



RARE BONE DISEASE: CHALLENGES AND SOLUTIONS

Rare bone disorders share five major challenges: (1) **Unknown Prevalence**, (2) **Common Misdiagnosis**, (3) **Lack of Effective Treatments**, (4) **Unmet Research Needs**, and (5) **No Novel Drug Therapies**

The Rare Bone Disease Advocacy Alliance has developed policy solutions to these challenges and will work, as a community, with lawmakers and other stakeholders to make them a reality.

| CHALLENGE | UNKNOWN PREVELANCE |
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| IMPACT | A lack of understanding about the number of individuals impacted by rare bone diseases limits the knowledge base required to support research and advocacy efforts. |
| SOLUTION | Leverage all publicly and privately available resources to support patient registries and data repositories. |

| CHALLENGE | COMMON MISDIAGNOSIS |
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| IMPACT | A lack of awareness of rare bone diseases by primary care providers often causes patients to set off on a “diagnostic odyssey” that results in unnecessary suffering and expense. |
| SOLUTION | Develop and conduct a symposium among government officials, advocacy organizations, researchers, professional medical societies and other stakeholders to determine methods for working collaboratively to improve understanding of rare bone disease within the medical community. |

Fibrous Dysplasia Foundation • HPP Choose Hope • Lymphangiomatosis & Gorham’s Disease Alliance • Lymphatic Malformation Institute
 MHE Research Foundation • Osteogenesis Imperfecta Foundation • Soft Bones, Inc. • XLH Network

1825 Eye Street, NW | Suite 600 | Washington, DC 20006 | Phone: 202.207.3655 | Fax: 202.575-3400

| CHALLENGE | UNMET RESEARCH NEEDS |
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| IMPACT | Insufficient funding for research with regard to natural history studies, animal models and other basic, translational and pre-clinical research means that rare bone diseases are not well understood and few treatment options are available. There is a need to encourage more researchers to enter the field of rare bone research to advance scientific discovery that will lead to treatments and therapies. |
| SOLUTION | Work with the National Institutes of Health (NIH) to develop dynamic methods for fostering interest among young investigators in privately- and publicly-funded rare bone disease research and encourage the Congress to increase funding for rare bone disease and related research. |

| CHALLENGE | LACK OF EFFECTIVE TREATMENTS |
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| IMPACT | Individuals with rare bone disease often face significant disability with few options to treat the pain and other symptoms of their disorder. |
| SOLUTION | Work with professional medical societies, government officials and other stakeholders to develop tools necessary to support patient care protocols, including palliative care and other methods to reduce discomfort, mitigate disability and treat symptoms. |

| CHALLENGE | NO NOVEL DRUG THERAPIES |
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| IMPACT | There are presently no drug therapies specific to rare bone diseases or that treat the underlying cause of the disorders. |
| SOLUTION | Encourage clinical therapy development by fostering closer collaboration between advocacy organizations, academic researchers, the biopharmaceutical industry, the NIH Office of Rare Disease Research and the Food and Drug Administration's Office of Orphan Products Development. |