RARE BONE DISEASE ALLIANCE

RBDA
RARE BONE DISEASE ALLIANCE

The Rare Bone Disease Alliance, originally organized in 2006 as a patient advocacy network, evolved into the Rare Bone Disease Alliance which includes patient organizations and thought leaders in the rare bone disease field.

The Alliance missions are: stimulating education on rare bone disease through working with professional medical and scientific societies, organizing its own meetings, and assisting patients and families affected by rare bone disease.

Alliance Patient Organizations

FD/MAS ALLIANCE

Fibrous Dysplasia Foundation

GACI Global
circulating hope

ifopa

International Fibrodysplasia Ossificans Progressiva Association (IFOPA)

THE JANSEN'S FOUNDATION

The Jansen's Foundation

"Reaching for a Cure"
LGDA
Lymphangiomatosis & Gorham's Disease Alliance

Lymphangiomatosis & Gorham's Disease Alliance

LMI
Lymphatic Malformation Institute

Lymphatic Malformation Institute

cause treatment cure
Melorheostosis ASSOCIATION

Melorheostosis Association

The MHE Research Foundation
Wings of HOPE as we REACH for the CURE to Multiple Hereditary Exostoses

The MHE Research Foundation

OSTEOGENESIS IMPERFECTA
OI
FOUNDATION
Unbreakable Spirit

Osteogenesis Imperfecta Foundation

The OsteoPETrosis Society

The Osteopetrosis Society

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Finding the Key to HPP

Soft Bones: The U.S. Hypophosphatasia Foundation

Sophie's Neighborhood

Sophie's Neighborhood

XLH NETWORK

XLH Network

The FD/MAS Alliance (previously known as Fibrous Dysplasia Foundation) is a community-led, 501(c)3 not-for-profit organization that works tirelessly to advocate for patients' needs. We help advance research to understand **Fibrous Dysplasia/McCune-Albright syndrome (**FD/MAS) and develop treatments while fostering a supportive, inclusive, and empowered community.

Each and every day, we try to nurture close collaborative efforts between patients and our Scientific and Medical Advisory Councils. Their volunteer members include world-class scientists and clinicians with expertise in FD/MAS pathophysiology, metabolic bone disease, and endocrinology.

Living with a rare disease is tough, and more often than not, FD/MAS patients often struggle to find answers to questions about their condition and connect with clinicians with FD/MAS expertise. For more than 15 years, the FD/MAS Alliance has improved patients' well-being through education, support, and research. Our recently revamped FD/MAS Alliance website provides patients with free educational content and the ability to contact and access members of the Medical Advisory Council and clinicians in their area equipped to evaluate and treat FD/MAS. Because patients are dispersed worldwide, FD/MAS Alliance exists primarily as an online community, regularly reaching thousands of patients and caregivers. Web-based educational programming helps patients navigate complex medical challenges and find experienced clinicians.

The name of this condition, FD/MAS can be confusing. Fibrous dysplasia (FD) and McCune-Albright syndrome (MAS) are debilitating diseases caused by a mutation of the same gene. FD is a condition in which bone is replaced by fibrous tissue, resulting in fractures, deformity, and pain. MAS is classically defined as the combination of FD, excess hormone levels, and cafe-au-lait birthmarks. An activating mutation of the same gene causes FD and MAS, and researchers have come to understand that both diseases exist on a spectrum.

Patient experiences depend on when the mutation occurred during their development and how many different cell types in their body are affected. Diagnosis and treatment can be difficult. The mutation's occurrence is uncommon and completely random, and cannot be inherited. FD/MAS affects people across gender, race, location, and socioeconomic backgrounds. We can address immediate needs, and more focused than ever, we are working to do more.

Research. Advocate. Collaborate.

For more information about the FD/MAS Alliance EIN: 02-0715210, please contact Adrienne McBride, Executive Director at amcbride@fibrousdysplasia.org or visit our website: www.fdmasAlliance.org

GACI Global began in 2019 when four families came together to form a 501(c)3 organization. The mission of GACI Global is to connect families affected by Generalized Arterial Calcification of Infancy (GACI) or Autosomal Recessive Hypophosphatemic Rickets Type 2 (ARHR2) caused by ENPP1 or ABCC6 Deficiencies to each other and to the medical community. The organization strives to provide current educational resources and supports ongoing research.

