Catching Blue Babies: Critical Congenital Heart Defects in Newborns at Moderate Altitude

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and Genetic Testing Symposium

Pediatric Heart Defects

- Critical Congenital Heart Defects (CCHD)
  - 1 in every 4 newborns with a heart defect
  - 7,200 newborns in the US every year
  - Cardiac procedure within 1st year of life

Newborn Screening for CCHD

- CCHD symptoms:
  - Low oxygen saturations → cyanosis
  - Blue baby syndrome

- Screening using pulse oximetry technique (!!!)

CCHD Screening at Altitude

- Partial pressure of O_2 decreases with increasing altitude → physiological issue
- Screening algorithm based on studies at sea level → efficacy issue
- Limited information on newborns with CCHD at altitude and their screening results
- Secretary HHS requested additional studies

Research Questions

- What are pulse oximetry results of newborns with CCHD born at altitude?
  - Delineate pulse oximetry values
- How did they respond to screening algorithm?
  - Performance of AAP algorithm at altitude

Methodology

- Retrospective cohort of ***true positives***
  - Using data from medical charts to recreate screening scenario
- Term/near-term newborn, 7 dx of CCHD, treated at Children's, 2003 – 2013
  - i2b2 → 343 cases
  - No electronic chart, > 48-hr admission, < 24-hr deceased or had cardiac surgery
  - Descriptive statistics → 158 cases
  - Demography, risk factors, O_2 saturations at 24-, 28-, 36-, 48-hr, screening results

Demography

- Male → 64%
- White/Caucasian → 69%
- Non-Hispanic/Latino → 64%

78% from CO

- KS, MT, NE, NM, TX, WY
  - Transferred to Children's for higher care within 24-hr after birth

Risk Factors and Diagnosis

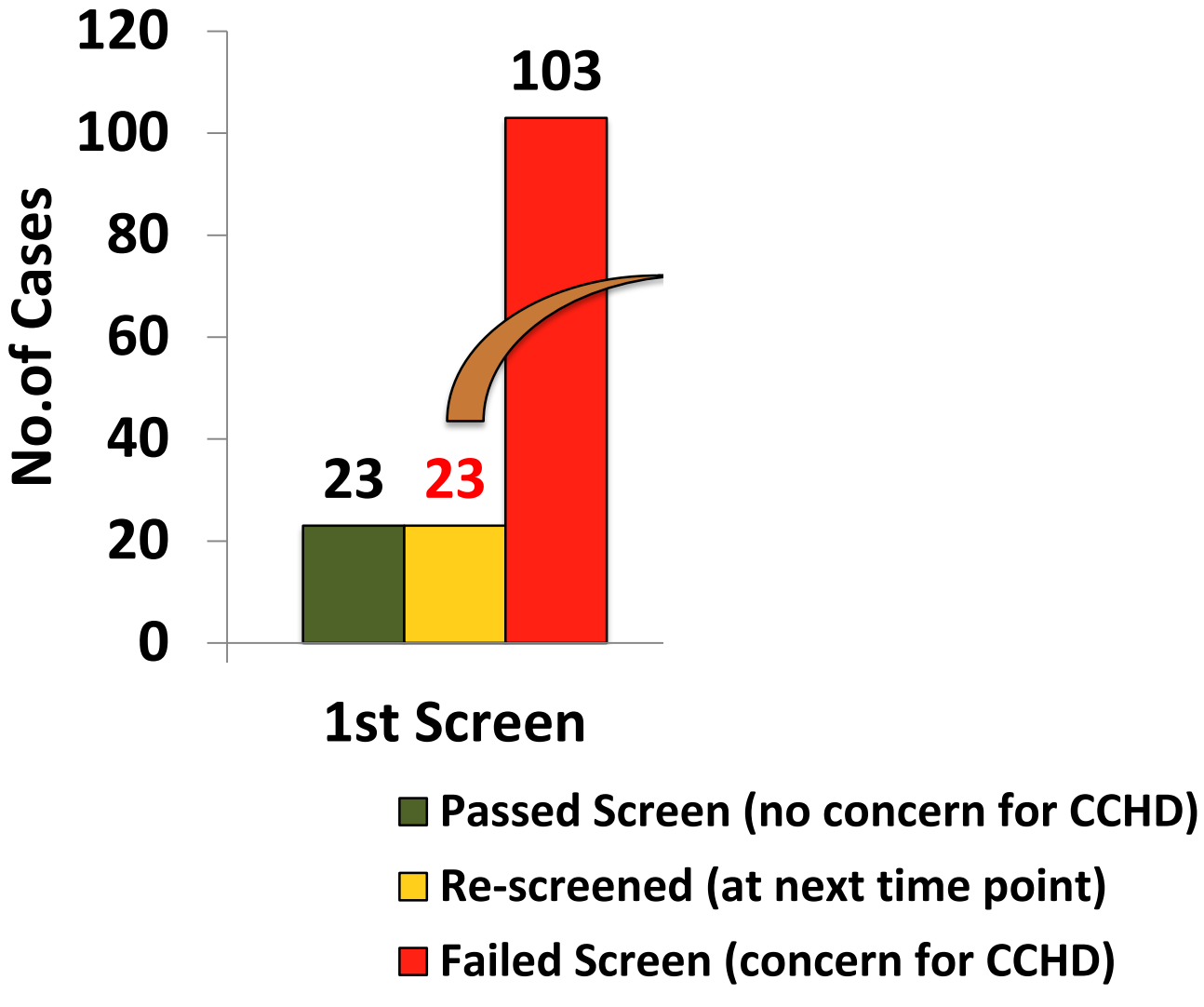
- Family history of CCHD, genetic disorder, and maternal diabetic status are ***not*** risk factors in our cases
- Top 3 diagnosis:
  - Hypoplastic Left Heart Syndrome (70%); Transposition of Great Arteries (30%); Tetralogy of Fallot (18%)
- Children's Hospital CO treated ~30 cases/yr

