Göteborg, 2023

Early detection of critical congenital heart defects in Sweden

- with a focus on coarctation of the aorta

Akademisk avhandling som för avläggande av medicine doktorsexamen vid Sahlgrenska akademin, Göteborgs universitet kommer att offentligen försvaras i föreläsningssal Tallen, Drottning Silvias Barnsjukhus Göteborg,

fredagen den 16:e juni klockan 9.00

av

Katarina Lannering

Fakultetsopponent:

Henrik Holmström (professor)

Institutt for Klinisk Medisin, Rikshospitalet, Oslo, Norge

Avhandlingen baseras på följande delarbeten

I. Lannering Katarina, Bartos Marie, Mellander Mats. **Late Diagnosis of Coarctation Despite Prenatal Ultrasound and Postnatal Pulse Oximetry.** *Pediatrics 2015 Aug Vol 136 (2) e406-e412.*

II. Lannering Katarina, Elfvin Anders, Mellander Mats. **Low false-positive rate of perfusion index as a screening tool for neonatal aortic coarctation.** *Acta Paediatrica 2020 Nov Vol 110 (6) p.1788-1794*

III. Lannering Katarina, Kazamia Kalliopi, Bergman Gunnar, Liuba Petru, Östman-Smith Ingegerd, Alenius Dahlqvist Jenny, Elfvin Anders, Mellander Mats. **Screening for critical congenital heart defects in Sweden. A retrospective national cohort study 2014-2019.** *Under revision*

IV. Lannering Katarina, Östman-Smith Ingegerd, Mellander Mats. **Enhancing neonatal screening for aortic arch obstructive lesions: the added value of using perfusion index.** *Manuscript*

SAHLGRENSKA AKADEMIN
INSTITUTIONEN FÖR KLINISKA VETENSKAPER

GÖTEBORGS UNIVERSITET

Göteborg, 2023

Early detection of critical congenital heart defects in Sweden - with a focus on coarctation of the aorta

Katarina Lannering, Avdelning för pediatrik, Institutionen för kliniska vetenskaper, Sahlgrenska akademin, Göteborgs universitet, Sverige, 2023.

Abstract

Introduction: Despite the implementation of prenatal and neonatal screening, newborn infants with critical congenital heart defects (CCHD) remain at risk of being discharged undiagnosed, particularly newborns with coarcation of the aorta (CoA). Recent studies have suggested that perfusion index (PI) may potentially identify additional cases of CoA.

Aims: To investigate the relative contributions of prenatal screening, pulse oximetry screening (POS) and newborn physical examination (NPE) to the early detection of CCHD with special attention to CoA. A second aim was to investigate the use of PI to improve the early detection of CoA.

Methods: Retrospective population-based cohorts of isolated CoA (*Paper I*) and CCHD (*Paper III*) were studied with respect to the contribution of pre- and postnatal screening methods to early diagnosis. PI was measured prospectively in healthy newborns to determine the false positive rate (*Paper II*). The sensitivity of PI to detect aortic arch obstructions (AAO), such as CoA, was studied retrospectively in newborns with AAO who were routinely screened with PI (*Paper IV*).

Results: In *Paper I*, three of 90 with CoA were diagnosed prenatally. Among 87 diagnosed postnatally, 4/19 (21%) born in units using POS screened positive. Forty-six (53%) were discharged undiagnosed. At readmission, 22 were in circulatory failure and one died at home. In *Paper III*, 264/630 (42%) with CCHD were diagnosed prenatally, 142 (23%) by POS and 86 (14%) as a result of NPE. Although prenatal detection increased significantly during the study period, 4 newborns died undiagnosed before discharge and 64 (10%) were discharged undiagnosed. Upon readmission 24 were in circulatory failure with one preoperative death. Of 184 with CoA, 55 (30%) were discharged undiagnosed. In *Paper II*, the false-positive rate of PI in 463 newborns was reduced to 0% by using repeated PI measurements and a threshold of <0.7% for a positive screen. In *Paper IV*, the sensitivity of PI to detect AAO in 38 cases could be increased from 45% to 76% by combining PI in right hand with POS and NPE.

Conclusions: POS and NPE remain important for the early detection of CCHD, complementing the increasing prenatal detection. While the overall pre-discharge detection of CCHD was high, and improvements were made in the prenatal detection of CoA, this defect was still frequently not diagnosed before discharge. Adding PI to CCHD screening has the potential to further improve early detection of CoA but requires additional evaluation.

Keywords: Critical congenital heart defect, coarctation of the aorta, neonate, newborn, early detection, pulse oximetry screening, perfusion index, neonatal screening

ISBN 978-91-8069-297-7(TRYCK)
ISBN 978-91-8069-298-4 (PDF)