GACI is an ultra-rare genetic disease that primarily affects the circulatory system. It causes an abnormal buildup of calcium within the walls of the arteries that can reduce blood flow to vital organs. GACI occurs in roughly 1/200,000 births and usually affects infants during the first 6 month of life. GACI occurs in males and females equally in populations all across the world. In addition to arterial calcification, individuals with GACI can exhibit a variety of medical features including respiratory distress, gastrointestinal issues, joint calcification, hearing loss, high blood pressure, stroke and heart failure. The survival statistics for GACI are currently estimated to be around 50%. Those who survive GACI may develop a rare form of rickets known as ARHR2. This can result in bone and joint pain, bone deformities, dental problems, calcification of ligaments and short stature. Presently, there is no curative treatment available for GACI/ ARHR2. However, there have been significant advancements in GACI/ARHR2 research including early stage research to develop enzyme replacement therapy.

For more information about GACI Global:

Christine O'Brien Phone: (617) 286-6202 Email: info@gaciglobal.org

Facebook: www.facebook.com/gaciglobal Website: www.gaciglobal.org

Fibrodysplasia ossificans progressiva (FOP) is one of the rarest, most disabling genetic conditions known to medicine; it causes bone to form in muscles, tendons, ligaments and other connective tissues. Bridges of extra bone develop across joints, progressively restricting movement and forming a second skeleton that imprisons

the body in bone. There are no other known examples in medicine of one normal organ system turning into another. The International FOP Association (IFOPA) is a 501c3 nonprofit organization that was founded in 1988 by Jeannie Peeper, a young woman with FOP, whose goal was to bring people with FOP together. Within one year of our founding the very first family fundraiser was held. It started the IFOPA's legacy of funding FOP research as we work to achieve our vision, a cure for FOP, accessible worldwide.

The IFOPA's mission is to fund research to find a cure for FOP while supporting, connecting and advocating for individuals with FOP and their families, and raising awareness worldwide. In addition to funding research, we support, educate and advocate for people with FOP and their families worldwide. We also bring researchers, clinicians and pharmaceutical companies together to discuss important questions and challenges of developing a treatment for FOP. Our most significant initiative in support of research is the FOP Patient Registry - the largest and most in-depth database of clinical and medical information about FOP and those who are living with the disease. The Registry includes both a patient and medical portal, as well as a patient-reported outcomes survey. We also provide FOP grant funding through a competitive application process and offer an open-access FOP mouse model and FOP Biobank. Patient and family education and support include an annual Family Gathering, webinars, mentorship, and Resilient Living and Independent Living programs. The place to come to learn about FOP, find community and keep up with the latest FOP clinical studies and trials (there are currently eight FOP trials in various phases) is ifopa.org.

Contact:

Michelle Davis, Executive Director
International FOP Association
+ 1 (816) 809-2772 cell | michelle.davis@ifopa.org
ifopa.org | facebook.com/ifopa | twitter.com/ifopa | instagram.com/cure_fop | youtube.com/ifopa

Jansen's Metaphyseal Chondrodysplasia (JMC) is an extremely rare skeletal dysplasia characterized by progressive growth plate abnormalities that affect most of the long bones.

JMC is usually diagnosed during childhood, based on a combination of radiographic and biochemical abnormalities. However, some patients are not diagnosed until adulthood.

The limbs of affected individuals typically show progressive changes that are caused by an abnormal regulation of chondrocyte growth and differentiation, eventually leading to short and bowed legs. X-rays during infancy show

demineralization and rickets-like metaphyseal changes. Fraying of the metaphyses is seen during childhood. Long bones do not grow normally and result in severely reduced adult height.

There is usually severe hypercalcemia and hypophosphatemia despite normal or undetectable serum levels of PTH or PTHrP.

Four different mutations in the gene encoding the PTH/PTHrP receptor (PTHR1) have been identified in several unrelated Jansen's patients.

The mission of the Jansen's Foundation is to bring awareness and support research in hopes of bringing about a cure to this debilitating skeletal condition.

