will not only help patients, but drive medical, surgical and pharmacological innovation. Organisation of specialised care, which is happening across Europe, provides an exciting platform for high quality, practice changing research studies'.



## An important attempt to improve the outcome of congenital heart disease in Europe

### New recommendations for the organization of care for adults with congenital heart disease and for physician training in the subspecialty of 'Grown-up Congenital Heart Disease' in Europe

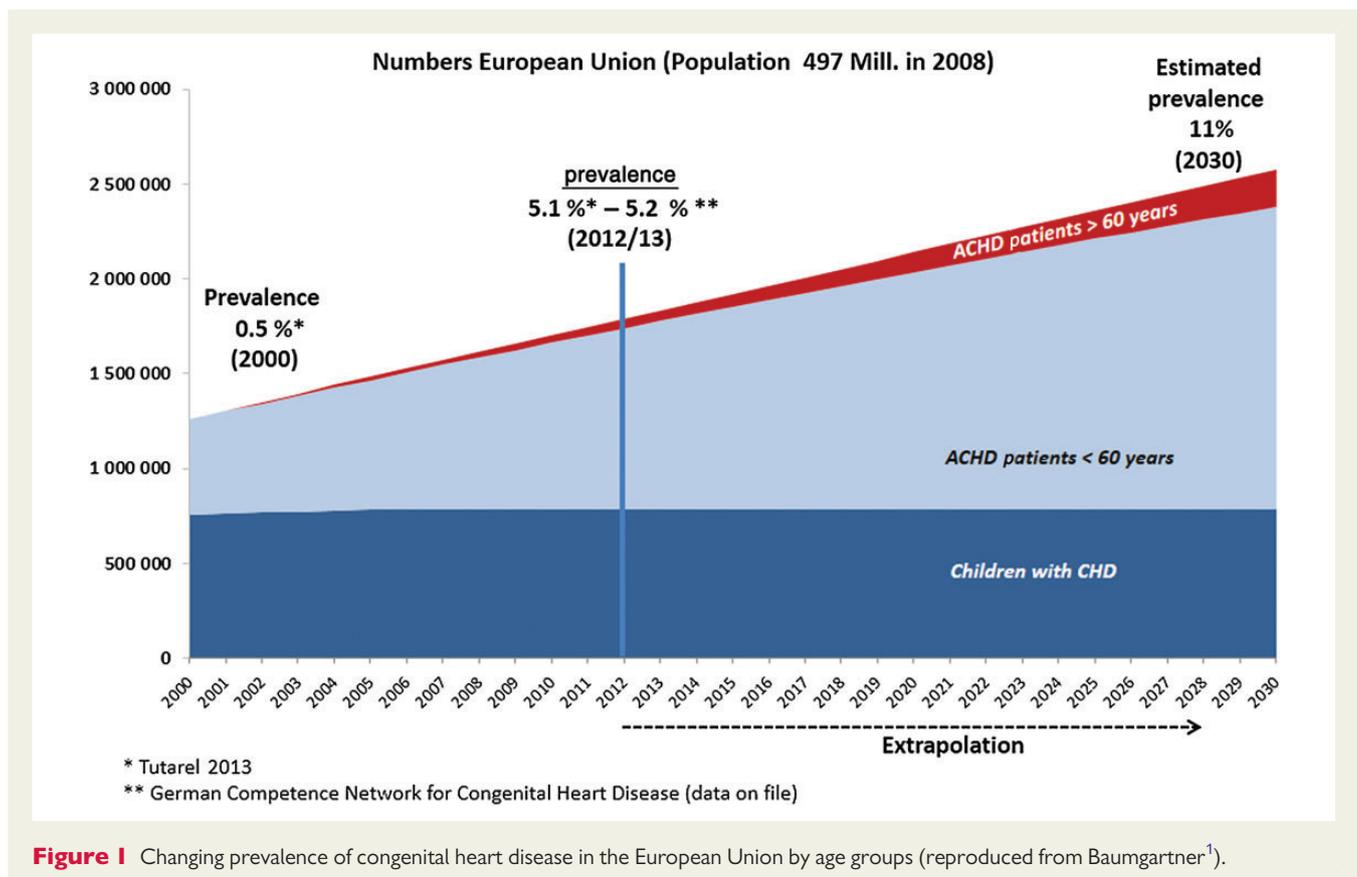
In the current issue of the *European Heart Journal*, the Working Group on Grown-up Congenital Heart Disease (GUCH) of the European Society of Cardiology (ESC) provides a position paper that sets standards for the organization of GUCH care and the training in this new subspecialty of cardiology.

