Neurological Considerations in Short Stature

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I would like to discuss some of the neurological problems commonly encountered in short-statured individuals. Frequently these problems are not recognized by parents, physicians or affected individuals themselves until they have progressed to the point of severe disability. They remain hidden for a variety of reasons: in part because of understandable focus on the cosmetic and mechanical aspects of short stature, in part because of the insidious nature of their progression. However, it is important that they be recognized early so that appropriate corrective action can be taken before they cause significant, and, in many case, irremediable dysfunction.

In this presentation, I would like to begin by summarizing a few general points about neurological dysfunction and about the salient features of all syndromes associated with short stature. After these introductory generalities, I will discuss specific examples neurologic dysfunction in two prototypic short stature syndromes: achondroplasia and the spondyloepiphyseal dysplasias.

Neurologic dysfunction refers to a failure of any portion of the nervous system to function properly, either because of chemical or mechanical dysfunction. Primary dysfunction refers to a failure of the nerve cells to function properly, either because they were not properly assembled or because of failure of a biochemical process intrinsic to these nerve cells. Such disorders are very difficult to treat. Indeed, in most such disorders curative treatment is impossible with currently available technology. Fortunately, such intrinsic nervous dysfunction is limited to only a few short-stature syndromes. More commonly, neurologic dysfunction is secondary. The nervous system is assembled properly and the nerve cells themselves are healthy until they are secondarily compromised by compression. This is fortunate, because prevention or alleviation of such compression can either prevent or reverse such secondary neurological dysfunction.

A second point about neurologic dysfunction is worth remembering. The nervous system contains many different parts, each with a specific function. Certain regions control movement in the arms and legs, others control respiration and control of the bladder, still others govern intellect and memory. For that reason, there can be a selective pattern of symptoms, reflecting with a great deal of precision which parts of the nervous system is being compromised, and which is functioning properly. For purposes of today's discussion I would like to draw your attention to two major subdivisions of the nervous system: the cen*tral nervous system*, comprised of the brain and the spinal cord, which lie under the protective bony coverings of the skull and the spinal column; and the *peripheral nervous* system, which are long cables that exit from these bony coverings to connect the brain and spinal cord with the limbs and the internal organs. Whereas the peripheral nervous system has a great capacity to repair itself after injury, the central nervous system does not. For that reason, special care must be taken to recognize dysfunction of the central nervous system before irreversible damage takes place.

My other introductory remarks concern the classification of short-stature syndromes. Accurate diagnosis is important not only for the geneticist providing counseling information, but also for the neurologist, orthopedic surgeon, and neurosurgeon charged with detection and amelioration of neurologic dysfunction. Different types of short stature predispose to different types of neurologic dysfunction. Predisposition does not imply that each individual with a particular syndrome will necessarily develop those complications. As in all of medicine, each person must be assessed individually. However, such assessment is greatly aided if parents and clinicians are alert to commonly encountered problems.

A major distinction is that between *proportionate* short stature and *disproportionate* short stature. In the former, the individual is short, but bodily proportions are the same as that of average individuals. Proportionate short stature results from nutritional, hormonal or psychosocial factors. Such syndromes are not associated with compressive neurologic dysfunction and will not be discussed further. In contrast, disproportionate short stature is typical of the *skeletal dysplasias*, genetically determined abnormalities affecting formation of bone or cartilage. In such syndromes, compressive neurological problems are common.

Currently, geneticists have recognized over 300 different skeletal dysplasias, each with a distinct pattern of neurologic complications. Fortunately for the neurologist dealing with such a wide spectrum of syndromes, several general principles have emerged. It is useful to consider two major categories of skeletal dysplasia: *short-limbed, of which the most common is achondroplasia*, and *short-trunk as occurs in the spondyloepiphyseal dysplasias.*

Achondroplasia

In achondroplasia, neurologic dysfunction results from *stenosis*, narrowing of a bony channel through which nerves or blood vessels pass. This can occur at either of several levels, resulting in (l) compression at the level of the craniocervical junction, where the upper part of the spinal cord joins with the brain; (2) hydrocephalus, an excessive collection of cerebrospinal fluid inside the head; and (3) compression of the spinal cord. Each of these produces a distinctive pattern of neurologic dysfunction.

