

Risdiplam Improved Motor Function in Late Treatment of Spinal Muscular Atrophy Type 1

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INTRODUCTION

Treatment for SMA is unavailable for patients with advanced disease. In clinical trials, patients with invasive ventilation or tracheostomy and those with history of hospitalization for a pulmonary event within the last 2 months at the time of screening were excluded from use of Risdiplam. There is limited documentation on outcomes for initiation of therapy in these patients.

OBJECTIVES

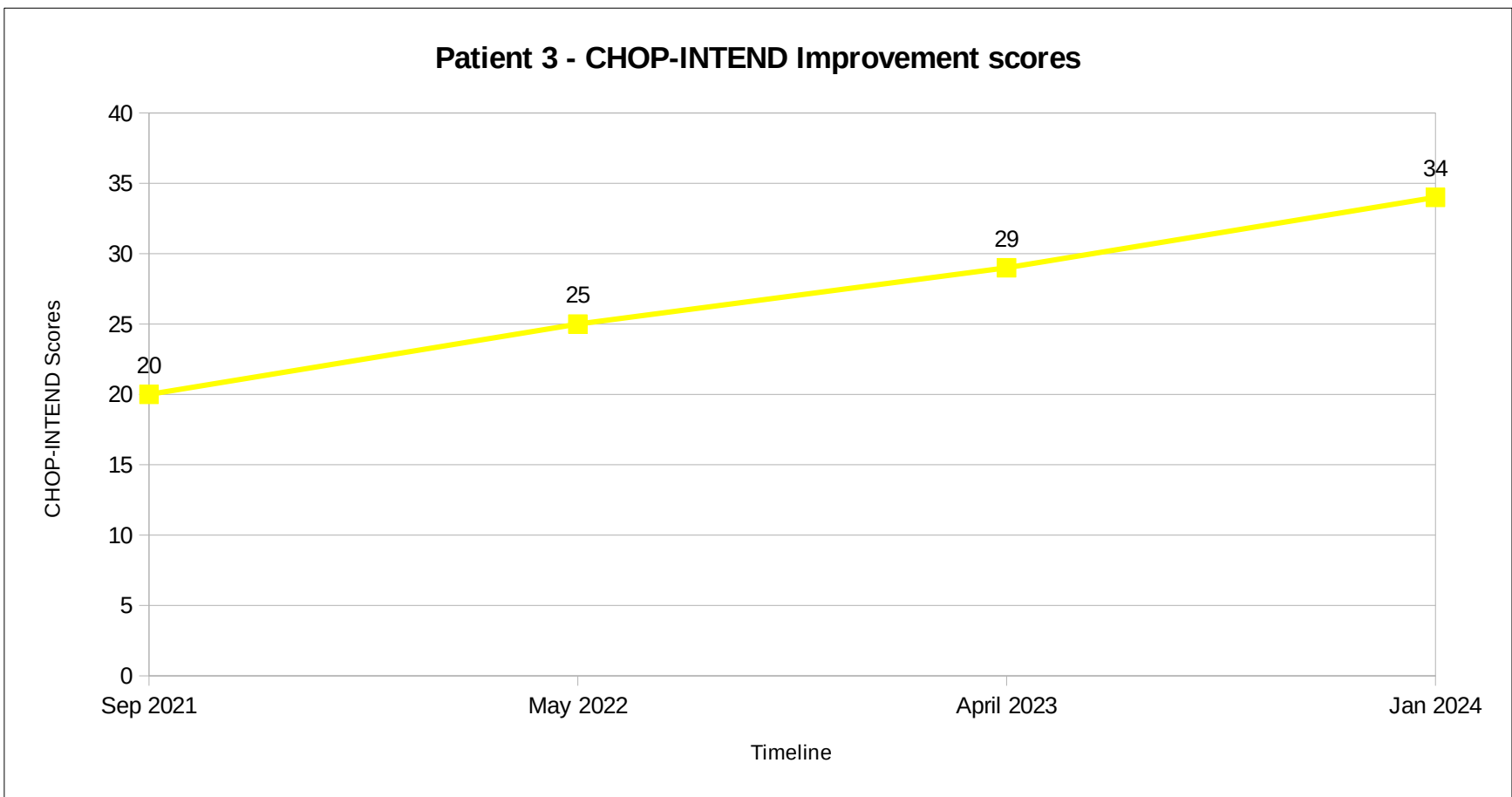
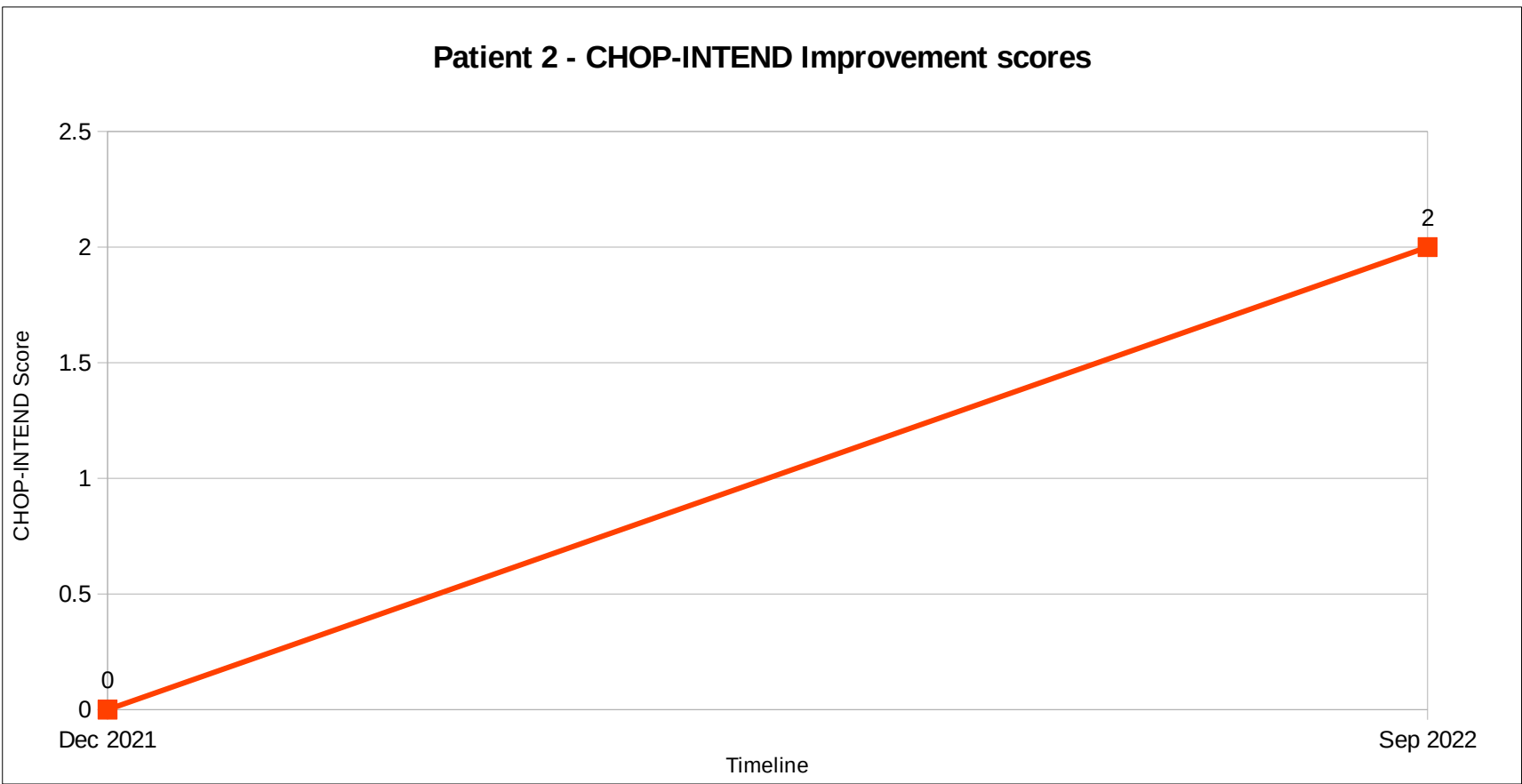
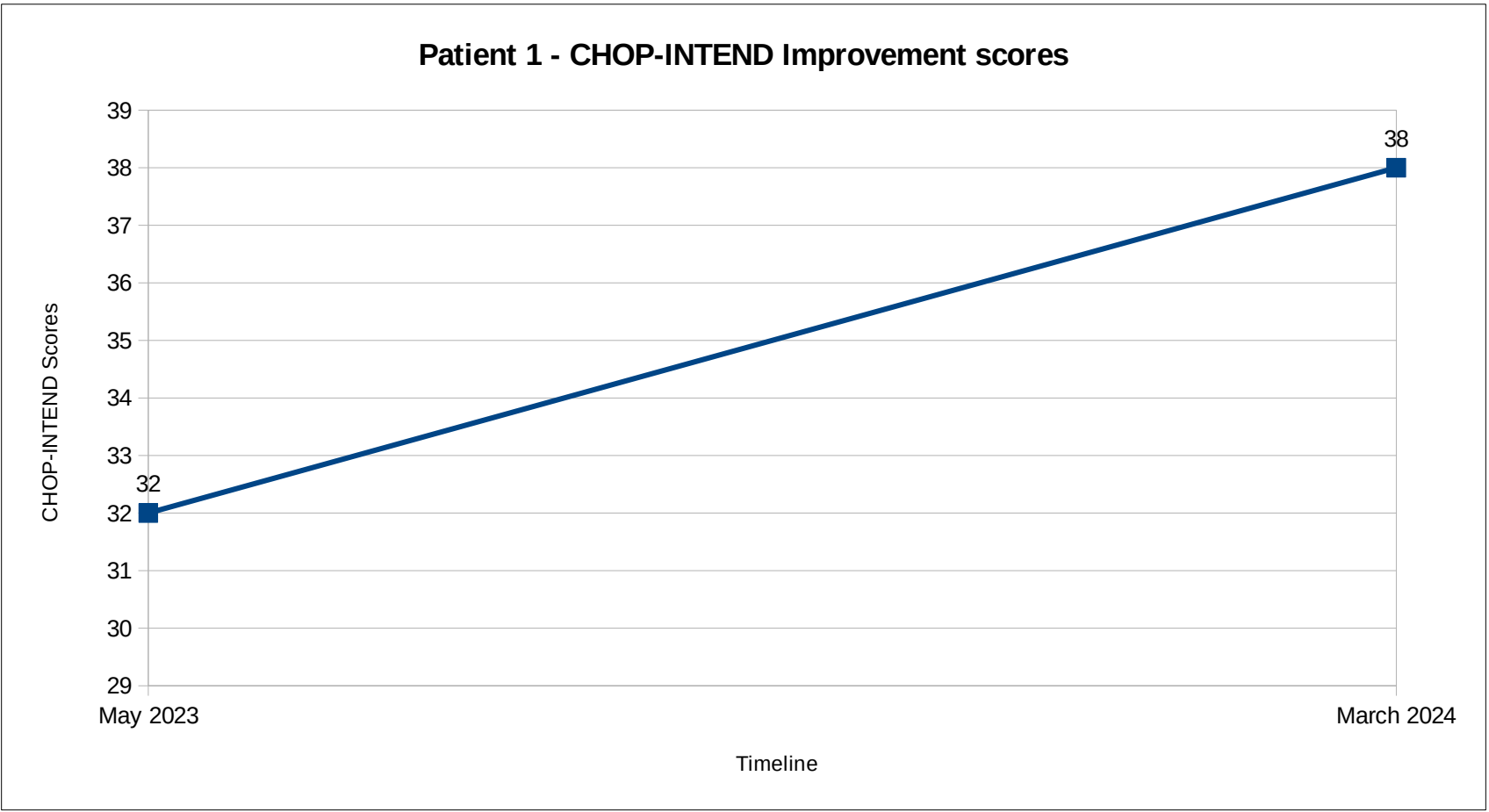
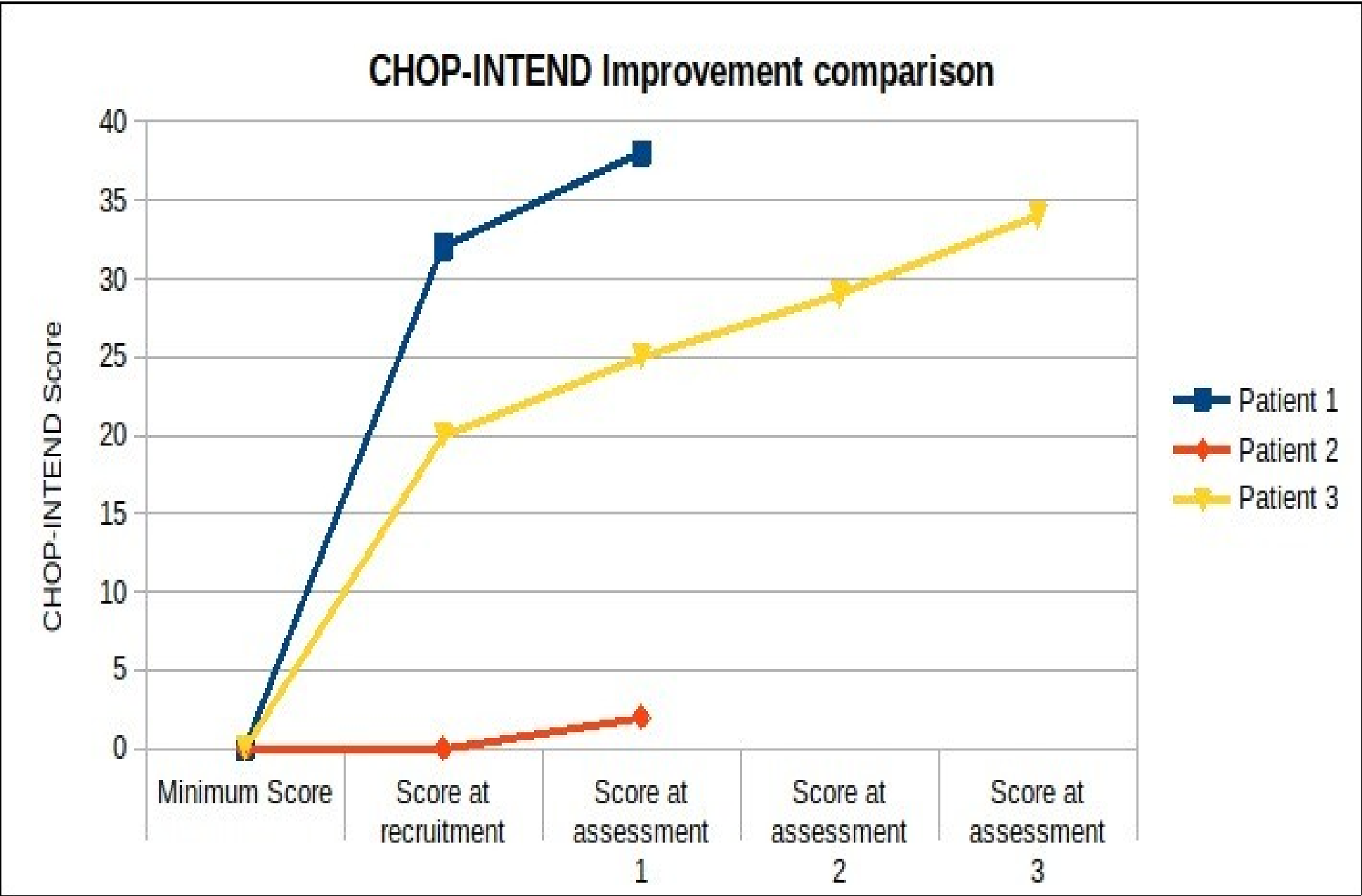
To document motor and respiratory outcomes for patients with SMA type 1 with late treatment starting points.

METHODS

We conducted a case series of five patients on follow up from September 2021 for SMA type 1 with severely limited motor skills and on treatment with Risdiplam. We assessed the disease course using the Children’s Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND).

RESULTS

We followed up five patients, the patients’ ages ranged from 3 months to 5 years at the time of recruitment into the compassionate Risdiplam program. Two patients were already on respiratory support and had gastrostomy tubes by the time Risdiplam was being initiated. Two patients died prior to initiation of Risdiplam due to aspiration pneumonia less than two months after diagnosis. Patient 3 improved from using respiratory support for 24 hours to 18 hours a day, complimented by occupational and physical therapy.



CONCLUSION

Risdiplam was associated with improved motor and respiratory outcomes in late treatment SMA type 1 patients.

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