Why is this of such great importance?

The major advances of paediatric cardiac surgery and paediatric cardiology during the last decades has resulted in an increasing number of patients with congenital heart disease who survive into

adulthood. While 90% of patients born in the 1950s with complex disease died during childhood, the majority born in the 1980s and later, survived beyond 18 years of age. Therefore, we are now faced with a new population of grown-ups with congenital heart disease (CHD) who need very special care.

In Europe, we currently estimate a population of ~2.3 million adults with congenital heart disease, which significantly outnumbers the paediatric CHD population of ~1.9 million and the numbers are expected to continue increasing (Figure 1).



**Figure 1** Changing prevalence of congenital heart disease in the European Union by age groups (reproduced from Baumgartner<sup>1</sup>).

Despite these impressive patient numbers, they nevertheless represent only a minority of the adult population with cardiovascular disease, considering that ~24 million may suffer from coronary artery disease as one of the major players. Thus, CHD is rarely seen in the non-specialized clinical practice of cardiology. In addition, the diversity of CHD and the resulting clinical problems are immense.

Although knowledge of adult CHD (ACHD) has increased in recent years, the spectrum of disease and the late complications are constantly changing due to the development of improved surgical techniques as the years go by. A typical example is transposition of the great arteries. In the current population of adults, there are many who underwent atrial switch operations and common late complications that need taking care of are, arrhythmias, failure of the systemic right ventricle, and baffle obstruction or leakage. However, adolescents and young adults who underwent arterial switch operations have a markedly different spectrum of late complications which include pulmonary stenoses, dilation of the neo-aorta with aortic regurgitation, and problems with the coronary arteries.

Although paediatric care of CHD has become very successful and enables the vast majority of patients to survive into adulthood, most defects cannot be cured and special efforts are required to maintain the good results of paediatric cardiac surgery. So far, a significant morbidity and excessive mortality are an issue, particularly in moderate and complex CHD. Optimal care with reduction of morbidity and mortality remains a special challenge.

In the current population of ACHD the vast majority of deaths are cardiac, related to CHD with only a few exceptions of milder lesions such as early corrected atrial septal defect and patent ductus arteriosus. The leading causes of death are heart failure, sudden death (mainly due to arrhythmia), and operative death at the time of surgical re-intervention.

It is possible that early detection of residual defects and sequelae with their potential for late complications, followed by timely intervention and re-intervention, can reduce late morbidity and mortality. However, this will require highly specialized care.

Besides surgical and medical problems directly related to the cardiac defect, it has become widely recognized that ACHD patients who have other medical issues (pregnancy, non-cardiac surgery, etc.) require special integrated care, as well as care for psycho-social issues. This has led to a general agreement that ACHD care requires superregional specialist centres and that special training programmes need to be established for the physicians and health professionals involved.

The ESC published practice guidelines for the management of GUCH in 2010 aiming to improve the care of these patients. However, due to space constraint, the document did not include recommendations for the organization of care for adults with congenital heart disease, nor for the training in the subspecialty of GUCH, although emphasis was given to their importance and their need, in order to improve the long-term outcome for congenital heart disease.

The current publication now makes recommendations for the standards and organization of care for adults with CHD, as well as for training in GUCH in Europe.

There is an ongoing discussion about who should primarily take care of this patient group—paediatric or adult cardiology—and where it should take place, in a paediatric or adult setting.

Although cardiology primarily started with structural heart disease—congenital and acquired valve problems—adult cardiology has developed along a completely different direction over the decades. The main focus has changed to coronary artery disease, arrhythmias, and heart failure. The adult cardiologist has therefore become more and more unfamiliar with CHD.

