Colorado Newborn Blood Spot Screening Program (CONBSP)

HCU Network America Round Table

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Name of presenter Gregory Bonn
Job title of presenter Newborn Screening Program Manager
Division of Disease Control and Public Health Response

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Colorado NBS

- The Colorado Newborn Bloodspot Screening Program (CONBSP) is part of the Colorado State Public Health Laboratory within the Division of Disease Control and Public Health Response (DCPHR) at CDPHE and tests for Colorado and Wyoming newborns for 39 life threatening genetic disorders.
- Annually, there are approximately 68,000 births in CO and WY.
- Colorado is a two screen state with samples submitted by 800 unique submitters. This translates into testing more than 130,000 samples using nine different methods for more than 700,000 individual tests annually.
- Annually, on average, testing identifies 700 affected newborns.there are 700 affected children identified due to this testing.
- Statute requires the CONBSP to operate six days a week, including holidays, in order to ensure timely reporting of abnormal results.The CONBSP is required by statute to operate 6 days a week including holidays in order to ensure timely reporting of abnormal results.
- 15 full-time staff members perform all testing, reporting, education, and stakeholder engagement.All testing, reporting, education, and stakeholder engagement is performed by 15 full-time staff members.

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Colorado Algorithm Fall 2021

- Two-screen state.
  - First Screen collected at 24-48 hours of life.
  - Second Screen collected at 8-14 days of life.
- NICU Algorithm
  - First Screen collected prior to 24 hours of life, all testing re-requested on second screen.
  - Hemoglobin AF sample results on first screen, all testing re-requested on second screen.
- Cutoff for Homocystinuria
  - Primary Analyte Methionine 100 umol\L
  - Secondary Ratio Methionine/Phenylalanine 1.0
  - First screen reported out as borderline with message send in a second screen.
  - Second screen reported to Children's Colorado Hospital Metabolic Specialists and PCP.

Colorado Algorithm: November 2021

- HCU webinar.
- Reviewed the historical cutoff and outcome data with MS Scientist.
  - PerkinElmer Cutoff Analyzer review.
  - Mayo CLIR review.
- Proposed new cutoff.
  - Met 48 umol/L Met/Phe 1.0
- Discussed update with Children's Colorado Hospital Specialists.
  - Concern about false positive calls.

Case Presentation

Summer 2022

6 yo M with developmental delay and bilateral pes planovalgus
presented to Children's Eye Center for visual impairment

Diagnosed with bilateral supero-temporal lens subluxation with high myopia
Referred to Cardiology with concern for Marfan syndrome
Found to have bilateral aortic root dilatation and mild pectus carinatum
Invitae Aortopathy Comprehensive Panel/Ehlers Danlos Syndrome
Panel: Heteroxygous for 2 pathogenic variants in CBS
c.1224-2A>C (splice acceptor)
c.1397C>A (p.Ser466*)
Homocysteine 320.5 (Ref: 6.6 – 14.8 mcmol/L)
Methionine 624 (Ref: 7-47 nmol/mL)

NBS results Met 53 Met/Phe 1.1

Follow-up

- August 2022 moved to Neobase 2 and QSight instruments.
- New Met 33 umol/L Met/Phe 1.0 Neobase 2 and QSight update.
- Regular review of cut offs.
  - All conditions.
- No increase in call outs.
- No other missed cases noted in Colorado based on Children's Colorado Hospital.

Thank you!

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