Hyperostosis Frontalis Interna

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DISEASE OVERVIEW

Hyperostosis Frontalis Interna is characterized by the thickening of the frontal bone of the skull. It is not clear that this disorder is actually rare. Some clinicians believe that it may be a common abnormality found in as many as 12 percent of the female population. The disorder may be found associated with a variety of conditions such as seizures, headaches, obesity, diabetes insipidus, excessive hair growth and sex gland disturbances. Increased serum alkaline phosphatase and elevated serum calcium may occur.

SYNONYMS

- Endostosis Crani
- Hyperostosis Calvariae Interna
- Morgagni-Stewart-Morel Syndrome

SIGNS & SYMPTOMS

The major feature of Hyperostosis Frontalis Interna is excessive growth or thickening of the frontal bone of the head. This excess growth can only be seen in an x-ray. As a result, scientists feel that this condition may be much more prevalent than suspected, but often goes undetected. Many people have no apparent symptoms.

Other conditions that may be found in patients with this disorder are: obesity, a condition in which secondary male sexual traits are acquired by a female (virilization); a central nervous system disorder characterized by a sudden, aimless, uncontrollable discharge of electrical energy in the brain causing a convulsion or loss of

consciousness (epilepsy); decreased vision; headaches; disturbances of the ovaries and testes (sex glands or gonads); excessive body hair; and/or diabetes. (For more information on these disorders, choose "Epilepsy" and/or "Diabetes" as your search terms in the Rare Disease Database).

CAUSES

Hyperostosis Frontalis Interna has been found in multiple generations suggesting that the disorder may be inherited as a dominant trait. It is not known if the disorder is autosomal dominant or X-linked. There are no known cases of male-to-male (father to son) transmission.

Human traits, including the classic genetic diseases, are the product of the interaction of two genes, one received from the father and one from the mother.

In dominant disorders, a single copy of the disease gene (received from either the mother or father) will be expressed "dominating" the other normal gene and resulting in the appearance of the disease. The risk of transmitting the disorder from affected parent to offspring is 50 percent for each pregnancy regardless of the sex of the resulting child.

AFFECTED POPULATIONS

Hyperostosis Frontalis Interna affects females 9 times more often than males. This disorder presents itself most often among the middle-aged and elderly but has also been found in adolescents.

DISORDERS WITH SIMILAR SYMPTOMS

Symptoms of the following disorders can be similar to those of Hyperostosis Frontalis Interna. Comparisons may be useful for a differential diagnosis:

Acromegaly is a slowly progressive, chronic metabolic disorder in which an excess of growth hormone causes abnormal enlargement of various tissues of the body and unusual height. Most conspicuously affected are the extremities, jaws, and face. The enlargement of soft tissue, especially of the heart, is a serious feature of this disorder. High blood pressure (hypertension) may be another serious consequence of Acromegaly. (For more information on this disorder choose "Acromegaly" as your search term in the Rare Disease Database.)

Paget's Disease is a slowly progressive disease of the skeletal system characterized by abnormally rapid bone breakdown and formation, leading to the development of bones that are dense but fragile. This disorder usually affects middle-aged and elderly people and most frequently occurs in the spine, skull, pelvis, thighs and lower legs. When it occurs in the skull it can cause hearing loss. (For more information on this disorder choose "Paget's" as your search term in the Rare Disease Database.)

Leontiasis Ossea or Virchow's Disease is a disorder in which there is an overgrowth of the bones of the face and sometimes of the cranium. This disorder causes a general enlargement and distortion of all the features.

The following disorders have been found in association with Hyperostosis Frontalis Interna. They are not necessary for a differential diagnosis:

Crouzon Disease is a genetic disorder characterized by abnormalities in the skull, face, and brain caused by premature hardening of the skull. The skull is made up of several bony plates initially joined by fibrous connective tissue which normally fuse together and harden over a period of several years after growth of the brain. Facial deformities are often present at birth and may progress with time. Vision disturbances and deafness may develop in some cases. (For more information on this disorder choose "Crouzon" as your search term in the Rare Disease Database.)

Galactorrhea is a condition in which there is a spontaneous flow of milk from the nipple.

Myotonic Dystrophy is an inherited disorder involving the muscles, vision, and endocrine glands. It can cause mental deficiency and loss of hair. Onset of this disorder commonly occurs during young adulthood although it can occur at any age and is extremely variable in degree of severity. Symptoms of this disorder may be tripping, falling, difficulty in moving the neck, lack of facial expression and a nasal sounding voice. (For more information on this disorder choose "Myotonic Dystrophy" as your search term in the Rare Disease Database.)

Diabetes Insipidus is due to an abnormality of anti-diuretic hormone (vasopresin or ADH) originating in the posterior lobe of the pituitary gland. The lack of this hormone on the kidney causes excretion of excessive quantities of very dilute (but otherwise normal) urine.

Excessive thirst is the major symptom of this disorder. (For more information on this disorder, choose "Diabetes Insipidus" as your search term in the Rare Disease Database.)

STANDARD THERAPIES

There is no known treatment for Hyperostosis Frontalis Interna. Seizures and headaches can be treated with standard medications.

Genetic counseling may be of benefit for patients and their families. Other treatment is symptomatic and supportive.

CLINICAL TRIALS AND STUDIES

Information on current clinical trials is posted on the Internet at www.clinicaltrials.gov. All studies receiving U.S. government funding, and some supported by private industry, are posted on this government web site.

For information about clinical trials being conducted at the NIH Clinical Center in Bethesda, MD, contact the NIH Patient Recruitment Office:

Tollfree: (800) 411-1222

TTY: (866) 411-1010

Email: prpl@cc.nih.gov

For information about clinical trials sponsored by private sources, contact:

www.centerwatch.com

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Programs & Resources

RARECARE® ASSISTANCE PROGRAMS

NORD strives to open new assistance programs as funding allows. If we don't have a program for you now, please continue to check back with us.

ADDITIONAL ASSISTANCE PROGRAMS

MedicAlert Assistance Program

NORD and MedicAlert Foundation have teamed up on a new program to provide protection to rare disease patients in emergency situations.
https://rarediseases.org/patient-assistance-programs/medicalert-assistance-program/

Rare Disease Educational Support Program

Ensuring that patients and caregivers are armed with the tools they need to live their best lives while managing their rare condition is a vital part of NORD's mission.
https://rarediseases.org/patient-assistance-programs/rare-disease-educational-support/

Rare Caregiver Respite Program

This first-of-its-kind assistance program is designed for caregivers of a child or adult diagnosed with a rare disorder.
https://rarediseases.org/patient-assistance-programs/caregiver-respite/

PATIENT ORGANIZATIONS

Children's Craniofacial Association (CCA)

NORD Member

Email: contactcca@ccakids.com

https://rarediseases.org/organizations/childrens-craniofacial-association-cca/

FACES: The National Craniofacial Association

NORD Member

Email: info@faces-cranio.org

https://rarediseases.org/organizations/faces-the-national-craniofacial-association/

NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases

Phone: 301-495-4484 Email: NIAMSinfo@mail.nih.gov

Fax: 301-718-6366

https://rarediseases.org/organizations/nih-national-institute-of-arthritis-and-musculoskeletal-and-skin-diseases/