Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy (SMA) is the most common lethal genetic disease in children.

The incidence of SMA is **one in 6,000 to one in 10,000 live births.**

Carrier frequencies in most populations are estimated to be one in 40 to one in 60 for this autosomal recessive disorder.

With just one assay, ARUP identifies patients affected with SMA, carriers of SMA, and those at increased risk to be "silent carriers."

2013436 Spinal Muscular Atrophy (SMA) Copy Number Analysis
This test is a first-tier diagnostic test for individuals suspected of having SMA and includes copy number for *SMN1* and *SMN2*. It may also be used for carrier screening in families with *SMN1* deletions. In addition, carrier screening for SMA should be offered to all individuals who are pregnant or considering pregnancy, as recommended by the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG).