

Trisomy 21 Update

A Newsletter from the Trisomy 21 Program at The Children's Hospital of Philadelphia

Winter 2007



Fourth Annual Trisomy 21 Symposium

Saturday, March 3, 2007

The *Fourth Annual Trisomy 21 (Down syndrome) Symposium* is scheduled to take place on Saturday, March 3, 2007 at The Children's Hospital of Philadelphia. This one-day symposium will provide parents with up to date clinical information and therapeutic approaches on select topics applicable to individuals with Trisomy 21. This year's topics include Self Advocacy, Neurology, Information on waivers through the MR system in Pennsylvania, Camp Pals and Dentistry. We are fortunate to have another group of dedicated and informative presenters including Erin Riehle from Project SEARCH.

Nearly a decade ago, the Cincinnati Children's Hospital opened a program called Project SEARCH. This program helps place people with cognitive disabilities (many with Down syndrome) in jobs within the hospital and teaches other organizations how to do the same. Project SEARCH provides employment and educational opportunities while remaining dedicated to workforce development that benefits the individual, community and workplace.

For more information on this year's symposium, please contact the Continuing Medical Education department at 215-590-5263.

Spotlight on:

Hillary Kruger, MD

Originally a native New Yorker, Dr. Kruger joined the Division of Child Development at The Children's Hospital of Philadelphia in 2003. "When Dr. Pipan invited me to join the T-21 team in 2005, I jumped at the chance. In recent years, the world has opened up in new ways for people with Trisomy 21. Through improved medical care, access to wider educational opportunities, and the chance to start off with Early Intervention services, children with Trisomy 21 have a greater ability to live healthy, productive and enjoyable lives."

In addition to the T-21 program, Dr. Kruger is a member of the Regional Autism Center staff, and the Cerebral Palsy program. After attending Harvard University as an undergraduate and New York University School of Medicine, she completed her pediatric residency at Bellevue Hospital/NYU. Dr. Kruger completed her fellowship in Developmental Pediatrics at the Rose F. Kennedy Center, part of Einstein Medical Center, Bronx, NY. She is subspecialty board-certified in Developmental and Behavioral Pediatrics.

Occupational Therapy

Children with Down syndrome have a wide variety of unique needs individual to each child and family. Occupational therapy can assist with the development of a variety of skills including fine motor coordination, body awareness, social skills, and sensory processing. These components are important pieces of functional skills like eating, self-care, playing, and learning. Occupational therapy can assist parents in developing ways to engage and interact with their children while providing opportunities for independence and growth. Your primary care physician can provide you with a prescription for occupational therapy. To set up an outpatient evaluation call: 215-590-5819.

Upcoming Educational Sessions:

Look for further information on our upcoming October educational session to follow. Topics for the Saturday half day forum will include: sexuality, educational rights & resources and a fathers perspective on T21.

Trisomy 21 Team

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Ask the Doctor

We welcome all questions! Please send to trisomy21@email.chop.edu.

Questions will be presented anonymously to our T21 team, and answers may appear in upcoming newsletters.

Please submit your questions about caring for children with Down syndrome. Subjects can include child development, neurology or other issues.

Why wear an orthotic?

Helen Milligan, M.P.T.

People with Down syndrome have increased joint mobility, hypotonia, decreased strength and sensory impairments. This affects their postural stability and their ability to achieve new motor skills.

When a person stands their body weight is transferred to their ankles and feet. It requires that the ankle and foot be stable and supportive. When people with Down syndrome stand their feet "fall" into pronation. This is due to the ligamentous laxity which exists at the ankle and foot joint. A pronated foot is an unstable foot.

When a person without ligamentous laxity walks, they place their foot on the ground and the foot and ankle initially move into pronation. This allows the foot to accommodate to the surface and permits shock absorption. Then they rapidly shift their weight to the outside portion of their foot into a supinated position. Now the foot is stable and supportive. This is an ideal posture to lock the foot and get it ready for push off during the swing phase of walking.

People with Down syndrome remain pronated throughout the stance phase of walking. This means they are always standing on an unstable foot. Children with Down syndrome are frequently forced to develop compensatory motor patterns to accommodate for the inherent instability when learning to stand and walk. They may lock their knees, maintain a wide base of support, take short steps, turn their feet out and avoid balance challenges.

Children and adults who persistently pronate may also develop muscle tightness at the outside of their feet, hamstrings, and low back due to these compensations.

The sure step orthotic was designed specifically for children with decreased muscle tone and pronation. It is a flexible supramalleolar orthotic (SMO) which uses compression at the heel to prevent pronation while permitting normal movement at the ankle and toes. Kathy Martin, P.T. completed a study in 2004 on children with Down syndrome who used this orthotic. Her study reported significant immediate and long term improvements in postural stability with the use of this flexible SMO.

Each individual requires a physical therapy evaluation to assess if orthotics are appropriate. Most people with Down syndrome would benefit from a flexible SMO but some will require an ankle foot orthotic (AFO) which controls both the knee and ankle joints. A pediatric physical therapist is able to make the appropriate orthotic recommendation. The appropriate orthotic can be an important adjunct in providing adequate postural stability at the foot and ankle in children with Down syndrome.