Contact:

Neena Nizar, Ed.D. President & Founder The Jansen's Foundation
PO BOX 115, Elkhorn, NE, 68022
Phone: 402-457-9886
www.thejansensfoundation.org
Email: neenan@thejansensfoundation.org

"Reaching for a Cure"
LGDA
Lymphangiomatosis & Gorham's Disease Alliance

The Lymphangiomatosis & Gorham's Disease Alliance (LGDA) is a patient-support foundation concerned with a spectrum of complex lymphatic anomalies (CLA's). CLA's is an umbrella term encompassing four overlapping but distinct rare diseases of the lymphatic system: Gorham-Stout Disease (GSD); generalized lymphatic anomaly (GLA); kaposiform lymphangiomatosis (KLA); and central conducting lymphatic anomaly (CCLA). CLA's are characterized by growth of abnormal lymphatic tissue involving both soft tissue and bone leading to bone loss, chronic pain and debilitating conditions which can be fatal. They can present at any age, but mostly affect children and young adults. CLA's do not show inheritability, and affect, with varying frequency, different bones - among them: the maxilla, mandible, clavicle, ribs, cervical vertebrae, femur and calvarium.

The LGDA focuses in two primary areas – patient support and research support. Through our continuing patient outreach we collect patient data in our International Lymphatic Malformation Registry www.lgdaregistry.org

Together with our biorepository, we promote and support collaboration among our international research network in conjunction with our research partner, the Lymphatic Malformation Institute www.lmiresearch.org

For information about the LGDA Patient Registry, Vascular Anomalies Centers,
or our International Research Network, contact: Lisa Klepper
Lymphangiomatosis & Gorham's Disease Alliance
email: lklepper@lgdalliance.org website: www.lgdalliance.org

LMI
Lymphatic Malformation Institute

The LMI is dedicated to accelerating research and ultimately finding cures, for patients with Complex Lymphatic Anomalies (CLAs)

We support our global community of researchers, medical professionals and patients by offering information, networking and funding to generate hope and solutions

Complex Lymphatic Anomaly (CLA) is an umbrella term encompassing four overlapping but distinct rare diseases of the lymphatic system; Gorham Stout Disease (GSD); Generalized Lymphatic Anomaly (GLA); Kaposiform Lymphangiomatosis (KLA); and Central Conducting Lymphatic Anomaly (CCLA). CLAs are characterized by growth of abnormal lymphatic tissue involving both soft tissue and bone leading to chronic and debilitating conditions that can be fatal.

The LMI was established in 2010 to support research that drives our understanding of the pathobiology and genetics of CLAs. The institute also supports and funds clinical and translational research to better define the impact of CLAs on our patients, testing of new therapies and finding biomarkers of disease activity and treatment response.
LMI fosters an interactive and collaborative research community inclusive of patients and research and medical professionals, where our research drives improved quality of life and ultimately cures for patients with CLAs.

For more information about CLA research, visit our website at www.lmiresearch.org. Or to submit an application for funding, please contact Dr. Michael Dellinger, Director of Research, at mdellinger@lmiresearch.org.

Melorheostosis is a rare and progressive disorder characterized by hyperostosis (thickening) of the cortical bone. Melorheostosis affects both bone and soft tissue growth and development.
Melorheostosis can result in severe functional limitation, extensive pain, soft tissue contractures, limb length inequality, and limb, hand and/or foot deformity. The age of diagnosis is typically based on the severity of onset and symptoms. On x-rays, the appearance of melorheostosis been likened to flowing, melted candle wax. Melorheostotic bone and soft tissue often do not react as unaffected tissue would to traditional surgical interventions. Melorheostosis patients are literally “One in a Million.”

The Melorheostosis Association is a 501(c)(3) not-for-profit organization dedicated to finding the cause, treatments and cure for melorheostosis. Our focus is on promoting greater awareness and understanding of this progressive disease and its manifestations through education, research, communication and advocacy efforts on behalf of those affected by it as well as those dedicated to alleviating it.

The National Institutes of Health (NIH) is conducting a Long Term Study of Melorheostosis and has identified two genes which cause the majority of Melorheostosis cases — MAP2K1 and SMAD3. NIH has also contributed greatly to the published information on Melorheostosis. We are extremely grateful to NIH and every participating patient as NIH continues its invaluable work in 2020.

The Melorheostosis Association - www.melorheostosis.org
410 East 50th Street, New York, N.Y. 10022.
Kathleen Harper, Founder and Chairman - Kathleen@melorheostosis.org

The MHE Research Foundation

Wings of HOPE as we REACH for the CURE to Multiple Hereditary Exostoses

Multiple Hereditary Exostoses / Multiple Osteochondroma Syndrome is a genetic bone disorder in which benign cartilage-capped bone tumors form throughout the body. Some patients may have as few as two tumors, but

most patients develop many more and the numbers of tumors can be in the hundreds. Exostoses/Osteochondromas can cause numerous problems, including compression of peripheral nerves or blood vessels; irritation of tendons and muscles resulting in pain; loss of range of motion; skeletal deformity; short stature; limb length discrepancy; chronic pain and fatigue; mobility issues; early onset arthritis; an increased risk of developing malignant tumor transformation (chondro-sarcoma) reported risk of 2%-5% over life time and autism relevant behavioral phenotype. It is not uncommon for MHE/MO/HME patients to undergo numerous surgical procedures. Surgery, physical therapy and pain management are currently the only options available. The prevalence of MHE/MO/HME is 1/50,000.