The craniocervical junction is where the top of the neck attaches to the bottom of the head. Through this region pass all the nerves that communicate between the brain and the rest of the body, with the exception of the head. In this region are also located the nerve centers that control breathing. Mild compression of the craniocervical junction impairs control of the arms and legs, making them stiff and exaggerating the reflexes. More severe compression interferes with the respiratory centers. If sufficiently severe, respiration can cease and the child can die from asphyxia. Compression occurs for two reasons. First, the *foramen magnum*, literally the big hole at the base of the skull through which the spinal cord passes, is smaller than it should be. Secondly, the foramen magnum in individuals with achondroplasia sits too far forward in the skull, kinking the brainstem backwards. The kinking can inadvertently be exaggerated if the achondroplastic infant is placed in a seated position before developing sufficient control of the neck to prevent the head falling back-

Election

From back page.

Gerry Lurtz has been appointed as the Vice President of Programs by the new Executive Board of LPA. Gerry is currently the District 5 Director, but will go out of office when District 5 has elections at their Regional Meeting this fall.

Mail-In Ballots

As with any new system that is first put into use there were some minor items that will be worked out in the next election in 1994. One item is that you must be 18 or older to vote (this is for both little people and average-size). Because of the way names and types of memberships are keyed into the computer, we must have the age of everyone who wants to receive a mail-in ballot for voting. **To accomplish this, Lee Kitchens, Database Manager, has requested that when you receive your 1993 Membership Renewal Form (in about late October of this year) you must put down your age if you want to receive a ballot. There will be more on this later on with your 1993 Membership Renewal Form, and in *LPA Today*.** ◆

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wards. This kinking is further exaggerated because of the infant's tendency to bend the neck backwards to admit air into a small chest through crowded upper airways. Indeed, a major diagnostic problem for clinicians is to determine if breathing problems in achondroplastic infants result form compression of respiratory centers in the craniocervical junction or from obstruction of the airway. Each of these requires a separate remedy. If the problem lies in the craniocervical junction, a neurosurgeon will unroof the top of the foramen magnum and the top cervical vertebrae, by removing bone. If the problem is in the airway, supplemental oxygen, tonsillectomy and adenoidectomy, or tracheostomy may be required.

A striking feature of all children with achondroplasia is the relatively large size of the head in comparison to the rest of the body. This results, in part, from *hydrocephalus*, the accumulation of an unusually large amount of cerebrospinal fluid inside the ventricles. Cerebrospinal fluid is normally produced inside the ventricles, cavities inside the brain, passes through holes to bathe the outside of the brain and spinal cord, and then is ab-

See NEUROLOGICAL, page 10.

Marge Carlisle
National President

Marge has been a member of Little People of America, and District 5 since 1968. She was a charter member and later President of the Motor City Micronauts Chapter. Marge held the position of District 5 Director from 1983-1989, and in 1986 chaired the National Conference in Dearborn, Michigan.

Marge is married to Butch, and has two sons. Professionally, she is a Special Education teacher in an urban city school.

Ruth Ricker
Senior Vice President

Ruth grew up in LPA and has been fortunate to attend every national conference since 1968. She has held several district and national positions including District One director and was chair of the 1983 national conference in Boston. She is also on the boards of the Human Growth Foundation and the Coalition for Heritable Disorders of Connective Tissue.

Ruth is employed as a Technical Assistance Specialist for the US Department of Education, Office for Civil Rights regional office in Boston. In her spare time she enjoys the Red Sox, movies, reading, music, walking and baby-sitting her two-year-old niece.

James Davis
National Treasurer

James Davis is a native of Georgia, where he currently lives with his wife, Joy They will celebrate their first wedding anniversary in April of this year. They met through the Little People of America.

In 1984 he earned his Bachelor's degree in Business Administration with a concentration in Accounting, and in 1988 I was awarded a Masters degree in Business Administration, both from Kennesaw State College. For the last four years, he has been employed by Boeing Computer Services as a Business Manager of a project with the U.S. Army.

Anthony Soares
Vice President of Public Relations

Anthony has been an active member of LPA since 1963. He has attended many local regional events in District 2 and served as the first Teen Coordinator for four years, eventually rising to Assistant District Director from 1989 to 1991.

Anthony is an Art Director with DDB-Needham Worldwide Advertising. Based at their New York Headquarters he works on print and TV advertising.

A former resident of Manhattan, he now resides in the Hudson river front City of Hoboken, NJ. where he currently serves on many committees for his condominium association. He loves to Rollerblade, collect CD's, and throw parties.

