



February 2022

Smith-Kingsmore Syndrome Foundation

Rare Disease Day is Monday February 28th!

You can get involved in multiple ways to support Rare Disease Day:

1. Join the [SKS Rare WOD](#). See the work-out below and visit this link [here](#) to learn more about how you can work-out to support SKS research
2. Go to [Twibbonize](#) to upload your Social media profile pic with SKS Rare Disease Day frame
3. Share your SKS Story with the SKS Foundation for our family story page. See the [story page](#) for examples and email us [here](#)
4. Share your story with Nord. See this link [here](#)
5. Share your SKS Story on social media and use hashtags #RareDiseaseDay & #smithkingsmoresyndrome

All of these speak to our community mission of raising awareness of this ultra-rare disease!

*click hyperlinks for more details

SKS Research Update



Current Research:

- Dr. Liu, Dr. Prada & the Cincinnati Children's SKS Team have submitted their first publication supported by SKSF funded research. This will be a comprehensive paper on SKS & will open up new avenues for scientific inquiry
- Dr. Yang, the SKS post-doc at UF, research continues to evaluate the differences between 20 + different MTOR variants

Future Research:

- Dr. Prada & Dr. Krueger, at Cincinnati Children's Hospital, are evaluating surveys collected and looking at brain function and behaviors. Look for the email from Lindsey E. Aschbacher-Smith requesting MRI and/or EEG data.

Want to Enroll in SKS Studies?

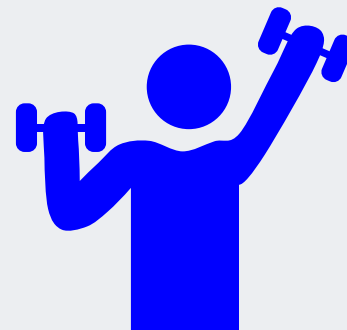
- Email Lindsey at Lindsey.Aschbacher-Smith@cchmc.org for more details.

Calling all German, Belgium & Netherland Families:

- Email Dr. Reijnders at margot.reijnders@mumc.nl for more information on the mTORopathy Expertise Center

February 26th

Workout for Rare Disease Day



The Smith-Kingsmore Syndrome
RARE Workout Of the Day
Saturday February 26th, 2022
Virtual Event



Powered By
Smith-Kingsmore Syndrome Foundation

#rarewod2022
#foundation #giveback
#smithkingsmoresyndrome
www.smithkingsmore.org

Mission: SKS Families, Friends, and Communities are virtually coming together for the **SKS RARE WOD** on February 26th. During a time of social distancing and quarantine this is your chance to celebrate by joining a cause worth working out for.

Execution: SKS Athletes compete at various levels to complete the **SKS RARE WOD**. SKS Athletes come together (in spirit & virtually) in order to raise awareness for **Rare Diseases Month** and celebrate the first ever **SKS RARE WOD**. Why such a hard workout? To "Be abnormal, among the normal." Just like these rare and extraordinary children.

SKS RARE WOD Athletic Levels:

Varsity: SKS Athletes compete in all four prescribed workouts. There are no substitution lifts/movements at the varsity level. Do the workout as prescribed.

Junior Varsity: SKS Athletes compete in two of the four **SKS RARE WOD** events with athlete choice of rest time in-between.

Relay: SKS Athletes compete as a team to complete all events of the **SKS RARE WOD**.

Athlete Choice: SKS Athletes pick any event to complete to the best of their abilities.

The Smith-Kingsmore Syndrome
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SKS RARE WOD GEAR

RFT: 5 Rounds

- 21x Double-Unders
- 15x Deadlift(Bodyweight)
- 9x Push-Press(1/2 Bodyweight)

AMRAP: 15 min

- 21x Pull-Ups
- 15x Thrusters (1/2 Bodyweight)
- 9x Toes to Bar

*Each round completed = 1min off overall time.

RUN: 5k

EMOM (Ascending 1,2,3...)

- Push-ups
- Air-Squats
- Burpees

*Each minute completed = 1min off overall time.



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RARE WALK

Dedicate a walk at any distance and pace to a child with a rare disease.

www.smithkingsmore.org

SKS RARE WOD NO GEAR

RFT: 5 Rounds

- 21x Jumping Jacks
- 15x (each leg) Lunges
- 9x Push Ups

AMRAP: 15min

- 21x Back Extensions
- 15x Burpees
- 9x V Ups

*Each round completed = 1min off overall time.

RUN: 5k

EMOM (Ascending 1,2,3...)

- Push-Up
- Air Squat
- Burpee

*Each minute completed = 1min off overall time.

RARE WOD 2022

It's time again for all the SKS athletes to sweat for SKS. Join us on for the world famous Rare WOD (workout of the day), powered by SKSF, on Saturday, February 26!

Click [here](#) to learn more about this event!

[Donate](#) today & support our SKS athletes!

2022 Foundation Initiatives

We continue to work with our dedicated SKS experts to raise awareness and increase research potential. Be on the lookout for these incredible initiatives:

- SKS Newly Diagnosed Informational Packet
- Up-To-Date SKS Report
- GeneReview SKS Report
- SKS NORD Report
- SKS Centers of Excellence - where medical professionals are up to date on providing evidence based care to people living with SKS



Smith-Kingsmore Syndrome Global Patient Registry Update February 2022

200+

Diagnosed
with SKS
Worldwide

69

Participants from
Australia, Belgium,
Brazil, Canada, France,
Italy, Japan, Kuwait,
Mexico, Netherlands,
New Zealand, Spain,
UK, USA

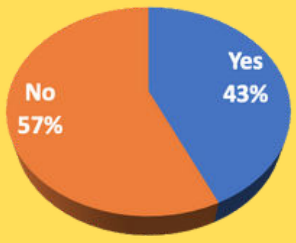


64% Male
36% Female

* 44 participants have completed SKS specific questionnaire

Seizures

43% have had at least one seizure



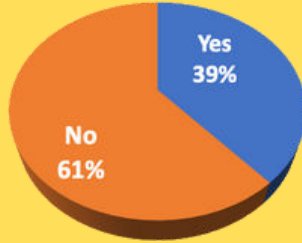
Interception & Body Temp Dysregulation

50% Have difficulty feeling full/hungry,
hot/cold, and/or overheats easily



Hyperphagia

39%



Neurodevelopmental & Behavior

45% Autism

79% Sleep Disturbances

45% Non-Verbal

23% Anxiety

All non-verbal participants have
intentional vocal sounds

86% Sensory Dysregulation,
Stimming and/or Self-
harming behaviour

96% Intellectual
Impairment and/or
Global Delay



To Start Your
SKS Registry Today