



# Early detection of and intervention for two newborns with critical congenital heart disease using a specialized device as part of a screening system

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## Abstract

Screening for critical congenital heart disease is a clinical method used for their early detection using pulse oximetry technology. This, followed by a diagnostic confirmatory protocol, allows timely therapeutic interventions that improve the newborn's outcome. According to Mexican birth statistics, approximately 18,000–21,000 neonates are born with a form of congenital heart disease each year, of which 25% are estimated to be critical congenital heart disease. We report two cases with an early critical congenital heart disease detection and intervention through an innovative critical congenital heart disease screening program implemented in two Mexican hospitals. They integrated a new automated pulse oximetry data analysis method and a comprehensive follow-up system (Cárdi-k®). Both cases were confirmed by echocardiogram, which served for an intervention in the first week of life, and the patients were discharged in good clinical condition. In addition, to the routine physical assessments, the critical congenital heart disease screening program (which includes echocardiogram for presumptive positive cases) should be implemented in a timely manner.

## Keywords

Cardiovascular, critical congenital heart disease, pulse oximetry, newborn screening

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## Introduction

Congenital heart disease (CHD) is the most common congenital malformation, being present in 1% of all U.S. national births and in 2.1–12.3 per 1,000 newborns worldwide.<sup>1,2</sup> No data are available to indicate the prevalence of CHD in Mexico; however, it is recorded as the second leading cause of death in children younger than 5 years of age. According to Mexican birth statistics, approximately 18,000–21,000 neonates are born annually with CHD,<sup>2</sup> and approximately 25% of them are born with a critical condition. Critical congenital heart disease (CCHD) is a condition that requires early therapeutic intervention.

CCHD screening is a clinical method used to detect CCHD using pulse oximetry (POX) technology, which measures peripheral capillary oxygen saturation (SpO<sub>2</sub>). In the United States, the Secretary of Health and Human Service's Advisory Committee on Heritable Disorders in

Newborns and Children (SACHDNC) established seven pathologies as the primary targets for CCHD screening.<sup>3</sup> Hypoxemia is usually present in newborns with those CCHD

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(e.g. in some cases, patients present with cyanosis), which occurs when both systemic and venous circulations are mixed.<sup>4</sup> Negative screening for POX does not exclude the presence of heart disease.

The introduction of CCHD screening by POX would prevent 4–6.5 deaths per 100,000 newborns worldwide.<sup>1</sup> These deaths currently occur in an apparently absence of clinical signs, as mild cyanosis is not visible to the human eye. Therefore, physicians should consider POX as a screening method that helps in closing the diagnostic gap, improving overall sensitivity when done in conjunction to fetal ultrasound and physical assessment in the well-baby nursery (WBN).

It is well-known that in most screening programs, POX readings are operator-dependent (i.e. the readings are visually obtained and typically handwritten), and there is an absence of technological links that automate the patient follow-up process for presumptive positive cases. Consequently, the methodology and results employed vary among health care institutions.

Here, we report two newborns who were detected to have CCHD, as determined by *Cárdi-k®*, a CCHD program implemented in two Mexican hospitals (Christus Muguerza Hospital Conchita (Monterrey, Mexico) and Hospital Médica Sur Lomas (CDMX, Mexico)) where these patients were born. This shows how a comprehensive screening program, complemented with good-quality pre- and postnatal care, could improve the outcomes of a newborn with CCHD.

## Case report

The CCHD screening program (*Cárdi-k®*) comprehended the detection and diagnosis phases for targeted CCHD. The first stage included POX screening performed by WBN staff measuring pre- and postductal SpO<sub>2</sub>. A specialized device was used for the screening, as described below. The automated interpretation followed the recommendations of the American Academy of Pediatrics (AAP).<sup>5</sup> For suspected newborn, a protocol was established in each hospital, where the WBN staff immediately notified on-call physician triggering pediatric and cardiologic evaluation procedures. Complete clinical examination and two-dimensional transthoracic echocardiography scan with pulsed-wave and color-flow Doppler imaging were performed for those presumptive positive newborns.<sup>6,7</sup> Afterwards, a trained team member communicated the findings to the patient's parents and, if indicated and authorized, the intervention protocol continued.

The main features of the computerized device—based on POX technology—include the following features: (1) to conduct simultaneous pre- and postductal SpO<sub>2</sub> readings; (2) to perform operator-independent data analysis, where statistical evaluations are performed in real time; (3) to generate immediate results and recommendations according to a standardized protocol that was saved and managed in a

central database; and (4) to receive automated system alerts (i.e. e-mails) that are sent directly to cardiologists, the CCHD screening program coordinator, and the newborn nursery director to perform confirmatory evaluations in presumptive positive cases.

### Newborn 1

This male newborn was born from a first gestational 31-year-old mother diagnosed with and treated for type II diabetes, hypertriglyceridemia, preeclampsia, and hypothyroidism who had a complete prenatal care. An urgent cesarean section at 35.5 weeks of gestational age (GA) took place due to fetal distress; the newborn's length was 47 cm (p66), weighed 2.5 kg (p51), and had Apgar scores of 9 and 10 at 1 and 5 min, respectively. The pediatrician considered this newborn to be healthy and he was subsequently transferred to the WBN.