The new insights gained in paediatric cardiology and the emerging problems of patients with operations not seen in the past have led to a lack of knowledge in the spectrum of ACHD and its late complications that we are now faced with. The interest in late outcomes and maintaining the successful treatment accomplished during childhood, together with the impression that adult cardiology may not take proper care of these patients, as well as the trend of decreasing numbers of children with CHD, has led to the integration of ACHD care into paediatric cardiology departments in some countries.

However, it appears that neither paediatric nor adult cardiology departments are most appropriate for the care of ACHD *per se*. The gap of knowledge in congenital heart disease—clinically, imaging and intervention—may be the problem of the average adult department. On the other hand, paediatric cardiology does not cover the needs of adults with congenital heart disease well. Long-term problems of the cardiac defects encountered during adult life differ from those seen in children.

Particular patient groups such as transposition of the great arteries with atrial switch or Eisenmenger patients are no longer or, only rarely seen in the paediatric population, whereas adult patients just require adult medicine. Paediatricians are unfamiliar with pregnancy pre-evaluation and care, with adult cardiovascular disease such as coronary artery disease, systemic hypertension, etc. and in particular, with adult co-morbidities. Therefore, it has been generally accepted that ACHD cardiology is a new specialty in addition to both, adult and paediatric cardiology. The document makes clear that trainees may come from either training track but need additional specific training in this new specialty as well as additional knowledge, in paediatric cardiology for adult cardiologists and adult cardiology and adult medicine in general for paediatric cardiologists.

Similarly, the document clarifies that superregional centres of excellence should not be simply programmes within paediatric or adult cardiology departments but have their own infrastructure that guarantees interdisciplinary collaboration.

If this document helps to convince governments and hospital administrations to establish such centres and to train physicians in the appropriate programmes, a major step towards improving the outcome for adults with congenital heart disease will have been accomplished.



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# GUCH/ACHD: what is required of the newest field in medicine?

**Cardiologists from the USA provide information from earlier experiences in this emerging specialty**



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(credit Sam Rogers)**



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The rate of growth of patients with grown-up congenital heart disease (GUCH)/adult congenital heart disease (ACHD) reaching adulthood is rapidly increasing, as advances in surgical and interventional techniques in infants and children have led to increased long-term survival.<sup>1</sup> Yet, most practitioners and health-care systems are currently poorly equipped to care for this increasingly complex patient population. Most adult cardiologists have little training and experience in congenital heart disease. On the other hand, paediatric cardiologists may have a thorough understanding of the anatomy and pathophysiology of congenital heart disease but limited knowledge of medical comorbidities associated with adulthood. These knowledge gaps highlight the need for developing formal training in GUCH/ACHD as the number of these patients grows.

As of 2010, there were an estimated 1.8 million GUCH/ACHD patients in Europe.<sup>2</sup> Additionally, the composition of this high-risk population continues to evolve so that we are seeing a general increase in the complexity of these patients along with its size. Thus, the challenges to developing adequate systems of care and training of physicians in the field of GUCH/ACHD are immense and it is vital that we meet these needs. This edition of the *European Heart Journal* includes an important position paper by the working group on GUCH of the European Society of Cardiology, which addresses both of these issues.<sup>3</sup> The recommendations of Baumgartner *et al.* related to the organization of care for adults with congenital heart disease build upon recommendations by previous groups, including The 32nd Bethesda Conference of 2000, and the American College of Cardiology/American Heart Association 2008 Guidelines for the Management of ACHD.<sup>4,5</sup>

At the core of these recommendations is the development of supra-regional 'Centres of Excellence' to provide comprehensive, expert care for GUCH/ACHD patients. It is recommended that such centres would aim to serve a general population of ~5–10 million people. However, despite such expert panel recommendations, care in many parts of the world remains compartmentalized for this population. For instance, in the USA the pattern of health-care systems addressing GUCH/ACHD involves a few, high-volume experienced centres and a large number of small- to medium-sized programmes. The most recent clinic directory of the Adult

Congenital Heart Association includes 112 programmes in North America with a median self-reported programme size of 463 visits per year (range 4–3772).<sup>6</sup> The geographic distribution of these programmes is also not uniform, with the New York City Metropolitan area alone hosting seven programmes, ranging in size from 38 to 2010 visits per year, while many other regions may be underserved.

