

# Transitions in Care in SMA

Case contributor and commentary:  
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## Patient #2: Fragmented adult care 21-year-old female

### Case Timeline



### Early life

The patient's parents recalled preclinical symptoms during her early life.

- » Symptoms began at 3 years of age
- » Slow runner
- » Difficulty getting up from the floor

### Age

13 -21

### Diagnosis

- » Diagnosed at 13 years of age
- » Diagnosis of 5q-SMA was genetically confirmed (SMN1 copy number 0; SMN2 copy number 3)
- » She noted a slow decline in her motor function over time
  - » Could not participate in the gym class in high school
  - » Needed to use the rails to go up the steps
  - » Could not put a suitcase in the overhead cabinet

### Adult SMA

Adult SMA is a practical term that describes individuals with genetically confirmed 5q-SMA, age 18 and older. They are best described based on disease severity using current functional status as severe non-ambulatory (unable to sit), less severe non-ambulatory (able to sit independently), and ambulatory (able to walk independently) groups.<sup>1,2</sup>

#### General health status

- » Fatigued
- » Migraine without aura

#### Management

- » Prescribed Vitamin D supplement
- » Followed with PCP for migraine
- » Migraine well controlled on Topiramate and PRN Excedrin

#### Mental health status

- » Followed with PCP for anxiety/ depression
- » Stable on current treatment
- » Met with a counselor a few times, but not regularly

#### Management

- » Prescribed selective serotonin reuptake inhibitor (SSRI)

#### Mobility status

- » Retained the ability to walk independently

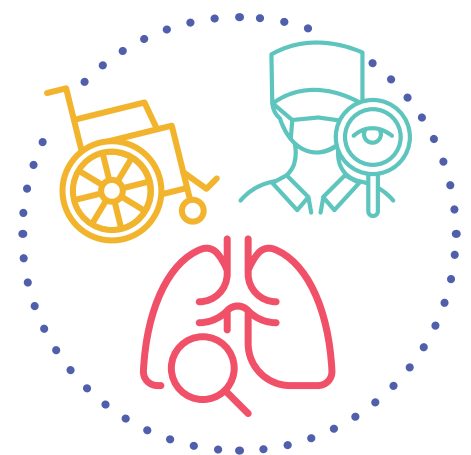
#### Management

- » Preferred to use wheelchair outside the house

#### Additional status updates

- » No pain
- » No sleep issues
- » No breathing difficulties

- » No GI-related symptoms
- » No recent infections or hospitalization
- » No tobacco, alcohol or illicit drug use



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### Disease-modifying treatment

Following FDA approval of nusinersen, the patient started treatment at a local clinic for 10 months.

- » Treatment and lumbar puncture were well tolerated, except for occasional transient headaches and low back pain
- » Patient reported subjective improvement of stamina and strength
- » Data on baseline and post nusinersen motor function assessments was not available

### Age

## 21 The patient relocated, which led to disruptions in the continuity of care.

#### Life change

- » Moved to a new state and rented an apartment with a friend
- » Established care at MDA Care Center
- » Requested continuation of nusinersen treatment (Next injection was due in 2 weeks)



#### Challenges to maintaining care

- » Shared records did not have documentation of the genetic testing or the motor function scales assessments
- » Patient was concerned about treatment delay. She experienced wearing off the treatment effect in the last month before the following dose
- » Nusinersen was approved by new insurance after obtaining genetic testing, functional assessments, and going through an appeal process. This delayed injection by about 4 weeks
- » Blood work up, including coagulation profile and complete blood count, and urine testing for protein were obtained prior to the injection

### Commentary

- » The high price of the medications resulted in a complex and lengthy insurance approval process.
- » Baseline and post-treatment motor function assessments are important to track treatment response and help with insurance approval (Table 1).
- » Communication is key for optimal care. Lack of a transition plan could result in fragmented, suboptimal and delayed care.

**Table 1: Commonly used assessments<sup>3,4</sup>**

Walkers <sup>5</sup> :	Sitters:	Non-sitters:
<ul style="list-style-type: none"> <li>» 6 minute walk test (6MWT)<sup>6</sup></li> <li>» Hammersmith functional motor scale expanded (HFMSE)</li> <li>» Strength measurement</li> <li>» Handheld dynamometry (HHD)</li> <li>» Maximum voluntary isometric contraction testing (MVICT)</li> <li>» Modified SMA Functional Rating Scale (SMAFRS)</li> <li>» Pulmonary function tests (PFT)<sup>7</sup></li> </ul>	<ul style="list-style-type: none"> <li>» Revised upper limb module (RULM)<sup>8</sup></li> <li>» HFMSE</li> <li>» SMAFRS</li> <li>» PFT</li> </ul>	<ul style="list-style-type: none"> <li>» Children's Hospital of Philadelphia's Adult Test of Neuromuscular Disorders (CHOP-ATEND)</li> <li>» SMAFRS</li> <li>» PFT</li> </ul>

