



Natural history studies are an important tool for understanding the etiology, range of manifestations and progression of rare diseases like Smith-Magenis syndrome (SMS). This workshop examines the guidelines for medical management and treatment for individuals with SMS developed by PRISMS Professional Advisory Board that were updated in 2018. Management involves evaluation for manifestations of SMS and treatment to mitigate associated symptoms and conditions. Ongoing surveillance recommendations as well as health promotion strategies will also be discussed.

I. Medical management recommendations for SMS

- A. At Diagnosis
- B. Across the lifespan – surveillance and transition to adulthood: caveats from SMS Natural History study that warrant attention and management intervention

II. Problem Identification in SMS - Personalized problem management

- A. Health promotions strategies & transition to adulthood
- B. Hands-on approach/practical tips that work for the child with SMS and entire family

III. Added Resources/Links:

SMS Medical Management Guidelines and Checklist (v1.2018), available at

<https://www.prisms.org/about-sms/living-with-sms/medical-management-guidelines/>

Bright Futures/AAP Recommendations for Preventive Pediatric care (2022):

<https://www.aap.org/en/practice-management/bright-futures/bright-futures-materials-and-tools/bright-futures-guidelines-and-pocket-guide/>

Apps for exercise and nutrition with general options for individuals with developmental disabilities that can be customized to the needs of the patient: <https://www.hwms.com.au/news/best-apps-for-people-with-disabilities>

Helpful overview for fitness promotion for children/youth with special health care needs:

<https://ucedd.georgetown.edu/documents/PhysicalActivityFactSheetFinal.pdf>

2022 Adult Preventive Health Guidelines: Ages 19 through 64 Years

<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation-topics>

2022 PCRC Treatment Recommendation: Constipation in Individuals with Smith-Magenis Syndrome:

Recognition and Recommendations for Treatment (AU: Gail Reiner, DNP, FNP-C, Myra Woolery, PhD, RN, APRN-CNS, CPON, and Ann C.M. Smith, MA, DSc (Hon.), CGC)

<https://www.prisms.org/constipation-treatment-recommendations-for-sms/>

MEDICAL MANAGEMENT OF SMS ACROSS THE LIFESPAN (SMITH, REINER)

Friday, August 5, 2022 9:45am-11:00am

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