At odds with the concept of a limited number of highly specialized centres, is the reality that many patients prefer or are unable to travel far from home to receive care at a regional centre. Less important (although still a fact of life) is the perception by some centres that there is value in keeping as many patients and procedures at their own institution. Of course, the optimal number of specialty GUCH/ACHD centres has not yet been defined and is arguably controversial. A more recent study suggests that the optimal ratio may be one centre per 2 million people in the general population.<sup>7</sup> Based on this ratio, we would anticipate the need for 150 centres in the USA, 17 centres in Canada, and 365 centres in Europe. These estimates suggest there might even be more of a shortage of adequately trained physicians for GUCH/ACHD patients than previously thought. Formalization of accreditation processes for GUCH/ACHD centres, certification of providers and pressure from payers, governments, and advocacy groups, may be required to further regionalize the care of these patients and spur the development of systems of care and further training.

It is timely to see the development of more formal training requirements for GUCH/ACHD from the European Society of Cardiology, which parallels a similar process in the USA. In December of 2012, the American Board of Medical Specialties announced the recognition of ACHD as the newest American subspecialty, with the development of training requirements which will lead to Board eligibility soon to be published by the American Council of Graduate Medical Education (ACGME). The European and American training requirements are overall similar in the structure of the recommended training programme. Both allow for GUCH/ACHD training pathways for those trained in either in paediatric or adult cardiology.

The creation of minimum criteria for trainee interpretation of studies and procedures tends to be relatively straightforward, as experts in a field have a sense for the minimum requirement for competence to practice independently. It is somewhat more difficult and sometimes more contentious to develop minimal criteria for patient care numbers, as it is more difficult to assess competency of trainees in their expertise of care for the entire gamut of GUCH/ACHD care. Both proposals recommend involvement with  $\geq 200$  inpatient admissions (difficult for all but a few centres in the USA), and  $\sim 400$  outpatient clinic visits.<sup>8</sup> While the European proposal recommends trainees perform  $\geq 50$  ACHD transoesophageal echocardiograms (TOEs), the American Board of Internal Medicine petition recommends a less stringent number of 25 TOEs. These minimal numbers will inherently limit training programmes to high-volume

centres, and potentially limit adequate numbers of trained ACHD providers to meet the rapidly growing need.

It is interesting to note that both the European Society's and American's ratio of minimum required outpatient visits to inpatient exposure is relatively low, at a ratio of just slightly  $>2$  to 1. This ratio is contrary to the trend, at least in the USA, to training in other fields such as, Internal Medicine and Paediatrics, where increasing emphasis has been placed on outpatient care. While cardiology-training guidelines in the USA have not paralleled this trend, we would argue that GUCH/ACHD care probably should. The large number of different cardiac lesions seen by congenital cardiologists, as well as the significant heterogeneity in the clinical status of these patients makes it apparent that a larger number of outpatient visits may be needed for trainees to be exposed to the entire spectrum of GUCH/ACHD patients. As indications for admission usually fall into a limited number of categories (including heart failure or arrhythmia), the requirement for a minimum of 200 inpatient evaluations may be somewhat higher than necessary to establish competency for the trainee.

A critical barrier to achieving such a goal at least in the many parts of the USA, is the distance many patients live from such tertiary care centres. While most of the surgical care can and should occur at the centre of expertise, many medical admissions will remain at the local hospitals closer to the patient's home and family, ideally with input from the GUCH/ACHD centre via long distance consultation. Novel innovations in telemedicine may be uniquely suited to the role of supporting remote centres in the care of GUCH/ACHD patients as it has been in other areas of medicine. Transfers would occur only when advanced therapies or studies are needed. Hence the critical number of admissions during the 2 years is unlikely to be achieved in all but a few of the largest training centres, leading to a paucity of cardiologists completing GUCH/ACHD training when compared with the rapidly growing need.