At the nursery, our CCHD screening was performed at 24h of birth, where the simultaneous SpO<sub>2</sub> levels were reported to be 88% on the right hand and 87% on the right foot. Both the device and the system directly alerted the personnel responsible for the screening program about these levels.

According to the protocol, the patient was assessed by pediatric cardiology and an echocardiogram was performed, where the newborn was diagnosed with critical pulmonary valve stenosis, coarctation of the aorta, and patent ductus arteriosus. The patient was transferred to a tertiary referral hospital for diagnostic and interventional catheterization, as well as for left thoracotomy where coarctectomy, aortic valve plasty, and ligation of a patent ductus arteriosus were performed 2 days after detection. There were no complications. Post-interventional evolution was satisfactory.

### Newborn 2

This male newborn was born from a third gestational 31 year-old mother (one delivery, one abortion) diagnosed with and treated for preeclampsia who had a complete prenatal care. He was born at 38.6 weeks GA by cesarean section due to pelvic presentation; the newborn's length was 49 cm (p45), weighed 3.425 kg (p72), and had Apgar scores of 9 and 9 at 1 and 5 min, respectively. While in the WBN, he presented with progressive distal cyanosis.

CCHD screening was performed, where SpO<sub>2</sub> levels of 50% on the right hand and 49% on the right foot were obtained. Following the physical assessment and the POX measurement, the newborn was immediately transferred to the neonatal intensive care unit (NICU). As in the previous case, both the device and system immediately alerted the personnel responsible for the screening program of this event.

A transthoracic echocardiogram was performed, which diagnosed the presence of critical pulmonary valve stenosis and a dysplastic pulmonary valve with anterograde flow not

assessed. Valvular opening was not observed. Confluent pulmonary artery branches were present. There was a patent ductus arteriosus of 5 mm with a cross gradient of 36 mm Hg. In addition, an atrial septal defect (ASD) of 9 mm with a right-to-left shunt and left septum deviation were evident. The right atrium was dilated, and there was severe tricuspid regurgitation (TR) with a Coanda effect. Furthermore, there was a small hypertrophic right ventricle, without sinusoids, with an intraventricular pressure of 99 mmHg. No ventricular septal defects were assessed. Left ventricular systolic function was preserved with a left ventricular ejection fraction (LVEF) of 59%, as determined by Simpson biplane, and with a shortening fraction of 40%. The left ventricular outflow tract was unobstructed. The pulmonary ring was of adequate size ( $Z=-0.7$ ).

The patient was transferred to a tertiary referral hospital for diagnostic and interventional catheterization, where pulmonary valvuloplasty with a balloon was performed 2 days after detection, once optimal conditions were achieved. No complications occurred during the procedure. The post-interventional evolution was satisfactory, and echocardiographic control revealed a pulmonary valve gradient of 4 mm Hg, an ASD measuring 6 mm with a predominant left-to-right shunt, a dysplastic tricuspid valve with severe insufficiency, moderate pulmonary insufficiency, a small hypertrophic right ventricle with diastolic dysfunction, an LVEF of 67%, and a small ductus arteriosus.

The patient was discharged with captopril and oxygen-free. During a 10-month clinical and echocardiographic follow-up to assess right ventricle function, it was evident that the tricuspid regurgitation improved with medication. The newborn's present condition is currently being maintained with hemodynamic stability under follow-up by pediatric cardiology.

The ethics approval was provided by the Ethics Committee ("Comité de Ética en Investigación de Christus Muguerza, Sistemas Hospitalarios, S.A. de C.V.") with the approval number CONBIOETICA19CEI00420170601. Furthermore, the legally authorized representatives provided written informed consent for patients' information to be published. We have obtained the necessary permission from the respective copyright holder to use all applicable tools in this report.

## Discussion

Both newborns had outstanding outcomes as a result of the actions of the hospitals' clinical staff, as well as in response to the prompt interventional catheterization that was performed. The physical assessment and POX screening that are routinely carried out in the WBN increase CCHD detection sensitivity.<sup>6,8-11</sup> Particularly, despite the low SpO<sub>2</sub> expected for the second newborn, this program allowed him to had a timely diagnostic approach and intervention as a result of the prompt notifications to the medical team and protocol put in place.

In relation to the POX routine performed in a screening program, there are technical limitations associated, such as the need for clinical staff members' subjectivity when assessing test performance and the lack of reliability during manual data transfer.<sup>12</sup> Based on previous reports,<sup>10,12</sup> the innovative nature of Cárdi-k®'s screening device may significantly reduce human error and increase confidence in the results. In addition, it efficiently communicated several results through a central database system, leading to rapid responses.

## Conclusion

To our knowledge, there is an absence of CCHD screening programs in Mexico. This study demonstrates strong evidence of potential benefits by two well-implemented CCHD programs as routine procedures before discharge. By extending this shared practice of employing technology as part of a comprehensive screening program, and implementing protocols for presumed positive and confirmed patients, clinicians and specialists will be able to provide their patients adequate and timely treatment.

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## Declaration of conflicting interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

## Ethical approval

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## Informed consent

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