World Report

When blue babies grow up

Thanks to advances in surgery, thousands of babies born with heart defects now grow up to live near-normal lives. But these survivors have unique medical needs for which most doctors are unprepared. Wendy Wolfson learns what happens to "blue babies" when they become adults.

Sara L, born in 1967, was a "blue baby" with transposition of the great vessels in her heart. At 3 days old, she had a Blalock Shunt at the University of Minnesota. When she turned 4, she went back for a Mustard procedure. Her lips and fingertips turned from purple to pink. But at 20, she started experiencing unpredictable bouts of arrhythmia that could only be treated by being electrocardioverted in the hospital emergency room.

Sara L got a pacemaker implanted and began taking cardiac drugs. "It was really scary", she recalls. "I had been brought up thinking that I was like everybody else, plus a few scars. Suddenly I had to think about how I was going to live my life . . . If I could live my life."

Happily, Sara fell in love, married, and is now the doting mother of a healthy baby. But having a child was huge gamble. "My husband left [the decision] up to me", explains Sara. "He said, 'this is your body and you will have to live with the consequences.' I was the one who pushed."

In the 1960s and 1970s, improvements in cardiac surgery and treatment enabled a new generation of children born with cardiac abnormalities to live to adulthood. Sara L is one of the estimated 750 000- 1 million young adults in the USA who had childhood open-heart surgery. While there are more than 35 classifications of heart defects, adults with heart abnormalities are divided between those whose illness impedes their daily life, and are generally be under the supervision of a cardiologist, and a deceptively healthy second group, who have undergone

childhood repairs but who have reached their 20s without apparent symptoms.

"There is a shortage of specialised care --we are endangered by our own lack of knowledge and lack of expertise", says Amy Verstappen, president of the Adult Congenital Heart Association (ACHA) an advocacy group set up by heart patients. "Our outcomes are excellent, but we have to be in ongoing appropriate care."

Heart patients who appear healthy and perceive themselves as 'fixed' may not be under the care of a cardiologist at all, says Verstappen. But sometimes, as in Sara's case, the heart's function changes. Changes may be imperceptible at first and can only be picked up by a cardiac specialist. A heart patient can miss "a window of opportunity", cautions Verstappen. "By the time you feel your cardiac symptoms you may have missed your opportunity for care. Rhythm problems could be a symptom that the anatomy is changing. There should be somebody looking at the structure and the electrophysiology because a heart with complex defects usually has funky wiring."

ACHA provides an online forum for heart patients all over the world. Although giving medical advice is strictly discouraged, connecting with others who have similar life histories is a revelation for many who grew up thinking that they were unique. Tips are exchanged on finding swimsuits that don't show the scars of open-heart surgery and where to find specialists.

Yet getting care is often not a straightforward process because specialists who are familiar with both paediatric and adult issues are unlikely to be located in many major urban centres.

These patients fall into a gap between paediatric and adult cardiology. Adult cardiac doctors are more attuned to issues of acquired heart disease, such as arteriosclerosis. Paediatric cardiac doctors may not be able to deal with the issues that adult patients face. Sara L stayed in New York City because she could find the specialised medical expertise she needed.

"As a person who has lived through all these medical treatments, I have a pretty clear understanding of my body", says Sara L. "It is very frustrating and frightening to go to a doctor and not have him understand the type of care I need. For us, the children with these type of defects, our medical treatment is often judgment-based. A lot of cardiologists have no concept of how our bodies work or what can affect us."

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Ensuring health

Lack of insurance cover is a particular problem for US patients with pre-existing conditions. In the USA, insurance is linked to employment. Younger patients may not have insurance, because they grow out of their parent's coverage by their early 20s, often when their heart function starts changing. "I had to make sure I always had a job, or stayed at jobs that drained me physically", says Sara L. "When I was at school, I applied for Medicaid and I was told by the intake clerk that if I was 16 and pregnant, I could get emergency Medicaid, but I just needed to go to a doctor to get my pacemaker checked."

Patient advocacy groups like ACHA and the International Society of Adult Congenital Cardiac Disease (ISACCD) lobby for improved care and education for patients and doctors. They organise conferences and refer patients to qualified doctors and reliable information. The American College of Cardiology's 32nd Bethesda Conference in 2001 published a framework of guidelines for care of adults with congenital heart disease. They cover issues including portability of insurance and accessibility of medical records, follow-up, medications and side-effects, preventing heart infections, exercise, birth control, career planning, insurance, dental care, and symptoms.

A model is being advocated of linking together existing medical centres into a network of regional centres of expertise that primary-care physicians can go to for guidance. This system needs a filtering mechanism to maximise visits for the most ill, but helps to centralise expertise for healthier heart patients.

Adults face not only the lingering effects of their childhood heart defects, but also the additional problems of adult-onset acquired heart disease. As congenital heart disease is transformed into a chronic condition, heart patients have to deal with a host of lifestyle issues that include staying healthy, day-to-day work, romance, and having children.

Little is known about the long-term effects of pregnancy, but an increasing number of women with congenital heart defects are having children. Deborah Gerasony, of Columbia Presbyterian Hospital in New York, has launched a study to collect information on women who have children to determine the effects of pregnancy and childbirth.

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An international problem

In the UK, Michael Gatzoulis, director of the Adult Congenital Heart Programme, Royal Brompton Hospital, London, says that congenital heart defects are "the most common inborn defect that exist".

Gatzoulis advocates a preventive approach which includes addressing patient lifestyle issues, such as investigating whether it is safe for an individual to be pregnant, and not just cardiology advice.

He emphasises that patients must be proactive. "Patients should know as much about their condition as they can", says Gatzoulis. He recommends patients see a cardiologist, avoid risk factors for acquired heart disease, exercise, look after their teeth to avoid bacterial infections, know the risks of pregnancy, and keep copies of their medical files.

"While insurance in the UK is not a problem there are not enough centres, not enough resources, and not enough trained cardiologists. Both the profession and public need a lot of education.

Most patients in the UK are lost to follow-up. Gatzoulis estimates that there are about 200 000 congenital heart patients in the UK. "We see up about 5500 patients in Brompton. There are other centres and they see about 5000-10000 for the rest of the UK. For the rest, who do they see? Undoubtedly they do not have good outcomes", he says. "They need regular assessment. There are a spectrum of lesions. No patient is the same. Even with the same lesion the patient is different. A significant number of patients would benefit from surgery and drug therapy."

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Learning fast

In the 1960s, surgeons developed new techniques of open heart surgery, says Michael Landzberg, director of the Boston Adult Congenital Heart group (BACH) centre at Children's Hospital in Boston. Students of the early pioneers went on to set up medical departments of their own and, as the discipline has started to mature, this expansion has continued.

Medical centres focusing on adult congenital care have now been set up in the USA in Boston, New York, San Francisco, Stanford, Los Angeles, and Minnesota; there are also centres in Canada and the UK. However, says Landzberg, the field has only really been established in the past 4 years. "We haven't done a lot", he says. Groups like the American College of Cardiology, and National Institutes of Health will have meetings in September to talk about future guidelines for management of individuals who survive neonatal heart problems. A conference on the issue is also planned for this autumn in the UK.

BACH runs a monthly support group for heart patients. Jonas Bromberg, who has led the group since 1996, says adult heart patients often feel very isolated. "Most patients had never met somebody who had a congenital heart defect", he explains. Groups like this help affected individuals "connect with other folks like them", he says.

"The typical person who has a congenital heart disease is an achiever." says Landzberg. "They have dealt with significant issues and have won."

Wendy Wolfson

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