Baumgartner *et al.* discuss important and timely aspects for the care of GUCH/ACHD patients. Formalization of the organization of care and training for these patients will be vitally important as the population continues to increase in size and complexity. This will require this newest field of medicine to mature on its own so as to meet the challenges that are certain to be raised, as we all strive towards optimizing care in GUCH/ACHD patients in both North America and Europe as well as worldwide.

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# Landmarks in the history of Cardiology I

## From pre-historic times to Galen's medical authority

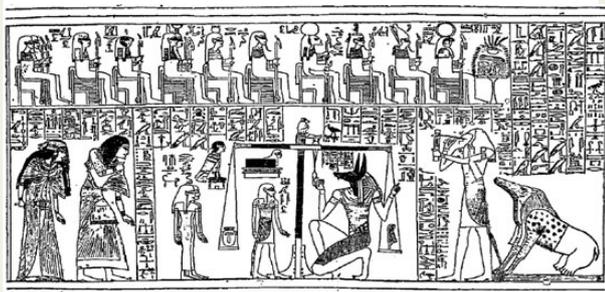


**Figure 1** The Pindal elephant.

In 10 000 BC a pre-historic man in Pindal, Spain, painted an elephant on the wall of a cave accurately depicting the position of the animal's heart (Figure 1). Similar illustrations from that period have been found in the Niaux Cave, France, where a bison is

painted with arrows embedded in its heart, demonstrating that early man recognized the vital importance of this organ.<sup>1</sup>

In ancient civilizations, the heart had not only a medical importance, but also a symbolic and spiritual one. Ancient Egyptians considered the heart as the seat of intelligence, thinking, and feeling.<sup>2</sup> Believing that the heart recorded all of the good and evil actions a person did while alive, it was the only organ left in the body during mummification. During the ritual of 'weighing of the heart' (Figure 2) a scarab was placed to secure the heart's silence concerning



**Figure 2** The 'weighing of the heart' ceremony.



**Figure 3** A Chinese physician feeling the pulse.

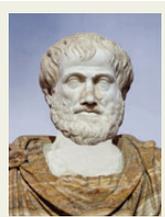
past transgressions. Anubis (the God of mummification) held a scale where the heart was weighed against the feather of Maat, the symbol of truth and eternity. If the heart was as light as the feather, it meant that the person would reach eternity. If it was heavier, the demon

Ammit would swallow the heart and the deceased would not have an afterlife.<sup>3</sup> Concepts and notions on the cardiovascular system were integrated in the Ebers papyrus dating back to 1500 BC. Without giving special attention to anatomy, ancient Egyptians did not know the difference between vessels, nerves, and tendons. The heart was linked to a number of vessels (*metu*) that were carrying among other things, air, blood, urine, and excrement. However, in several passages from Ebers, heart diseases were more or less identified as angina, heart failure, and ventricular fibrillation. Pulse was recognized as a result of the heart beating and feeling; the pulse was an important part of examination, but not as critical as it was for Ancient Chinese medicine, which placed the pulse in the centre of its nosology (*Figure 3*).<sup>2</sup>

Ancient Chinese tradition was opposed to autopsies and so, physicians' medical knowledge was mainly based on tradition and empiricism. The heart was considered the seat of emotions and the pulse was interpreted by the concept of 'yin-yang'. Chinese physicians described >200 pulse varieties and developed special techniques for pulse examination based on their perception of the pulse wave at the radial artery.<sup>4</sup>

Medicine and cardiology received an important boost, because of ancient Greek thinking and philosophy, which provided a rational approach to medicine, disconnecting it from religion and superstition.

