1 Do people with EDS suffer from dislocations, and if so, how do they manifest?
2 Do you necessarily have to have dislocations for an EDS diagnosis?
3 What other symptoms do EDS sufferers experience? Gastrointestinal, vision, etc.?
4 What are some of the faulty proteins implicated in EDS?
5 What are the various kinds of EDS that have been identified?
6 What are the various genes that have been implicated in EDS? And what genes tangentially affect these genes, potentially affecting disease phenotype?
7 Do transcriptomic tools have a role in EDS research? To what extent can transcriptomic tools help us identify
8 What role do epigenetics play in EDS research? With the epigenomic reads that long-read sequencing tools provide, what could we theoretically do to help EDS sufferers?
9 Since EDS is a full-body disorder, what diagnostic tests could be used to elucidate the areas of the body suffering from EDS pathology, aside from genome sequencing?
10 What proteomics tools are ideal for EDS diagnostic research? Spatial proteomics, visual proteomics, single-cell proteomics? Protein sequencing?
11 Have certain EDS subtypes been definitively linked to certain genes? If so, could you list the subtypes and the genes associated with them?
12 In TNXB-associated EDS, what is the relationship with other genes in the genome that contribute to better or worse phenotypes?
13 What does EDS stand for?
14 Are there different types of EDS?
15 What is connective tissue?
16 How does EDS affect my digestion and gastrointestinal system?
17 How is EDS diagnosed?
18 Are postural orthostatic tachycardia and EDS related?
19 Are people with EDS prone to neck problems?
20 Given the recent findings about how collagen affects the extracellular matrix structure, what implications do these findings have for the development of targeted therapeutic interventions?
21 Explain how tenascins influence cell shape growth and migration
22 Is there significant clinical overlap between myopathic EDS and collagen VI–related myopathies?
23 Explain the effect that damaged extracellular matrix structure due to inadequate collagen affects ECM has on the role of acting as an

epigenetic informational entity through reduced intracellular signals via distinct cell surface receptors.

24 Explain how tenascins in people with EDS are suppressed during wound healing, nerve regeneration, and tissue involution, and in pathological states including vascular disease, tumorigenesis, and metastasis.
25 What are the definitions of EDS and HSD?
26 What are the fundamental aspects and symptoms of EDS and HSD?
27 What are the different types of EDS and their characteristics?
28 What is the genetic basis and what are the causes of EDS and HSD?
29 What are the modes of inheritance and inheritance patterns of various EDS types?
30 What are the considerations for genetic testing and counseling for EDS and HSD?
31 What is the diagnostic criteria for EDS and HSD?
32 What is the diagnostic process and what is the role of various healthcare professionals in diagnosing EDS?
33 What are the differential diagnoses and overlapping conditions with EDS and/or HEDS?
34 What are the most common symptoms of EDS and HSD?
35 How does systemic involvement (e.g., cardiovascular, gastrointestinal, neurological) present in EDS and HEDS?
36 What are the potential complications and comorbid conditions associated with EDS and HSD?
37 What are the management strategies, treatment options, and lifestyle adjustments for patients with EDS and HSD?
38 What are the non-pharmacological approaches to managing EDS and HEDS?
39 What are the pharmacological treatments for specific symptoms of EDS and HEDS?
40 Are there surgical interventions for EDS and HEDS?
41 What is the impact on daily activities and quality of life for people with EDS and hEDS?
42 What strategies can improve functionality and well-being in EDS and HEDS patients?
43 What are reliable sources of information for patients and families dealing with EDS and HEDS?
44 What is the importance of patient advocacy and support groups for EDS and HEDS?
45 What strategies can help cope with the challenges of living with EDS and HSD?
46 Where can I find information on the real experiences of individuals with EDS/HSD and community resources?

