• On December 23, 2016, the U.S. Food and Drug Administration (FDA) approved the use of nusinersen (brand name: Spinraza), a new drug treatment for spinal muscular atrophy (SMA) in children and adults.

• Boston Children’s Hospital’s Spinal Muscular Atrophy Program has been involved in clinical trials of nusinersen since 2011, under the leadership of neurologist Basil Darras, MD. Our multi-specialty SMA team helped to develop standard ways to measure patients’ SMA symptoms and the changes in these symptoms over time. These results are being used to measure the effectiveness of nusinersen and other treatments.

• We’re here to help answer any questions you may have about this new treatment for SMA. Below are answers to questions we’ve received so far. For more information, please contact our Spinal Muscular Atrophy Program at 617-355-8235, Monday through Friday, 8:30 to 5:00 p.m. U.S. Eastern time.

Can my child use nusinersen?

• To be treated with nusinersen, your child must have the SMN1 mutation that causes this disease.

• The FDA approved the use of this drug for all types of SMA with an SMN1 mutation, from infant-onset SMA Type 1 to adult-onset SMA Type 4.

• Talk with your child’s doctor to see if nusinersen is right for your child.

How does nusinersen work?

• SMA is caused by a change in the survival motor neuron gene 1, also known as SMN1. All people with SMA have a copy of the survival motor neuron gene 2, called SMN2. But an important protein is missing in the SMN2 gene.

• Nusinersen uses synthetic (man-made) genetic material called antisense to replace the missing protein in SMN2. This helps people to make enough SMN protein needed for healthy motor neurons.

Will my child benefit?

About the ENDEAR trial

• The greatest effects of nusinersen have been seen in infants with SMA Type 1, who participated in the ENDEAR trial. These children were enrolled before 7 months of age and now range in age from 1.5 years to 3 years old. In the trial, study investigators saw infants reaching developmental milestones like head control, rolling over, and sitting and standing. A small number of children were able to walk with support.

• The ENDEAR trial was blinded, which means participants did not know if they received nusinersen or a placebo (a pill or liquid that doesn’t contain any medicine). We’re now learning who received the active drug and who received the placebo.

• Historically, 90 percent of children with SMA Type 1 have passed away before their second birthday unless given aggressive respiratory support.

About the CHERISH trial

• The CHERISH trial studied the effects of nusinersen in children ages 2 to 12 with SMA Type 2. The changes were positive.

• Children with severely limited mobility, chronic respiratory failure, swallowing problems and severe joint contractures or severe scoliosis were not included in the CHERISH trial. Therefore, the effects of nusinersen in children with more advanced SMA Type 2 are uncertain and will likely vary from child to child.

Nusinersen for treatment of SMA Type 3 and 4

• Nusinersen has been studied in SMA Type 3, but not in a controlled way. Open-label studies show some signs of benefit — for example, some patients were able to walk longer on a 6 minute walk test.
• We don’t yet know whether nusinersen will help people with adult-onset SMA (Type 4).

Overall, the results from clinical trials show that the earlier treatment starts and the longer a child is treated, the better the outcome will be. Nusinersen does not work in everyone. Also, we still don’t know how long the benefits will last, since we have only followed children for, at most, several years. The risk and benefits of this drug will be reviewed with the patient, family and medical team on a regular basis.

How is nusinersen given?
• Nusinersen is injected directly into the spinal canal (intrathecally). This requires a lumbar puncture or spinal tap.
• The care team individualizes the injection process based on each child’s health and needs.
  • It’s important to be still during the lumbar puncture. Some children may need anesthesia to sleep through the procedure.
  • Children with spinal fusions may need an interventional radiology procedure to safely give the drug.
• We start treatment by giving 4 higher doses of nusinersen. After that, we give a lower dose of this drug once every 4 months.
• We believe this treatment will most likely need to be given for life.

How do I know if nusinersen is covered by my health insurance?
• It’s not yet clear how much of the cost health insurance providers will cover for nusinersen.

• We encourage you to talk with your health insurance provider, as coverage varies from plan to plan.
• We are working with insurance companies and the drug’s manufacturer, Biogen, to help make the treatment as accessible as possible to families.
• Biogen offers a family support group called SMA360° that can help you to look into insurance benefits.

My child is in the open-label trial. Can he or she still receive nusinersen?
• Children already participating in the open-label trial (known as CS11) can continue receiving the drug at no cost until the trial is complete.
• However, the trial led by Biogen is not accepting new patients.

What’s happened with Biogen’s expanded access program (EAP)?
• Now that the drug has been approved by the FDA, this trial is no longer recruiting (taking) participants. However, children already enrolled may be able to continue receiving the drug at no cost through this program until families get insurance authorization for the commercial drug.

Is there a shortage of the drug?
• No, Biogen has said that it has enough supply. There may be a time delay of several weeks before the drug is available for our hospital to give.

What if I have other questions?
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