The Top 25 questions (in order of priority as agreed at the final workshop). For information about other questions received by the PSP, please see further down this spreadsheet	H = health or social care professional; O = organisations representing people with MBD; P= person with MBD; R= relative/carer/friend of person with MBD; T = Other.			
Qn. No.	**Original questions**	**Category of Respondent**	**Type of rare metabolic bone disorder**	**Previous related research studies**
1. What is considered a good outcome of treatment in rare bone metabolic disorders? How can this be measured in studies of new treatments?	For FD:Correlation between markers and QOL?	H		Determinants of impaired quality of life in patients with fibrous dysplasia.Majoor BCJ, Andela CD, Bruggemann J, van de Sande MAJ, Kaptein AA, Hamdy NAT, Dijkstra PDS, Appelman-Dijkstra NM.Orphanet J Rare Dis. 2017 Apr 27;12(1):80.
	Biomarkers for efficacy	H		Physical function is impaired but quality of life preserved in patients with fibrous dysplasia of bone.Kelly MH, Brillante B, Kushner H, Gehron Robey P, Collins MT. Bone. 2005 Sep;37(3):388-94
	For FD: Better characterisation of outcomes - what is correlation between marker changes and radiological progression/improvement?	H		Health-related quality of life and a cost-utility simulation of adults in the UK with osteogenesis imperfecta, X-linked hypophosphatemia and fibrous dysplasia. Forestier-Zhang L, Watts L, Turner A, Teare H, Kaye J, Barrett J, Cooper C, Eastell R, Wordsworth P, Javaid MK, Pinedo-Villanueva R
	X: anti-FGF23 treatments in XLH-how will outcomes in trials be measured meaningfully?	H		https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3235275/
	In each of these areas there are is little by way of high quality outcome data as to clinically relevant end points. Where data is available it tends to be predicated on laboratory or imaging data rather than clinically relevant data	H		http://www.thebonejournal.com/article/S8756-3282(08)00897-1/fulltext
	Little or no evidence on quality of life, cost effectiveness or similar	H		https://www.ncbi.nlm.nih.gov/pubmed/12794240
2. What is the cause of pain in people with rare metabolic bone disorders?	FD:What is the source of bone pain?	H		Pain in fibrous dysplasia of bone: age-related changes and the anatomical distribution of skeletal lesions.Kelly MH, Brillante B, Collins MT. Osteoporos Int. 2008 Jan;19(1):57-63.
	My main questions would be pain There is little known about fd and pain is bad with it Why is the pain with fd	P	F	
	Why am I constantly in pain?	P	O	
	How do hormones impact on PFD lesions? There is a definite connection between pain levels and menstrual cycle, what is this due to?	P	F	
	Is there research to suggest there is any link to decreased strength and ability / increased pain and problems around the time of an OI women's periods?	P	O	
3. What is the psychological impact of having a rare metabolic bone disorder and how can patients and their families best be supported?	What is the psychosocial impact of fibrous dysplasia ?	H		
	Support during pregnancy and for mums with children with oi	P	O	
	Does having OI and lots of injuries affect the brain etc do we become socially creatively different. . I read somewhere that traumatic incidents in early life have a long term affect. There's not a lot about the pychological impact of breaking 145 bones glfor instance.. if you scanned our brains would we be different ?	P	O	
	psycological help	P	X	
	help to cope with life with FD.	P	F	
	Pain relief and PTSD reduction in acute settings (i.e. preventative medazolam)	P	O	
	Better support for families	P	O	
	What are ways to mentally stay strong when you have lived with XLH your whole life?	P	X	
	Is it linked/caused my depression?	P	F	