Daniel Margulies
Vice President of Membership

Daniel has been a member of LPA for 20 years, and an officer in some capacity for most of the time. He has served as National President in addition to many other offices. Daniel has also co-chaired two national conferences. In addition to being the Vice President of Membership he serves as Editor of *LPA Today*.

Daniel is a graduate of the University of Arizona with a degree in Mechanical Engineering. He worked for the Department of Defense as a Test Engineer at the nuclear submarine yard in California for 15 years. After that for a couple of years, he worked as Marketing Director for a nonprofit employment agency for people with disabilities. Currently he and his brother now own and operate their own graphic art company.

Ron Piro
Vice President of Resources

Ron Piro joined the LPA in 1986, and has attended District 1 activities regularly, and has attended all the national conferences since Philadelphia in 1987. He was appointed Vermont's first State Coordinator in 1988. He has served as a director on the LPA Foundation Board.

Professionally, Ron is an electrical engineer for IBM in Burlington, Vermont, where he has designed computer chips for the past ten years. He is also active in the community, serving as president of the Georgetown Condominium Association for five years, and as Membership Director for the Northwestern Vermont Model Railroad Society. ◆

WEDDINGS

Penelope Blake and Steve Goodwin were married on September 27, 1991 in Victoria, British Columbia.

LPBC Newsletter, November 15, 1991

Orsola Pannuccio and John Williams were married on June 9, 1991 in Australia. John is from Canada and Orsola is from Australia.

Little People of Ontario News, January, 1992

Connie Valuckas and Nick Mowery were married on July 25, 1992 in Puyallup, Washington. Nick is from Dallas, Texas and Connie is from Washington.

Monica and Butch Patton had a perfectly beautiful wedding. We all wish them the best.

Northcoast Breeze, July 14, 1992

Deborah Stinger and Glenn Vowell (right) were married on May 16, 1992 in Kelowna, British Columbia, Canada, where they will also reside. Deborah is a native to Kelowna, and Glenn is from North Carolina. Deborah is the LPBC Vice President, and Glenn is the LPBC Treasurer.

Little People of British Columbia, June, 1992

Engagements

Karen Bolling and Charles Elder of Norton, Virginia have announced their engagement. No date set at this time.

Delmarva Mini News, March, 1992

Robin Zeltner of Willowbrook, Illinois and **Don Snider** of Georgetown, Illinois. both of District 6 have announced their engagement. A May, 1993 wedding is planned.

New Additions

Danielle Nicole seven months old; parents Herb and Sue Berzins.

Little People of Ontario, June, 1992

Phranquie Mae born on may 11, 1992 to Beth Ann Smith.

Mid-Michigan Chapter, June, 1992

Neurological

From page 9.

sorbed into the blood stream. Blockage of either the cerebrospinal pathways or the veins that drain blood out of the head can cause hydrocephalus. If mild, this will only cause enlargement of the head. In our experience, this is usually a self-limited problem not requiring surgical intervention. However, in some children with achondroplasia, hydrocephalus can be more severe, causing headaches and stiffness of the legs. In more extreme cases, hydrocephalus can cause impairment of intellect, drowsiness, and vomiting. In unusual cases, hydrocephalus can progress to produce blindness and deafness. For these reasons, all infants with achondroplasia should have periodic measurement of head circumference and neurological performance. These measures should then be compared to standards compiled for achondroplastic children. If there is a sufficient departure from expected performance, a neurosurgeon can insert an *intraventricular shunt*, a plastic tube that can drain cerebrospinal fluid from the ventricles into the abdomen or the heart.

Unlike stenosis at the level of the foramen magnum, stenosis in the lower spine usually does not affect individuals with achondroplasia until later in life. The spinal cord and its associated nerves pass through openings inside the bony vertebrae. In achondroplasia, these openings are smaller than they should be. Furthermore, the backward tilt of the lower spine — *the thoracolumbar spine* — typically found in all individuals with achondroplasia, further narrows these passageways. Initially, symptoms are only noticed after prolonged walking. The legs first begin to tingle and get heavy. Sometimes there is mild aching of the lower back. If walking continues, the legs become numb and weak, resulting in a fall. All individuals with achondroplasia learn to avoid such falls by stopping to rest when tingling first occurs. Frequently they will squat or bend forward, reversing the backward curvature — the *lordosis* — of the lumbar spine. Within minutes, the symptoms stop and walking can continue. As thoracolumbar stenosis progresses later in life, the distance that the person can walk before having to rest decreases. It is our experience that if the patient can walk 200 meters without having to rest, the neurological examination is normal at rest and no permanent damage to the nervous system has occurred. However, if the symptoms are ignored beyond this point, permanent weakness, impairment of feeling, and loss of bladder control will occur. For that reason, we recommend that neurosurgical relief of thoracolumbar stenosis be undertaken when symptoms are sufficiently severe.

