Healthcare Professional Letter for Smith-Lemli-Opitz Syndrome (SLOS)

Patient Name:_____

Date of Birth:_____

Dear ER/Consulting Physician,

_____has a rare metabolic disorder called Smith-Lemli-Opitz Syndrome (SLOS). SLOS is a genetic disorder resulting in patients producing insufficient endogenous cholesterol necessary for growth and development. Some children with SLOS have minor manifestations of the condition; others may have many of the defects listed below, in addition to other manifestations not listed.

Manifestations of SLOS:

- General: prematurity, failure to thrive (postnatal), developmental delay
- Neurobehavioral/Psychiatric: variable degrees of intellectual disability, self-aggression and self-injurious behaviors, hyperactivity, attention-deficit hyperactivity disorder (ADHD), autism spectrum disorders, sleep disturbances
- Craniofacial: microcephaly, malformations (including agenesis of the corpus callosum), cerebellar hypoplasia, bitemporal narrowing
- Face: ptosis, epicanthal folds, short and upturned nose, narrow hard palate, cleft palate, cataracts, low-set ears, micrognathia, facial anomalies
- Extremities: polydactyly or syndactyly, proximally placed thumbs, abnormal palmar creases
- Gastrointestinal: feeding difficulties (poor feeding/suckling), gastroesophageal reflux, constipation, pyloric stenosis, Hirschsprung disease, elevated 7-dehydrocholesterol (7-DHC) labs
- Heart: congenital defects and cardiac malformations

Because of the possibility of internal malformations, patients with SLOS should be evaluated carefully, especially for heart and kidney defects.

Treatment:

Treatment for SLOS is directed at symptom management. Patients may be undergoing treatment with any of the following:

- Cholesterol supplementation
- Cholic acid treatment
- Other management solutions for specific symptom management (eg, antioxidant supplementation)

In addition to your standard work-up, depending on presenting symptoms, please consider the above typical signs and symptoms of SLOS when considering a differential diagnosis or initial laboratory work-up.

If_____presents with an illness or if you have any questions regarding this diagnosis, please call the primary care provider, at_____or Dr._____, at_____.

Sincerely,

Name:_____

Phone:_____

For more information on SLOS, please visit:

Smith-Lemli-Opitz Foundation
smithlemliopitz.org

National Organization for Rare Disorders (NORD)
rarediseases.org

National Institutes of Health (NIH) Genetic and Rare Diseases Information Center (GARD)
https://rarediseases.info.nih.gov

Human Phenotype Ontology (HPO)
hpo.jax.org

Note: the organizations listed above are not affiliated with Mirum Pharmaceuticals, Inc.