EXOME SEQUENCING FOR NEWBORN SCREENING

In a recent paper in the AJHG, Roman and her coworkers blindly sequenced the exome 106 children: 17 with inborn errors of metabolism, 28 with hearing loss, and 61 healthy newborns, to simulate a newborn screening. The study detected a medically actionable variant in 4 of 106 newborns that would have been missed by the standard newborn screening procedure. They also found that there were on average 1.8 reportable carrier variants per child. These findings per se are not novel, but the comparison to the standard newborn screening is new. The paper provides us with a glimpse of what we can expect if exome sequencing were to be used for newborn screening programs as a diagnostic tool. The authors stress the importance of finding a balance between increasing sensitivity, to maximize case finding, versus establishing stringent thresholds to reduce false positives.