

2. 8 Biliary Atresia

Biliary atresia is a disorder that leads to the bile ducts closing off in the first few months after birth and (if untreated) eventual liver failure. It is the leading reason for liver transplantation in children. There is evidence that the prognosis improves if it is detected early.

Harpavat et al[1] reported preliminary results of screening for biliary atresia using direct bilirubin measurements in newborns. An excerpt from the abstract is pasted below:

"Of 124,385 newborns in the screening study, 49.2% were female, 87.6% were of term gestational age, 70.0% were white, and 48.1% were Hispanic. Screening identified the 7 known infants with biliary atresia with a sensitivity of 100% (95% CI, 56.1%-100.0%), a specificity of 99.9% (95% CI, 99.9%-99.9%), a positive predictive value of 5.9% (95% CI, 2.6%-12.2%), and a negative predictive value of 100.0% (95% CI, 100.0%-100.0%)."

- A. Assume that all of the numbers in the abstract are correct. Create a 2×2 table for the results above. (2 points)
- B. What was the prevalence of biliary atresia in this population? (2 points)
- C. The 100.0% negative predictive value looks really good! But the 95% CI (100.0%-100.0%) for NPV looks suspicious, since the lower 95% CI limit for the sensitivity is only 56%. If the false-negative rate could be as high as 44%, can we really be confident that the negative predictive value is very close (within rounding error of) 100%? Explain. (2 points: 1 pt for correct yes/no, 1 pt for explanation)

1. Harpavat S, Garcia-Prats JA, Anaya C, Brandt ML, Lupo PJ, Finegold MJ, et al. Diagnostic Yield of Newborn Screening for Biliary Atresia Using Direct or Conjugated Bilirubin Measurements. JAMA. 2020;323(12):1141-50.