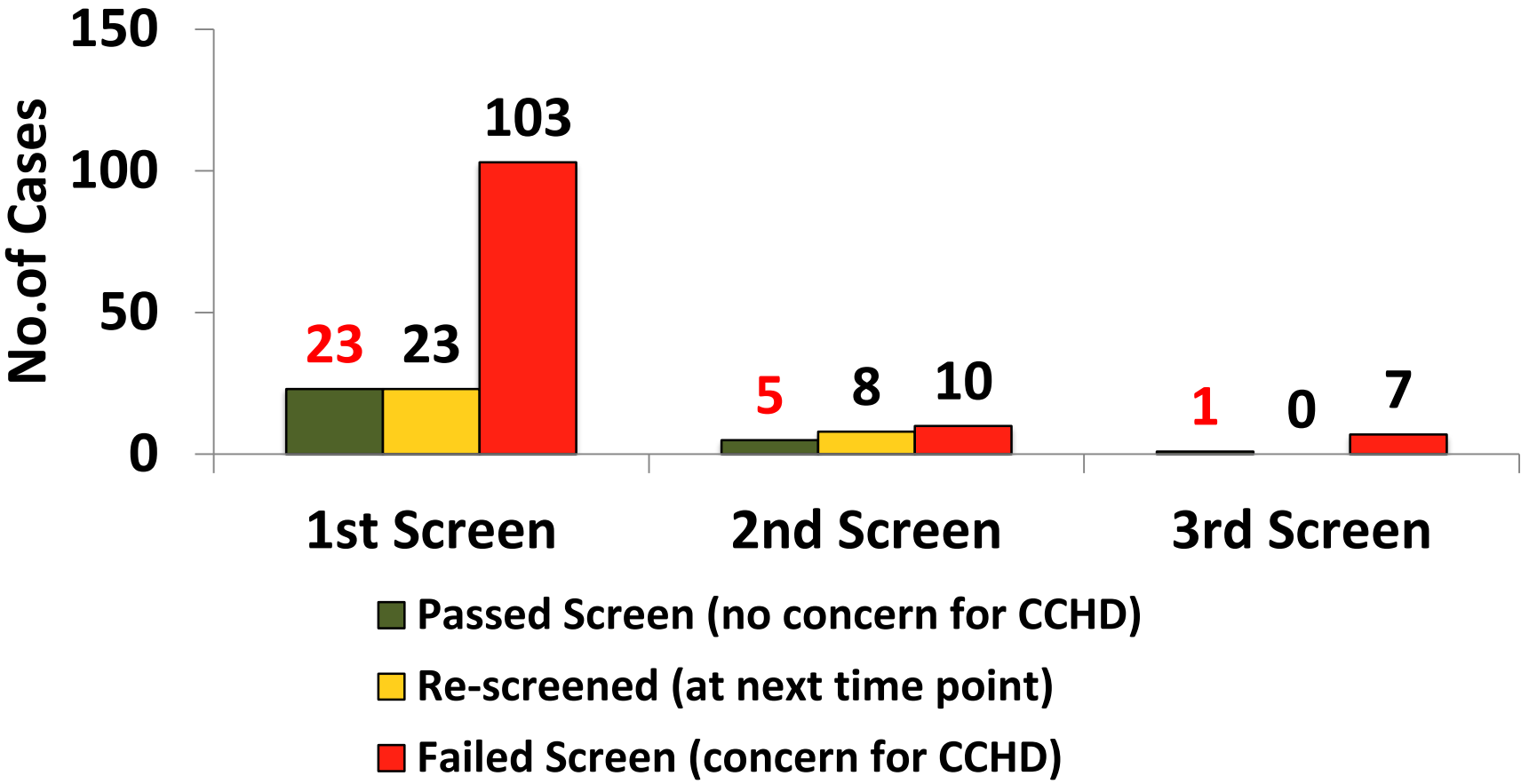
Comparative O_2 Saturation Values

Diagnosis	Altitude	Statistics	Preductal (%)	Postductal (%)
CCHD	Moderate	Average	87.1± 7.2	87.8 ± 6.3
CCHD	Moderate	Range	70.0 – 100.0	67.0 – 100.0
Healthy	Moderate	Average	97.2 ± 1.9	97.2 ± 2.1
Healthy	Moderate	Range	88.0 – 100.0	88.0 – 100.0
Healthy	Sea Level	Average	98.3 ± 1.4	98.9 ± 1.6
Healthy	Sea Level	Range	94.7 – 100.0	94.7 – 100.0

(Samuel et al., 2013, Acta Paediatrica) (Wright et al., 2014, Pediatrics)

Screening the Positives





- 29 out of 149 false negatives (19.5%)
  - 80.5% screen capture rate
  - Comparable to national data (76.5%)

(Thangaratinam et al., 2012, Lancet)

CCHD Treatments

- Prostaglandin (PGE) & O2 supplementation
  - 72% given PGE; 54% given O_2 (158 cases)
  - Comparable to false negatives (29 cases)
- Failed screening even after treatments:
  - Newborns at altitude have more severe defects
  - Current screening algorithm is sufficiently applicable and effective at altitude

In Summary

- First comprehensive review of newborns already diagnosed CCHD
  - Novel approach
  - Delineate average and range of O_2 saturations
  - Evaluate performance of current screening algorithm performed at altitude

Implication & Future Direction

- 80.5% capture rate of current screening algorithm performed at altitude
  - Helpful in moving forward with CCHD screening efforts in CO
- Combine with data of true negatives to get a complete assessment of the performance of screening algorithm at altitude

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