Infants diagnosed with dup15q syndrome are at risk to develop infantile spasms. Diagnosing and treating infantile spasms is an urgent matter.

Infantile spasms are a severe form of epilepsy. The onset is usually in the first year of life, typically between 4-8 months, and rarely after 18 months. Prompt diagnosis and treatment are critical, but this is challenging because infantile spasms can be mistaken as colic, reflux, or a startle reflex.

Dup15q syndrome is one of the most common known causes of infantile spasms. As many as 40% of individuals with seizures present initially with infantile spasms; of this group, approximately 90% subsequently develop other seizure types. Children with dup15q syndrome who have epilepsy, particularly those with infantile spasms are likely to have a greater level of impairment in terms of cognitive and adaptive functioning relative to those without epilepsy. Identifying and controlling seizures early could be extremely beneficial for children with dup15q syndrome.

**Primary Care Physicians must understand the “red flag” words alerting to the possibility of infantile spasms when caregivers are describing symptoms. It is important to note that in an individual child, all of the spasms usually look identical.**

**Movements:**
- small crunches
- belly tensing
- head drops
- head bobs
- uncontrolled movements
- a quick wide-eyed stare/ eyes rolling up
- the raising of the shoulders and arms

**Timing:**
- Length of Individual Spas: 1 sec
- Repeats every 5 to 15 seconds: 15 sec
- Over a span of several minutes: Minutes

Infantile spasms often happen one after another in a series called a cluster with 5 to 15 second pauses in between.

- Clusters may occur once or several times a day.
- Clusters are common several minutes after waking.
- After a cluster, children usually go back to normal behavior.
- In most cases, there is no warning of an oncoming spasm.

Neurologists must begin treatment within the first seven days of an infantile spasms diagnosis to have the best chance to prevent long-lasting symptoms. Physicians should maintain a low threshold of suspicion when considering if a child could have infantile spasms.

If you suspect infantile spasms, refer the child to an urgent visit in front of a neurologist and order an EEG immediately. Most children, but not all, will have EEG readings of hypsarrhythmia. The main treatments include hormonal therapy and vigabatrin, an anti-seizure medicine. Do not hesitate to refer to a neurologist, even if you are unsure of a definitive diagnosis.

Learn more at infantilespasms.org