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Perspective

A Short Note on Achondroplasia

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DESCRIPTION

Achondroplasia is a genetic disorder whose major feature is dwarfism. In this condition, the arms and legs are short, while the torso is typically of normal length. Persons affected have an average adult height of 131 centimeters for males and 123 centimeters for females. Treatments can include support groups and growth hormone therapy. Struggle to treat or prevent complications such as obesity, hydrocephalus, obstructive sleep apnea, middle ear infections or spinal stenosis can be required. Achondroplasia is the most common cause of dwarfism and affects about 1 in 27,500 people. Most individuals with achondroplasia can be predictable to have a normal life expectancy.

Symptoms

People with achondroplasia usually have normal intelligence levels. Their side effects are physical, not mental. At birth, a child condition will have a short stature that's considerably below average for age and sex, short arms and legs, particularly the upper arms and thighs, in comparison to body height, short fingers in which the ring and middle fingers can also point away from each other, a suspiciously large head compared to the body, an abnormally large, prominent forehead, an underdeveloped area of the face between the forehead and upper jaw.

Health problems in an infant have decreased muscle tone, which may cause delays in walking and other motor skills, apnea, which contains brief periods of slowed breathing or breathing that stops, hydrocephalus, or "water on the brain", spinal stenosis, which is a contraction of the spinal canal that can compress the spinal cord. Children and adults with achondroplasia may have difficulty in bending their elbows, be obese, experience regular ear infections due to narrow passages in the ears, develop bowed legs, develop an abnormal curvature of the spine called kyphosis or lordosis, develop new or more severe spinal stenosis.

Causes

Achondroplasia is produced by a mutation in *Fibroblast Growth Factor Receptor 3 (FGFR3)* gene. This gene is primarily responsible for making the protein, fibroblast growth factor receptor 3. This protein gives to the production of collagen and other structural components in tissues and bones. When the *FGFR3* gene is mutated, it interferes with protein interacts with growth factors leading to difficulties in bone production. Cartilage is unable to develop into bone, causing the individual to be unreasonably shorter in height. Achondroplasia can be inherited through autosomal dominance. In couples where one partner has achondroplasia, there is a 50% chance of transient of the disorder onto their child in every pregnancy. In some situations where both parents have achondroplasia there is a 50% chance to the child that he/she may have achondroplasia, 25% chance the child will not, and a 25% chance that the child will inherit the gene from both parents resulting in double dominance and leads to severe or lethal bone dysplasia.

Epidemiology

Achondroplasia is one of numerous congenital conditions with related presentations, such as orthogenesis imperfecta, multiple epiphyseal dysplasia tarda, achondrogenesis, osteoporosis, and thanatophoric dysplasia. One comprehensive and long-running study in the Netherlands found that the prevalence determined at birth was only 1.3 per 100,000 live births. Another study at the same time found a rate of 1 per 10,000.

Diagnosis

Diagnostic tests may include measurements, appearance, imaging technology, genetic tests, family history and hormone tests. A regular part of medical exam is the measurement of height, weight and head circumference of child. At every visit, pediatrician will plot those measurements on a chart to show child's current percentile status for each one. This is essential for identifying abnormal growth, such as delayed growth or a disproportionately large head. Many different facial and skeletal features are associated with each of several dwarfism disorders.

Children's appearance may also help the pediatrician to make a diagnosis.

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Doctor may order imaging studies, such as X-rays, because certain abnormalities of the skull and skeleton can designate which disorder the children is suffering with. In case of growth hormone deficiency, several imaging devices may also reveal the delay in maturation of bones. A Magnetic Resonance Imaging (MRI) scan may reveal abnormalities of the pituitary gland or hypothalamus, of which both play a vital role in hormone function.

Genetic tests exists various known causal genes of dwarfism-related disorders, but these tests often aren't required to make an accurate diagnosis. Healthcare professionals suggest a test only to differentiate among possible diagnoses when other evidence is indistinct. If pediatrician believes that the children might have Turner syndrome, then a special lab test may be done that evaluates the X chromosomes which are extracted from blood cells.

Pediatrician may take a history of prominence in siblings, parents, grandparents or other relatives which helps to determine whether the average range of height in your family includes short stature. Doctor may suggest hormone test that measure levels of growth hormone or other hormones that are dangerous for childhood growth and development.

Treatment

The treatment may include hormone therapy, limb lengthening, ongoing health care, surgical treatments, etc.

Surgical procedures may precise complications in people with disproportionate dwarfism which include improving the direction in which bones are growing, stabilizing and improving the shape of the spine, increasing the size of the opening in bones of the spine (vertebrae) to improve pressure on the spinal cord and placing a shunt to remove excess fluid around the brain (hydrocephalus), if it occurs.

Lifestyle and home remedies

Issues predominantly critical for children with inconsistent dwarfism include use an infant car seat with stable back and neck supports; avoid infant devices such as swings, umbrella strollers, carrying slings, jumper seats and backpack carriers; adequate head and neck support; provide good posture by providing a pillow for the lower back and a footstool when child is sitting; healthy diet; encourage participation in appropriate entertaining activities, such as swimming or bicycling, better to avoid sports that involve collision or impact, such as football, diving or gymnastics.

CONCLUSION

Achondroplasia is a rare disease that includes unusual shortness. It is a genetic disorder. Although the future may include pathway driven therapies, there will be proceeding with a requirement for quality clinical examinations about the regular history and ideal mediations for the squeals of achondroplasia.