DIASTROPHIC DYSPLASIA

Also available on the web: http://pixelscapes.com/ddhelp

With love and gratitude, this booklet is dedicated to the memory of
DR. STEVEN E. KOPITS, friend and healer of diastrophics
and all little people, and to ALL doctors who strive for answers
in their care of little people.

DIASTROPHIC SIBLINGS

DIASTROPHIC DYSPLASIA

by Mary Carten and Vita Gagne

Diastrophic Dysplasia (DD) is a very rare type of dwarfism. It occurs once in every 500,000 births in the United States making it the third largest type of dwarfism. Diastrophic dysplasia is inherited in an autosomal recessive pattern which means that both parents must carry the DD gene for a child to be born with diastrophic dysplasia. For couples who have a child with DD, there is a 25% chance with each subsequent pregnancy that the child will be diastrophic. Prenatal diagnosis can be accomplished by chorionic villus sampling and DNA linkage studies. Ultrasonography is also a highly accurate prenatal diagnostic method.

A diastrophic baby may be born with, or develop, the following characteristics:

1. Severe shortening of limbs
2. Cleft palate
3. Ear deformities (85% of cases)
4. Progressive deformities and contractures of joints (100% of cases)
5. Progressive hip dysplasia (70% of cases; dislocation, 22% of cases)
6. Typical hand deformities, including "hitchhiker" thumbs (100% of cases)
7. Severe clubfoot (almost 100% of cases)
8. Progressive spinal curvature (lumbar lordosis ["swayback"], 100% of cases; scoliosis [s-shaped curves], 80% of cases; cervical kyphosis [abnormal neck flexion], percentage unknown)
9. Early degenerative changes of joints (100% of cases)

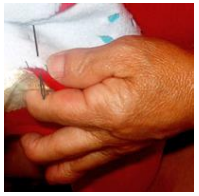
Newborn diastrophic babies average 42 centimeters (16.5 inches) in length. Adult height varies widely, averaging 87-140 cm (34-55 inches). In most infants with DD, the bone connecting the thumb to the wrist is unusually small and oval shaped, causing the thumb to deviate away from the hand and into the so called "hitchhiker" position. Other fingers may also be abnormally short and joints between certain bones of the fingers may be fused together, causing restricted movement of the finger joints.

Despite these limitations, DD children learn to improvise when it comes to their hands. No surgical invention is warranted or needed. If given the opportunity, most DDs are capable of even the most complex manipulation tasks.



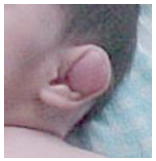




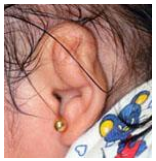



THOSE CAULIFLOWER EARS

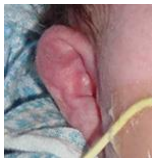
Most newborns with diastrophic dysplasia develop blister-like sacs filled with fluid on the upper part of the ears. These swollen sacs will drain naturally, but most often leave a misshapen upper ear. The new school of thought is to tape gauze pads over the ears when the swelling first appears and use a gauze headband for compression until the swelling subsides, usually within 3-4 weeks. This procedure helps to reshape the ear and keep it close to the head as the swelling diminishes. This swelling of the ears does not affect hearing.



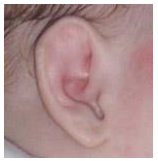
Typical ear swelling



Resolution of same ear



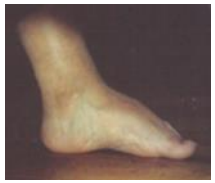
Typical ear swelling #2

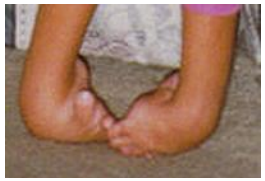


Ear #2 after compression method

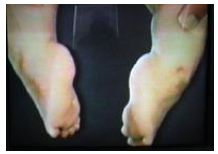
DIASTROPHIC CLUBFEET – UNIQUE AND COMPLEX

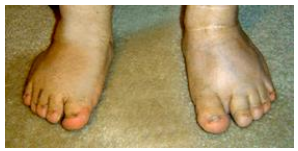
Diastrophic children are usually born with clubfoot deformities, having more components than a clubfoot in an average-sized child. These deformities may range from very mild to severe. Operations to correct this problem are usually done at age 1-2 years, and may include release of the heel tendon and often more involved procedures to correct the other components of the DD clubfoot. The great toe often presents in a "hitchhiker" position requiring correction. Many of these children and adults require custom-made shoes to accommodate their wide feet.


Mild DD foot

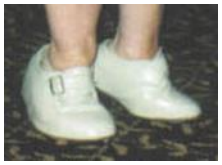

Severe DD feet


Typical DD infant feet


Rear view of feet before correction


Same feet after correction


Regular shoe with added strap


Custom made shoes


Pedorthist adapted athletic shoes


Pedorthist adapted dress shoes

CERVICAL AND SPINAL CHANGES

The trunk can be deformed by excessive lumbar lordosis (swayback) that develops early in life. Scoliosis, which is a side-to-side curvature of the spine, may also begin in infancy, becoming more severe with weight bearing and leading to both trunk deformity and barrel chest. Kyphosis, which is an in and out or s-shaped curvature, accompanies the scoliosis in variable degrees, and the resultant kyphosis [abnormal flexion] of the neck vertebrae, may cause catastrophic neurological problems. Because of these problems, it is very important to have your DD child followed by an orthopedist, preferably a dwarfism specialist, who will monitor your child's spine.

