ANN & ROBERT H. LURIE CHILDREN'S HOSPITAL OF CHICAGO

Department of Pediatric Neurology
225 East Chicago Avenue | Chicago, IL 60611

NEUROLOGY ASSESSMENT

Patient:	Ethan Williams
DOB:	2025-06-10
Age:	7 months
MRN:	ETHAN_W
Assessment Date:	2026-01-25
Evaluating Physician:	Dr. Anna Kowalski, MD, PhD

CHIEF COMPLAINT

Progressive generalized hypotonia and motor regression in 7-month-old male with genetically confirmed SMA Type 1

HISTORY OF PRESENT ILLNESS

7-month-old male born full-term with normal birth history. Parents noted poor head control and generalized hypotonia at 3 months of age. Referred to pediatric neurology at 4 months. EMG showed diffuse denervation pattern consistent with anterior horn cell disease. Genetic testing at 5 months confirmed SMA Type 1 (homozygous SMN1 deletion, 3 SMN2 copies). Progressive motor decline with loss of previously acquired limb movements. Currently unable to sit independently, poor head control, weak cry, feeding difficulties requiring intermittent NG supplementation. No prior gene therapy (Zolgensma) or antisense oligonucleotide therapy.

MOTOR FUNCTION ASSESSMENT

SMA Type:	Type 1
Motor Milestones:	Never achieved independent sitting
Current Motor Function:	Severe generalized hypotonia, poor head control, weak cry, tongue fasciculations, paradoxical breathing pattern
CHOP INTEND Score:	18 (Severely reduced motor function (max score 64))
Feeding Status:	Oral feeding with modified consistency; intermittent NG supplementation
Respiratory Status:	No ventilatory support required. SpO2 93-96% on room air. BiPAP during sleep.
Ventilator Dependent:	No

ASSESSMENT AND PLAN

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

Dr. Anna Kowalski, MD, PhD

Pediatric Neurology

NPI: 1654789321

Date: 01/25/2026