Moyamoya syndrome is a disease that affects certain arteries in the brain. In this handout, you will learn about moyamoya syndrome, including its symptoms and causes. You will also learn how doctors test for and diagnose moyamoya syndrome.

WHAT IS MOYAMOYA SYNDROME?
Moyamoya syndrome is a disorder that causes certain arteries in the brain to constrict (tighten or shrink) and thicken. The arteries affected are called internal carotid arteries. They help carry blood to the brain. The constricted blood vessels can put a person at a higher risk for a stroke (when blood supply to the brain is interrupted by a blockage or blood flow issue).

Moyamoya syndrome is 3 times more common in people with Down syndrome than people without Down syndrome. Doctors do not know why people with Down syndrome have a higher risk of developing moyamoya syndrome.

WHAT ARE THE SIGNS AND SYMPTOMS OF MOYAMOYA SYNDROME?
People with moyamoya syndrome may not have any symptoms for many years. Over time, the thickening of the internal carotid arteries can cause different neurological (brain and nerve) related issues. The most common sign of moyamoya syndrome is a stroke.

- Ischemic strokes are caused by lack of blood flow to the brain because of constricted blood vessels. This is the most common cause of stroke in young people with moyamoya syndrome.

- Hemorrhagic strokes are when poorly formed blood vessels burst or tear because of the constricted blood vessels. This type of stroke is less common in young people with moyamoya syndrome.

HOW DO DOCTORS DIAGNOSE A STROKE?
If a person is diagnosed with moyamoya syndrome, it is important for them to see a neurosurgeon (brain and nerve surgeon) with expertise in moyamoya syndrome as soon as possible. Doctors can diagnose a stroke through the following tests:

- Magnetic resonance imaging (MRI) scan. An MRI is a safe and painless test that uses magnets and computer technology to take pictures of the brain. It does not use radiation.

- Magnetic resonance imaging of the arteries (MRA). This is also a safe, painless test that uses magnets and computer technology to take pictures of the arteries in the brain and neck.

- Computerized tomography (CT) scan of the brain. In some cases, doctors may use a special dye to make the pictures clearer. CT scans do use radiation.

What are the signs of a stroke?
- Weakness or difficulty moving arms or legs
- Numbness, tingling or a feeling of heaviness in the arms or legs
- Slurred speech
- Trouble swallowing or food/liquid falling out of the mouth
- Sudden weakness in the dominant hand
- Inability to speak or language that does not make sense
- Unsteadiness or falling to one side
- Facial weakness
- Severe and sudden headaches, possibly with vomiting
- In rare cases, seizures

If your child has any of these symptoms, see a doctor right away or call 911. Children who are actively having a stroke can be given medicine or have treatments that can help lessen the amount of injury to the brain. Remember, TIME = BRAIN!
HOW DO DOCTORS TREAT MOYAMOYA SYNDROME?

Treatment depends on whether a person with moyamoya syndrome has had a stroke. There is no unique treatment for stroke associated with moyamoya syndrome. Surgery can be done to prevent further strokes from occurring.

- **If they have NOT had a stroke and do not have symptoms**, early diagnosis of moyamoya syndrome is an important step in preventing a stroke. Surgery can also prevent a stroke.

- **If they are HAVING a stroke**, doctors focus on treating the stroke. If a stroke has already occurred, there is no unique treatment for moyamoya syndrome; patients will be treated for the stroke, instead. This will likely require admission to an intensive care unit (ICU) for close monitoring and possibly medical or surgical intervention. Surgery may be done after your son/daughter is stable, but this can take several weeks.

- In rare cases, blood pressure medications can help manage symptoms before surgery.

WHERE CAN I LEARN MORE ABOUT MOYAMOYA SYNDROME IN PEOPLE WITH DOWN SYNDROME?

- Down Syndrome Program at Massachusetts General Hospital and MassGeneral Hospital for Children  
  www.massgeneral.org/downsyndrome

- **Boston Children’s Hospital**  
  http://www.childrenshospital.org/centers-and-services/programs/f_-_n/moyamoya-disease-program

- Stanford University Medical Center  
  https://stanfordhealthcare.org/medical-clinics/moyamoya-program

- UCLA Health  
  https://www.uclahealth.org/cerebrovascular/moyamoya-disease

- **Children’s Hospital of Philadelphia (CHOP)**  
  https://www.chop.edu/conditions-diseases/moyamoya-disease

- National Organization for Rare Diseases  
  https://rarediseases.org/rare-diseases/moyamoya-disease/

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