The spondyloepiphyseal dysplasias

In contrast to achondroplasia, the neurological sequelae of spondyloepiphyseal and many other short-trunk dysplasias, occur because of bony instability rather than stenosis. That is to say, the openings in the spinal vertebrae are sufficiently large to permit easy passage of the spinal cord and its associated nerves. However, the vertebrae are not tightly interlocked. This can cause the bones to shift across each other, stretching the underlying nerves or spinal cord. The shifting can be gradual or it can be abrupt, with potentially serious consequences. Different patterns of such bony instability are seen in many of the skeletal dysplasias.

Dr. Orest Hurko

The most serious pattern of bony instability occurs where the two uppermost vertebrae — the *atlas* and the *axis* — attach to each other and to the *occipital bone* in the base of the skull. In several of these disorders, the *dens*, a bony peg that

inserts into an opening inside the atlas, is not well developed. As a result, these two bones can slide across each other, damaging the underlying spinal cord. Over the years, such atlanto-axial instability results in progressive difficulty with walking, stiffness in the legs, as well as impaired sensation and bladder control. Abrupt atlanto-axial dislocation resulting from a rapid backward jerk, can result in total paralysis or death. Such tragedies can result from a fall, or by manipulation of the neck during induction of anesthesia in preparation for otherwise routine surgery by an operative team that is not mindful of the instability of the neck in such individuals. For these reasons, patients with spondyloepiphyseal dysplasias and related disorders should be carefully assessed clinically and with X-rays of the neck to determine if the instability of the neck is sufficiently severe to require fusion. In such an operation, discussed in greater detail by Dr. Basset, a bone graft is placed to permanently connect these bony elements to ensure that they stay carefully in register.

Instability can occur in the lower spine as well, causing curvature of the back. This curvature can be of several types, depending on the type of skeletal dysplasia. In some dysplasias, curvature resulting from insecure attachment of the vertebral bodies results in a forward curve — a k*yphosis.* In others, the resulting rotation of the spine is from side to side — a *scoliosis. Kyphoscoliosis is* the occurrence of rotation in both directions. In addition to being cosmetically disfiguring and uncomfortable, such abnormal curves can alter the configuration of the chest wall, impairing breathing, and stretch the underlying spinal cord, weakening the legs and the bladder. It is important that such curves be identified early in life. Uncorrected, they will progress until the child reaches maturity, worsening the resulting damage. In some instances, such progression can be halted by application of an external brace. In other instances, fusion with bone grafts or internally placed metal rods is required.

Conclusion

In this brief overview, we have discussed briefly some of the neurological complications encountered commonly in some of the skeletal dysplasias associated with short stature. Knowledge of such basic syndromes are important both to parents and physicians, both to prevent avoidable injuries and to undertake corrective action before permanent neurologic damage occurs. However, it should be stressed that this has been a compilation of what *can* happen as opposed to what necessarily *will* happen. As in all of clinical medicine, each case needs to be evaluated individually. In this way unnecessary surgery can be avoided for those individuals who do not need it, but proper corrective care can be given to these individuals who do. ◆

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by Angela Muir Van Etten

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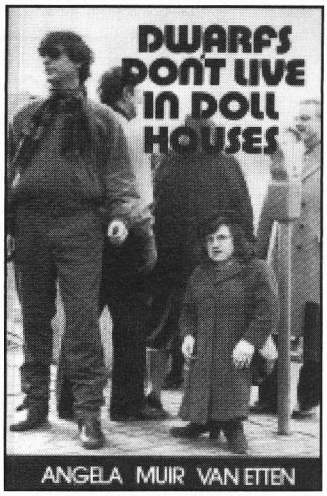

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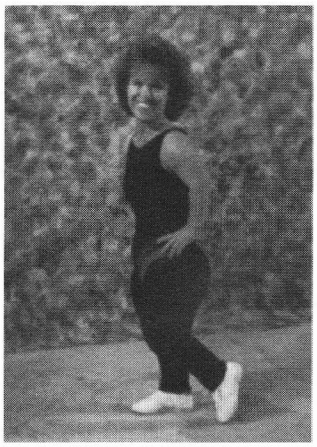

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