WHY MOBILITY BECOMES A PROBLEM

The combination of marked limitation of motion of all major joints together with a tendency toward dislocation and subluxation [partial dislocation] are characteristic of diastrophic dwarfism. Virtually every joint is likely to develop stiffness. This is due to the severe deformities of the long bones and the growth plates of the long bones, as well as soft tissue contractures, which cause shrinkage and shortening of the muscles. Progressive dislocation of the hips and knees (including the knee cap) are often observed. Hip dislocation occurs in about 22% and hip dysplasia has been reported in 70% of diastrophics.

DOING IT MY WAY

Children with diastrophic dwarfism may reach motor milestones of development more slowly than their average-sized counterparts. Diastrophic dysplasia does not affect intelligence, although speech can be affected by cleft palate. Some average examples of motor milestones among children with DD are as follows:

- Head control, 2.5 months
- Roll side to side, 5-12 months
- Sitting unsupported, 7-12 months
- Crawling, using a commando crawl, 7-18 months
- The diastrophic "scoot", 8-15 months. This is when the child is sitting on the floor scooting along on his/her bottom. You have to see this to appreciate it.
- Pulling up to standing, 12-24 months
- First steps, 15-24 months
- Walking, 24+ months

Many of these children will have had several surgical procedures in the first 48 months of life, delaying accomplishment of "normal" motor milestones. A diastrophic child with

clubfeet and/or dislocated hips could spend months in long-leg casts or a “spica cast” (waist to toes) making mobility impossible. The diastrophic child with cervical kyphosis of the neck will undergo surgery/fusion and be placed into a “halo” neck traction device for months. During these lengthy recoveries, parents can provide extra opportunities for their child to use his/her hands with various toys, games, art projects, etc. Once the casts are removed, physical therapy, including swimming, is recommended to loosen the stiffness and to strengthen the muscles and bones. Depending on the age of the child, potty training, walking and self-dressing might need to be re-taught.


Biking


Swimming Therapy


Physical Therapy

DIASTROPHIC ADULTS

An adult diastrophic usually walks with a forward tilt appearing to be falling forward. This is caused by a number of factors, such as deteriorating hips and knees as well as progressive spinal curvatures. Weight gain may also be a factor as many adults must rely on a motorized scooter for mobility. Hip and knee replacements are helping many adults regain some of their lost mobility. Some adults are fortunate enough to be able to continue using walkers and canes.

ANESTHESIA

The administration of anesthesia to a diastrophic dwarf should be closely monitored. It is **very important that prior to any surgery** that all diastrophics have x-rays of their cervical spine to determine if they have abnormalities, particularly kyphosis or abnormal motion. The placement of the endotracheal tube for breathing during general anesthesia typically requires the neck be hyperextended for the

insertion. This can be dangerous if the cervical spine has kyphosis or abnormal motion since compression of the spinal cord and or nerve roots could occur, leading to paralysis. Infants and children sometimes have very small tracheas and require special skill in tube placement.

PREGNANCY FOR DIASTROPHIC WOMEN

Pregnancy for adult DD females can and does happen. A DD female generally will have an average-sized baby, unless her partner is also a dwarf, and sometimes even then the baby will be average-sized. Consultation with a genetic dwarfism specialist before pregnancy occurs is suggested.

A DD female’s height, general health and internal area (space between bottom of ribs and hips) are some of the factors that determine pregnancy outcome. A complete exam by an obstetrician will determine if a DD female should attempt a pregnancy. The obstetrician will decide what will be a “safe total weight gain”. This is a very important item to remember because it will affect breathing, ability to continue daily activities, the amount of room the baby will have to grow, and the duration of pregnancy. Most women report breathing difficulties as the pregnancy advances. Many pregnant DDs must sleep upright late in their pregnancy in order to breathe normally. There are no reported major complications during pregnancy, other than the usual “morning sickness” that seemed to last longer than expected, elevated blood pressure, cold feet and legs as the pregnancy progressed, and excessive weight gain.

Serial ultrasounds should be done to monitor the baby’s growth and position. All deliveries are done by caesarean section and are usually planned to take place within 2-4 weeks of actual due date. This is a major complication even if it is planned. Tests are done of the baby’s lungs to ensure full development. General anesthesia is used most often, although there have been reports of success with an epidural. In some cases, the baby must remain in the hospital a few extra days to gain those extra ounces before being released. Recovery for the new DD mom should be a speedy and enjoyable time of getting to know her new little treasure.

Mother and Daughter (age 1)

Mother and Daughter (adult)

Fathers can be Diastrophic too!

RESOURCES

Little People of America, Inc.
1-888-LPA-2001
Little People of America (LPA) Online: http://lpaonline.org/

Diastrophic Help website: http://pixelscapes.com/ddhelp/
Online Medical Dictionary:
http://www.nlm.nih.gov/medlineplus/mplusdictionary.html
National Organization for Rare Disorders, Inc.:
http://hw.healthdialog.com/kbase/nord/nord482.htm

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