The MHE Research Foundation is a nonprofit 501 (c)(3) organization working in support of researchers, physicians and families dealing with Multiple Hereditary Exostoses / Multiple Osteochondroma Syndrome.

Our Mission is to **REACH**:
Research – to support researchers in order to one day find a treatment/cure. **E**ducation – to provide clinical information, to benefit both families and physicians. **A**dvocacy – to bring awareness about this disease throughout the world.
Clinical – to provide resources enabling families to locate the medical care.
Hope – the research, clinical care and informational resources will bring a better quality of life to the families affected by this disease.

For more information:
Sarah Ziegler, Vice-President & National Director of Research The MHE Research Foundation
8019 Harbor View Terrace, Brooklyn, NY 11209 Phone: 561-315-9149
Email: sziegler@paleyinstitute.org
Web site: www.mherf.org

IMPERFECTA OI FOUNDATION
Unbreakable Spirit®

Osteogenesis imperfecta (OI), also referred to as brittle bone disease, is a rare and variable genetic disorder that is characterized by fragile bones. OI is caused by a mutation in a gene that affects bone formation, bone strength, and the structure of other tissues. The incidence of OI is approximately 1 in 15-20,000 live births. People with OI experience broken bones from infancy through puberty. The frequency typically decreases in the

young adult years but may increase again later in life. Other than broken bones, respiratory issues including asthma are often seen. Short stature, rib cage deformities and spine curves can make breathing problems more severe. Other medical characteristics may include hearing loss, brittle teeth, loose joints, cardiac issues, and basilar invagination.

The OI Foundation is committed to the mission of improving the quality of life for individuals affected by OI through research to find treatments and a cure, education, awareness and mutual support. As a strong advocate for and partner of the Rare Disease Clinical Research Network's Brittle Bone Disorders Consortium, the OI Foundation collaborates with physicians, researchers, and other medical advisors to accelerate a better understanding of OI and improve treatment options for people living with OI.

A few major activities of the OI Foundation include the OI Registry, national and regional conferences, meetings and resources for clinicians and medical professionals, and newsletters and podcasts for constituents. In partnership with the Rare Bone Disease Alliance, the OI Foundation hosts the monthly Rare Bone Disease teleECHO Clinic Series, an educational program designed to build capacity to better diagnose and treat rare bone diseases safely and effectively. The OI Foundation also hosts the OI teleECHO Clinic Series, which focuses on topics specific to osteogenesis imperfecta.

The OI Foundation is the only patient advocacy organization in the United States dedicated to meeting the needs of the approximately 50,000 people living with OI.

For more information, please contact the OI Foundation at
656 Quince Orchard Rd., Suite 650, Gaithersburg, MD 20878
www.oif.org
(844) 889-7579 or Bonelink@oif.org

The OsteoPETrosis Society

Osteopetrosis is a rare congenital bone disorder in which bones are abnormally dense and brittle. This results from an imbalance between the formation and breakdown in the normal maintenance of the bone. There are several types of osteopetrosis of varying severity. Symptoms often include frequent fractures, infections, and chronic anemia and blindness or deafness can occur.

The mission and purpose of The OsteoPETrosis Society is to provide education and support to patients and caregivers, as well as medical professionals who are dealing with osteopetrosis. In addition, we plan to support research into an osteopetrosis treatment and cure.

We hold annual patient meetings with informative sessions dealing with various aspects of the disease. Literature for people with osteopetrosis have been published. A Medical Advisory Panel, comprised of doctors with extensive knowledge and experience with osteopetrosis guide us in these goals and insure that our information is accurate.

For more information:

Patrick Birdsall, President,
The OsteoPETrosis Society
448 Mine Road, Asbury, NJ 08802
Email: patrick@osteopetrosis.org

Soft Bones
Finding the Key to HPP

Soft Bones, The US Hypophosphatasia Foundation, was formed in 2009 to provide information and a community to educate, empower and connect patients living with hypophosphatasia (HPP), their families and caregivers.