The eminent philosopher Aristotle (384–322 BC) identified the heart as the most important organ of the body. He believed that it was the first organ formed during embryonic life and the last to stop working. It was connected with all sense organs and muscles via the blood vessels and it produced the body's heat. He interpreted the heart's



pulsation as the result of a boiling movement in the blood, which caused it to press against the walls of the heart and erroneously concluded that the heart was a three-chambered organ. Aristotle considered the heart the organ of intelligence, establishing a cardio-centric model that survived well into the Middle Ages.<sup>5</sup>

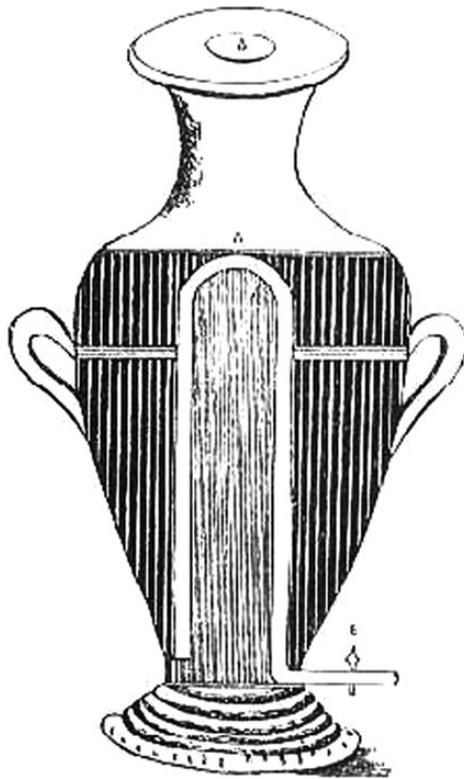
In the Hippocratic corpus several passages on cardiac anatomy, disease, and treatment are mentioned. The position of the heart and the anatomical relationships of the ventricles in the body were accurately described, the independent contraction of the atria was noted, and cardiac valves were identified. Physical examination of the chest, auscultation, and vascular pulsation were mentioned. The succussion splash was noted in empyema and in some passages a pleural friction rub is indicated. Clubbing fingers were described and pitting oedema was distinguished from the indurate oedema of local inflammation. The association of dyspnoea in heart disorders was pointed out, while an excellent description of congestive heart failure with cardiac cirrhosis was provided. Clinical descriptions recognizable nowadays as Cheyne–Stokes respiration, angina, myocardial infarction, and Stokes–Adams syndrome are identified. Wounds of the heart were known and thoracocentesis was performed. Phlebotomy was recommended in the treatment of dropsy along with mild exercise and a dietary regimen. Finally, obesity-related risk was noticed: 'Those who are constitutionally very fat are more apt to die quickly than those who are thin'.<sup>6</sup>

The high standards and value of medicine that were established by Hippocrates and his followers are reflected in the work of Herophilus of Chalcedon (350–280 BC) and Erasistratus of Ceos (304–250 BC), members of the Alexandrian School of Medicine who contributed to the study of cardiovascular anatomy by performing several vivisections. Herophilus supported the fact that arteries carried blood and not air, as was believed. He also studied the body's pulse rate and, using a 'water clock', developed methods to document pulse strength and rhythm (*Figure 4*). Erasistratus mentioned that blood in the body must move similarly to the way sap moves in trees. He mapped the veins and arteries and concluded that the heart functioned like a pump that moves blood.<sup>7</sup> Unfortunately, only fragments of their writings survived, which do not allow us to assert with certainty that blood circulation was understood, but neither do they allow one to conclude the opposite.



In the second century AD, Galen (130–201), one of the most outstanding physicians of antiquity, contributed to the knowledge of the cardiovascular system. He recognized that the heart pumped blood through the arteries and demonstrated the importance of heart valves in preventing blood from flowing backwards. As his experiments were based on animals, some of Galen's conclusions were erroneous: his blood circulation theory was based on the notion that venous blood was passing through invisible pores in the heart's septum, was moving from the right ventricle to the left, and was mixed with inhaled air from the lungs.<sup>8,9</sup>

Galen's writings dominated medieval medicine, taking on an almost religious sacredness. Even though medical practice was highly regarded, anatomy being related with the dead was considered immoral and non-religious. It was the period when experimental studies on anatomy and physiology ceased and Galen's blood



**Figure 4** Herophilus' water clock.

circulation model was passed on to successive generations of physicians for almost 1500 years.



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