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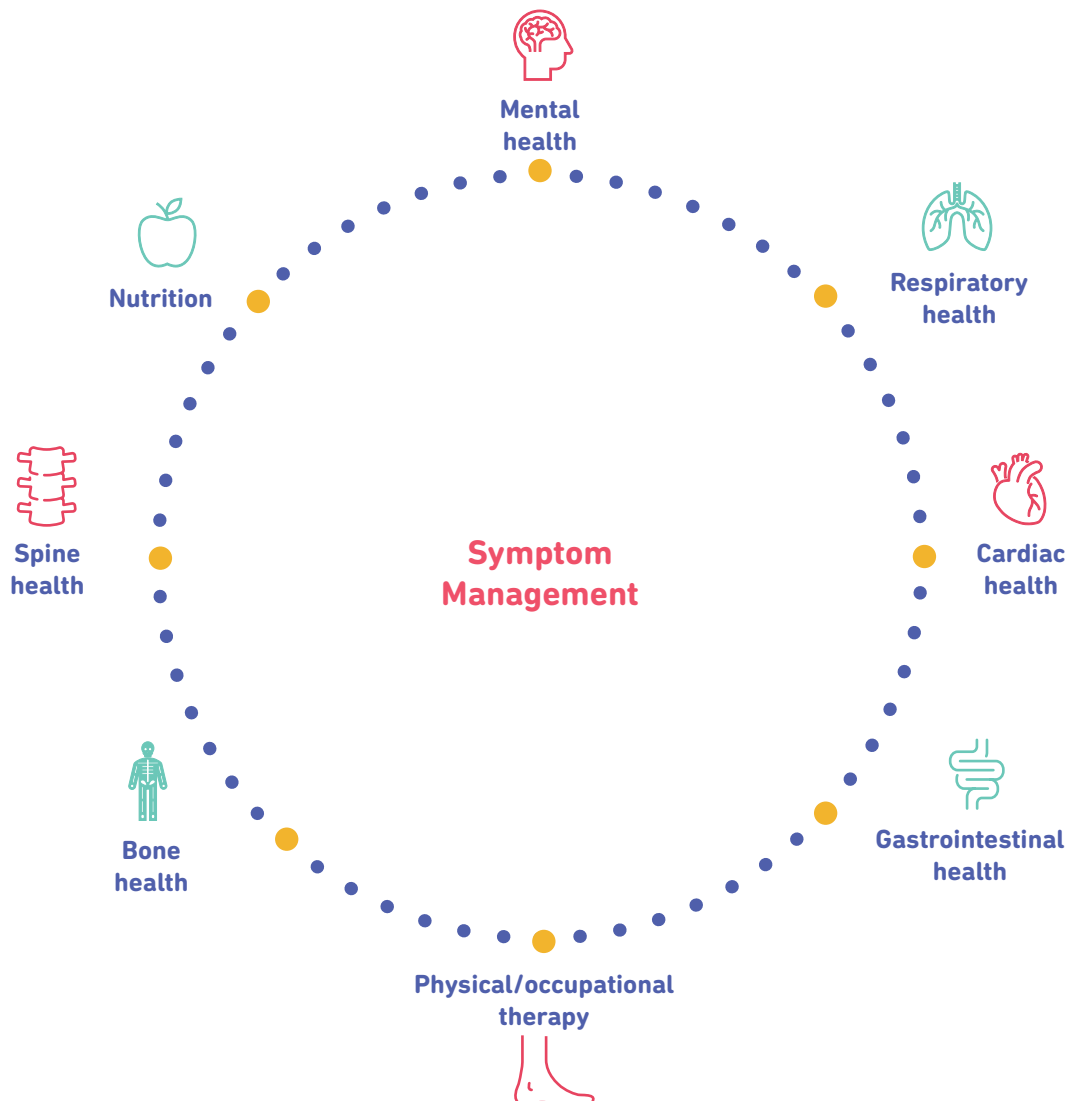
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### Adult clinics vs. pediatric clinics

Adult clinics differ from pediatric clinics in the following:

- » Patients are seen as self-reliant. The focus of medical team interactions shifts from the family and parents to the patient, who is viewed as a more autonomous decision-maker.
- » Shorter visit duration
- » Higher expectations from patient to adhere to care
- » Additional focus on medical issues related to adulthood

### Multidisciplinary Care



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The patient was referred for the establishment of care. Based on diagnosis of 5q-SMA adult SMA, the following multidisciplinary evaluation and management plan was implemented.

### Physical and occupational therapy (PT/OT)

- » Met with PT for assessments (6MWT, HFMSE, and HHD) to track response to treatment — Was instructed on stretching, range of motion, muscle strengthening, and aerobic exercise
- » Referral was placed for wheelchair repairs

### Respiratory care

- » Met with the respiratory therapist — Had an office PFT that was normal
- » Influenza and pneumococcal vaccinations were up to date

### Neuromuscular and mental health

- » Migraine, anxiety, and depression were discussed
- » Patient was provided with contact information of a new primary care physician and behavioral health clinic and advised to schedule appointments

### Bone health

- » Vitamin D level was normal — Was advised to continue the supplements

### Nutrition

- » Patient was connected with registered dietitian services in the clinic
- » BMI was normal
- » No nutritional needs were reported at this stage

### Genetic testing

- » Patient was made aware of the availability of the genetic counselor

## Future treatment goals and expectations were discussed with the patient.

- » Patient was assessed for side effects related to the medication and lumbar puncture
- » Patient was updated on research and the drug- pipeline (discussion occurred prior to approval of risdiplam for SMA in adults and infants/children)

### Recommendations for optimizing care in adults with SMA

Adults with SMA are encouraged to play an active role in their care. In addition to the use of disease-modifying medications, a comprehensive care plan delivered by a multidisciplinary team is crucial for better outcomes. The international standard of care documents<sup>9,10</sup> are valuable; however, there are gaps pertinent to the adult SMA population. For instance, data on pain, women's health, and mood disorders are limited. Individuals with SMA are encouraged to participate in research. Information about research studies can be found by asking their providers, checking [clinicaltrials.gov](https://clinicaltrials.gov), or visiting [mda.org](https://mda.org) and using the "Clinical Trials Finder Tool."

### Key learning points

- » Coordinated transition of care for adults with SMA is crucial for the continuity of care
- » A multidisciplinary team approach is important for optimal care
- » It is essential, despite the limited data in the adult SMA population, to screen and treat issues related to mental health

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### References

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