

PHOENIX CHILDREN'S HOSPITAL

Department of Pediatric Neurology
1919 East Thomas Road | Phoenix, AZ 85016

NEUROLOGY ASSESSMENT

Patient:	Sofia Reyes
DOB:	2026-01-15
Age:	2 weeks (17 days)
MRN:	SOFIA_R
Assessment Date:	2026-02-01
Evaluating Physician:	Dr. Michael Torres, MD

CHIEF COMPLAINT

Presymptomatic SMA detected via newborn screening — urgent request for early treatment initiation

HISTORY OF PRESENT ILLNESS

3-week-old female born full-term (39 weeks, birth weight 3.2 kg, Apgar 8/9) via uncomplicated vaginal delivery. Arizona newborn screening returned positive for SMA (SMN1 deletion) on day 5 of life. Confirmatory genetic testing confirmed homozygous SMN1 deletion with 4 SMN2 copies. Currently completely asymptomatic with age-appropriate motor milestones. Neurological examination at 3 weeks shows normal tone, reflexes, and spontaneous movements. Pediatric neurologist recommends immediate treatment initiation to preserve motor neurons before symptom onset. Literature strongly supports that presymptomatic treatment leads to significantly better outcomes than waiting for symptom onset.

MOTOR FUNCTION ASSESSMENT

SMA Type:	Presymptomatic — type not yet determined
Motor Milestones:	Age-appropriate — normal muscle tone and reflexes on neurological exam at 3 weeks
Current Motor Function:	Normal for age. Active spontaneous movements, appropriate tone, symmetric reflexes. No fasciculations. Normal suck and swallow.
Feeding Status:	Normal breastfeeding
Respiratory Status:	Normal. No respiratory compromise.
Ventilator Dependent:	No

ASSESSMENT AND PLAN

Recommend initiation of Spinraza (Nusinersen) therapy based on clinical assessment.

Dr. Michael Torres, MD

Pediatric Neurology

NPI: 1432567891

Date: 02/01/2026