	Since having my wrist fused I have developed complex regional pain syndrome which has drastically made my disability worse. I have had very little treatment for this which has been very disappointing and has affected my mental health.Depression must be a common occurrence with people with lifelong disability and much more support is needed for this.Mental health services don't seem to know how to help people that predominantly have a physical challenge and in my experience my depression has not been taken seriously.	P	O	
	More support for something to do in life. I'm fed up being written off.	P	O	
	Long term effects on life - mental health	R	O	
4. What can be done to prevent rare metabolic bone disorders in the first place, or to stop them from getting worse?	Help to prevent age related deterioration	P	O	
	Again can treatment prevent reduce lack of mobility injury etc	P	O	
	Is there anything that can be done to prevent the calcification of ligaments etc	P	X	
	Other than surgery can anything be done to slow down progressive kyphosis/scoliosis?	P	O	
	Is there anything that can be done to prevent the disease?	R	O	
	If XLH patients will suffer more than general with weaker bones as they age, are there any medications or treatments which can be taken to slow down the weakening process?	R	X	
5.What are the best ways to manage fatigue linked to rare metabolic bone disorders?	Fatigue is very common in OI of all ages, why is this? and how can this be improved?	P	O	
	Fatigue- what can be done for this?	P	O	
6. What are the best forms of surgery to treat bones and joints in people with rare metabolic bone disorders?	OI: What are the optimal surgical techniques for the management of fractures in osteogenesis imperfecta?	H		A small case series suggests that hip and knee arthroplasties are effective in treating arthritis associated with XLH, but osteotomy may also be required if there is severe deformity (Larson AN et al, J Arthroplasty 2010; 25: 1099-1103). Other small case series report correction of lower limb deformities with osteotomy, external fixation and insertion of an intramedullary nail (Kocaoglu M et al, J Bone Joint Surg Br 2011; 93: 52-56, Song HR et al, Acta Orthop 2006; 77: 307-314) . Correction of deformity has also incuded leg lengthening using distraction osteogenesis by Ilizarov or Heidelberg external fixation (Matsubara H et al, Arch Orthop Trauma Surg 2008; 128: 1137-1143, Choi IH et al, J Pediatr Orthop 2002; 22: 626-631).
	All: What is the optimal way of managing skeletal deformity in all three conditions?	H		Functional and radiological outcomes of a minimally invasive surgical approach to monostotic fibrous dysplasia. Rosario MS1,2, Hayashi K3, Yamamoto N1, Takeuchi A1, Miwa S1, Taniguchi Y1, Tsuchiya H1. World J Surg Oncol. 2017 Jan 5;15(1):1. (n=12)
	When is the best time to do surgery?	H		Surgical treatment for fibrous dysplasia of femoral neck with mild but prolonged symptoms: a case series. Nishida Y1, Tsukushi S2, Hosono K3, Nakashima H4,5, Yamada Y6, Urakawa H7, Ishiguro N8. J Orthop Surg Res. 2015 May 10;10:63. (n=8)
	Surgical interventions: For whome? When? Which techniques? Pharmacological pre-treatment?	H		Valgus osteotomy in combination with dynamic hip screw fixation for fibrous dysplasia with shepherd's crook deformity. Li W1, Huang X, Ye Z, Yang D, Tao H, Lin N, Yang Z. Arch Orthop Trauma Surg. 2013 Feb;133(2):147-52. (n=21)
	What can be done to help lax joints	P	O	Surgical treatment of dysplasia fibrosa and defectus fibrosus with bone allografts. Tomasik P1, Spindel J, Miszczyk L, Chrobok A, Koczy B, Widuchowski J, Mrozek T, Matysiakiewicz J, Pilecki B. Ortop Traumatol Rehabil. 2010 Jan-Feb;12(1):58-66.
	Why does my femurs get thinner because of rods?	P	O	Valgus osteotomy combined with intramedullary nail for Shepherd's crook deformity in fibrous dysplasia: 14 femurs with a minimum of 4 years follow-up. Yang L1, Jing Y, Hong D, Chong-Qi T. Arch Orthop Trauma Surg. 2010 Apr;130(4):497-502..