Interview with:

C. Alter, MD, Division of Endocrinology

Children with Down Syndrome are at higher risk for endocrinologic disorders. Which disorders are most common in these children?

One of the more common endocrine issues with DS is thyroid disorders. Thyroid disease develops slowly, often over years. However, the laboratory studies will be abnormal well before the onset of symptoms. Thus, the purpose of the thyroid function screening is to diagnose and treat thyroid disease well before the onset of symptoms.

All infants born in the U.S. get tested for hypothyroidism as part of the newborn screening test. Pediatricians are notified if there are any abnormalities. All children with Down Syndrome should have blood drawn for thyroid function tests (free T4 and TSH levels) at age 6 months, 12 months then every year throughout their lifetime, even if a child or adult is asymptomatic.

Hypothyroidism (not having enough thyroid hormone) is most common, affecting approximately 25 percent of individuals with DS. It can have onset at any time in life, causing fatigue, dry skin, constipation, cold intolerance, poor school performance, poor growth, increased weight, a low pulse, delayed puberty, and mood changes. In children less than three, thyroid hormone is required for brain myelination, a key process in brain maturation, and thus hypothyroidism can directly affect cognitive development if untreated.

Children with Down Syndrome can also have hyperthyroidism (too much thyroid hormone). Treatment of hyperthyroidism is more complex and requires careful guidance by an endocrinologist, careful medication followup, and frequent laboratory studies.

How is hypothyroidism treated?

The treatment of hypothyroidism requires daily administration of a small pill (thyroid hormone replacement) which can be swallowed or chewed. In addition, thyroid function tests need to be checked every 3 to 6 months.

What is free T4 and TSH?

T-4 measures the amount of thyroid hormone that is active in the blood. Thyroid hormone is made by your thyroid gland, which is located in the front part of your neck. It is called free T4, because it measures the thyroid hormone that is not bound to protein and thus available for your body to

use. TSH (thyroid stimulating hormone) is another hormone, made in the part of your brain called the pituitary, that tells the thyroid gland to make thyroid hormone. If the TSH is high it means that your brain is sensing that you don't have enough thyroid and is trying to get the thyroid gland to make more.

What do you do when children have elevated TSH but normal T4?

An elevated TSH implies the pituitary is demanding that the thyroid produce more thyroid hormone (T4). An elevation in TSH occurs in 30 to 50 percent of children with DS. When a TSH is only mildly elevated (6-10 uIU/ml) it often goes back to normal without treatment and a repeat thyroid function test is recommended in 3 months. If a TSH is over 10, or if it is steadily rising, then I recommend treatment. Because thyroid hormone is so important to brain development in the first 3 years of life, I will treat younger children with mildly elevated TSH (above 6) with replacement thyroid hormone.

Why do children with DS have thyroid disorders?

We don't know exactly, especially in younger children. In older children and adults thyroid dysfunction is more commonly related to an auto-immune process. We know that children with DS are at higher risk for autoimmune disorders including thyroid disorders and Type 1 diabetes.

Is there any reason to regularly screen for Type 1 diabetes without symptoms?

Unlike thyroid disease which can take years to present, Type 1 diabetes symptoms develop quickly presenting with excessive thirst, urination, and fatigue. More important than annual blood or urine screening, families should be reminded frequently about these symptoms. I don't feel that annual lab screen for diabetes is needed.

Children with DS often have short stature. What role does growth hormone (GH) have to play in children with DS who are very short?

Growth hormone has been studied in children with DS. Most centers feel that growth hormone should be considered in children with DS who test deficient in growth hormone (a minority of cases) and where improvement of adult height by 2-3 inches would dramatically improve the quality of life. Growth hormone only comes as an injectible medication. Thus, if a child is not able to appreciate the differences in height, daily injections of growth hormone (as many as 4,000 total) may not be in the best interest of the child and family.

Resources Available Through the T21 Program

Educational consultant

Our educational consultant is a parent advocate who can assist families in the process of creating an individualized education program (IEP).

Care notebook — Look for the session at the 3/3 symposium.

This is an organizational tool to keep all medical and educational materials in one place, making it easy to retrieve the right piece of information for the doctor, the school, the babysitter and the rest of the family. Call or e-mail to sign up for a one-on-one or group session, at which you will learn how to use the notebook, including which records to keep or shred and how to maintain records on an ongoing basis. Contact us at 267-426-5360 or trisomy21@email.chop.edu.

Symposium — Come join us Saturday, 3/3

The symposium takes place each year in March to inform families and professionals about current trends that will benefit people with trisomy 21.

Resource directory

The resource directory, updated each year, is on the Trisomy 21 Program Web site. Go to www.chop.edu and type “Trisomy 21” in the search box and choose the first option. Then choose “Resources for Families” from the list on the left side of the page. The resource directory is under “Downloadable Material.”

Educational Sessions

These half day sessions will held on Saturday's from 9 a.m.-12 p.m. and will include a variety of educational topics specific to T21. Look for details on the upcoming October session to follow.