

ARIZONA NEWBORN SCREENING PROGRAM

Arizona Department of Health Services — State Public Health Laboratory
250 North 17th Avenue | Phoenix, AZ 85007

NEWBORN SCREENING REPORT

Newborn:	Sofia Reyes
DOB:	2026-01-15
Sex:	Female
MRN:	SOFIA_R
Collection Date:	2026-01-20
Screening Program:	Arizona Newborn Screening Program

SCREENING RESULTS

Condition	Result	Action Required
Spinal Muscular Atrophy (SMA)	POSITIVE — SMN1 deletion detected	URGENT: Confirmatory genetic testing and pediatric neurology referral required
Cystic Fibrosis	Normal	None
Sickle Cell Disease	Normal	None
PKU	Normal	None
Congenital Hypothyroidism	Normal	None

CRITICAL RESULT — IMMEDIATE FOLLOW-UP REQUIRED

SMA screening POSITIVE: SMN1 gene deletion detected. This result requires immediate confirmatory genetic testing (SMN1/SMN2 copy number analysis) and urgent referral to pediatric neurology. Early treatment in presymptomatic SMA is associated with significantly better motor outcomes.