47 What ongoing research is being conducted on EDS and HSD?
48 What recent findings and future prospects exist in the study of EDS and HSD?
49 What are the therapeutic targets and potential future treatments for EDS and hEDS?
50 Why are patient registries and collaborative research efforts important in EDS and HEDS?
51 Can you provide hypothetical scenarios to apply knowledge to practical, clinical situations involving patients suspected of or diagnosed with EDS or HSD?
52 What is the long-term outlook for individuals with EDS and HSD?
53 What are the known symptoms of TNXB-related clEDS, Ehlers-Danlos syndrome?
54 What other genes are associated or might be associated with TNXB variants that cause clEDS Ehlers-Danlos syndrome?
55 What is the function of the TNXB gene and what parts of the body does it support?
56 Can you list all known variants that cause TNXB-related clEDS?
57 Can you explain the pathogenic variant of the TNXB gene: NM_001365276.2(TNXB) .7440delinsAC (p.Tyr2480Ter)?
58 Who are the doctors, geneticists, and scientists who specialize in TNXB gene research?
59 What vitamins, supplements, foods, etc., should people with TNXB clEDS avoid?
60 What are the biggest complications of TNXB clEDS?
61 Do TNXB clEDS patients have normal life spans?
62 How is TNXB-related EDS detected?
63 What type of genetic testing is best for finding pathogenic variants of the TNXB gene?
64 What is the sensitivity and specificity of genetic testing for the TNXB gene?
1 What is the most commonly documented ALPL mutation?
2 What is the most commonly documented ALPL mutation resulting in phenotypic Hypophosphatasia?
3 How many unique ALPL mutations have been documented from 1950 to current day?
4 What effect does Hypophosphatasia have on on the lungs?
5 How is Hypophosphatasia inherited?
6 What role does TNSALP have in the body?
7 Does Hypophosphatasia cause pseudofractures?
8 Do baby teeth fall out with roots intact?

9 What are the 2 main blood test you should have done to test for HPP?
10 What is the mortality rate in infants with hypophosphatasia?
11 Severe forms of hypophosphatasia affect an estimated how many people?
12 How does hypophosphatasia affect the brain?
13 What does Alkaline Phosphatase do to pyridoxal 5-phosphate so it can cross the blood brain barrier?
14 Are patient with Hypophosphatasia b6 toxic or deficient in the CFS and the cells of the body?
15 Which treatments should be avoided in patients with HPP?
16 Are patient with Hypophosphatasia b6 toxic or deficient in the CFS and the cells of the body?
17 Which is most accurate about emerging therapies for the management of HPP?
18 At what location are the DNA mutations found for hypophosphatasia?
19 What is the relationship between hypophosphatasia (HPP) and Ehlers Danlos Syndrome (EDS)?
1 What are the underlying genetic mutations responsible for different forms of hypophosphatasia, and how do they affect enzyme function?
2 How does the ALPL gene mutation lead to the clinical manifestations of hypophosphatasia?
3 What are the biochemical pathways disrupted by hypophosphatasia, and how do these disruptions lead to skeletal abnormalities?
4 How do different mutations in the ALPL gene correlate with the severity and onset of hypophosphatasia symptoms?
5 What are the diagnostic criteria for distinguishing between different subtypes of hypophosphatasia?
6 How does the clinical presentation of hypophosphatasia vary between perinatal, infantile, childhood, and adult forms?
7 What are the key differences in the pathophysiology of hypophosphatasia between adults and children?
8 How does hypophosphatasia affect the development and mineralization of teeth, and what are the dental implications?
9 What are the current treatment options for hypophosphatasia, and how effective are they in managing the disease?
10 How does enzyme replacement therapy work in treating

hypophosphatasia, and what are its limitations and potential side effects?

11 What are the potential long-term outcomes and complications of untreated hypophosphatasia in both pediatric and adult patients?

12 How can prenatal diagnosis of hypophosphatasia be performed, and what are the implications for pregnancy management?

13 What are the roles of alkaline phosphatase and pyrophosphate in bone metabolism, and how are they altered in hypophosphatasia?

14 How does hypophosphatasia impact overall metabolic processes, beyond its effects on the skeletal system?

15 What are the potential novel therapeutic targets for hypophosphatasia based on current molecular and genetic research?

16 How do lifestyle and environmental factors influence the severity and progression of hypophosphatasia?

17 What are the psychosocial impacts of hypophosphatasia on patients and their families, and how can they be addressed?

18 How can multidisciplinary care improve the management of hypophosphatasia, and what specialists should be involved?

19 What are the current challenges in developing effective gene therapy for hypophosphatasia?

20 How do animal models contribute to our understanding of hypophosphatasia, and what are the limitations of these models in translating findings to human patients?