Hypophosphatasia is an inherited (genetic), ultra-rare, metabolic (chemical) bone disease of broad-ranging severity that causes life-threatening disease in approximately one per 100,000 live births. People with the condition have low levels of the enzyme alkaline phosphatase, which impairs the mineralization of bones. Normal mineralization is essential for hard and strong bones. Without it, bones become weak and soften and teeth may fall out prematurely. Depending on the severity of the skeletal disease, symptoms can include deformity of the limbs and chest, pneumonia, recurrent fractures, premature tooth loss and pain. While there is currently no cure for hypophosphatasia, treatment is directed towards preventing or correcting the symptoms or complications.

The Soft Bones Foundation catalyzes research of this rare bone disease through awareness, the CoRDS International Hypophosphatasia Contact Registry, and fundraising efforts. Soft Bones also advocates for access to treatment and educates lawmakers and brings attention to the needs and gaps in the care of patients affected by hypophosphatasia around the globe.

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For more information visit www.SoftBones.org.

Sophie's Neighborhood is a 501(c)(3) organization formed to promote research that will lead to an effective therapeutic or cure for a severely crippling bone and kidney disorder, Multicentric Carpotarsal Osteolysis Syndrome ("MCTO").

MCTO is an ultra-rare genetic disorder affecting around 50 patients worldwide. It is caused by amino acid substitutions over a short domain in the MafB gene, in which all mutations appear within a section of pairs in the transactivation domain. Heterozygous, gain-of-function missense variants in the N-terminal transactivation domain cause the progressive disease phenotype.

The disorder is characterized by aggressive osteolysis of the carpal and tarsal bones in the hands and feet, as well as the joints at the ends of the long bones. Nephropathy, often leading to end stage kidney failure, as well as eye disorders such as corneal clouding, and craniofacial anomalies are all characteristics of MCTO. The disease is often first misdiagnosed as juvenile arthritis or chronic kidney disorder due to its onset features of inflammation or lowered kidney function. But eventually medications do not prove to be effective and the genetic diagnosis is only then discovered. MCTO was initially thought to be caused by excessive activity of osteoclasts, but treatments with anti-resorptive medications like bisphosphonates and denosumab have not been effective in stopping the disease progression. Therefore, data suggests there are likely problems in bone formation, in osteoblasts, chondrocytes and other cells responsible for modeling bone.

On average, between the ages of 4 and 14 many children with MCTO develop locked and dislocated joints at the knees and elbows disabling their ability to walk or rotate or bend their arms. Further, severe contractures of fingers and toes, as well as wrist and ankle deformities create life-altering disabilities. Patients rely on splinting, physical and occupational therapy, and some symptom easing medications such as anti- inflammatories to protect and maintain functionality to any degree possible. There is currently no effective treatment that exists for patients with MCTO. The disease requires more research into the underlying molecular mechanisms and a better understanding of MafB, to determine a therapeutic target, and an approach to a treatment or cure.

For more information, or for research interest please contact:

Lauren Rosenberg
Sophie's Neighborhood Co-Founder
hello@sophiesneighborhood.org
646-275-7881
3215 5th St
Boulder, CO 80304

X-linked hypophosphatemia is a rare bone disorder characterized by low serum phosphorus levels and poor bone mineralization. Symptoms may include lower limb deformities (bow or knock-knee), altered gait, short stature, spontaneous tooth abscesses, bone and muscle pain, arthritis, bone spurs, and tinnitus or hearing loss.

The XLH Network, Inc. began in 1996 as an informal collection of patients and families dealing with X-linked hypophosphatemia. Now a 501(c)3 nonprofit corporation, the organization connects affected families around the world with knowledgeable clinicians, active researchers, and up-to-date information on diagnosis and treatment.

Our mission is to promote XLH awareness and education for affected families, medical professionals, and the community at-large; to support physicians and other providers of medical care for better diagnosis and treatment; to create resources and a community for affected individuals and their families so they can understand and cope with the complications of the disease; and to foster the search for a cure.

For more information, visit our website, www.XLHNetwork.org, or contact us at executivedirector@xlhnetwork.org. Our website provides links to a growing body of information about XLH, along with the latest news. You can find us on Facebook at Facebook.com/XLHNetwork or follow us on Twitter @XLH_Network. For those affected by the condition, including patients and family members, there is a platform for private discussions (forum.xlhnetwork.org).