	Is multiple surgical intervention a good idea past a certain age in terms of bone regeneration and healing?	P	X	Fibrous dysplasia of bone: management and outcome of 20 cases. Keijser LC1, Van Tienen TG, Schreuder HW, Lemmens JA, Pruszczynski M, Veth RP. J Surg Oncol. 2001 Mar;76(3):157-66; discussion 167-8. (n=20)
	What is the long term management of our joints? I have so many joints that are bad I couldn't have them all replaced.	P	X	Fibrous dysplasia of the proximal part of the femur. Long-term results of curettage and bone-grafting and mechanical realignment. Guille JT1, Kumar SJ, MacEwen GD. J Bone Joint Surg Am. 1998 May;80(5):648-58.
	Is it now an option for OI type III children/adults to have spinal surgery?	P	O	What Is the Role of Allogeneic Cortical Strut Grafts in the Treatment of Fibrous Dysplasia of the Proximal Femur? Majoor BC1, Peeters-Boef MJ2, van de Sande MA2, Appelman-Dijkstra NM3, Hamdy NA3, Dijkstra PD2. Clin Orthop Relat Res. 2017 Mar;475(3):786-795. (n=30)
	Do knee/hip replacements help in the short or long term?	P	X	
	Due to joint hypermobility joint operations haven't been very successful so other ways of maintaining preserving joints needs to be found/ implemented.	P	O	
	Will my bones sustain knee or hip replacements.	P	O	
	Why are there still many surgeons who differ greatly in the way they carry out operations and treatments following fractures?	P	O	
	Surgery - should one have bone grafting in e.g. Tibia ? If so - what sort of graft? If a nail is put into , say, tibia ,should it be taken out at some stage?	P	F	
	If a long bone has already had telescopic rods or pins in it for over 10 years, is it as effective and safe to re-do the same operation if the metal work moves and becomes ineffective?	P	O	
	Why are bone grafts not recommended by some doctor's?	P	F	
	Are there any effective surgical options to help prevent worsening of scoliosis in adults with OI?	P	O	
	Operation outcomes for people with OI (as opposed to information applying to "normal" patients)	P	O	
	What should I ask before a surgery? I broke my arm (radius) and was told the surgeon would use steel to fix it. I had no idea whether that was a good option or not.	P	O	

	Surgeries for leg straightening	P	X	
	Will it be a good idea to see an orthopedic surgeon to follow up on the surgeries I had many years ago?	P	X	
	Can diseased bones be completely replaced from the bone bank?	R	F	
	Most common operation / procedures (successes / failure rates).	R	X	
	What benefits are there to having more surgery if fibrous dysplasia is active?	R	F	
7. What are the benefits and side effects of drug treatment for people with rare metabolic bone disorders in the short and long term? What is the optimal length of treatment?	FD: Do bisphosphonates have any benefits other than the relief of pain?	H		XLH has generally been treated with an activated vitamin D metabolite (calcitriol or alfacalciol) and phosphate supplements, but their effectiveness is limited. A recent randomised controlled trial demonstrates that use of the anti-FGF23 monoclonal antibody (KRN23; Burosumab) reverses biochemical abnormalities in XLH, increasing serum phosphate and 1,25(OH)2D. concentrations (Imel EA et al,J Clin Endocrinol Metab 2015; 100: 2565-2573). Phase III clinical trials of Burosumab are underway and NICE are conducting a Highly Specialised Technologies Evaluation of Burosumab, which is due to be published in October 2018.
	FD: Do other treatments (anti-IL6, denosumab) have any role to play in treating fibrous dysplasia?	H		A randomized, double blind, placebo-controlled trial of alendronate treatment for fibrous dysplasia of bone. Boyce AM1, Kelly MH, Brillante BA, Kushner H, Wientroub S, Riminucci M, Bianco P, Robey PG, Collins MT. J Clin Endocrinol Metab. 2014 Nov;99(11):4133-40. (n=24 adults)
	FD:What about denosumab?	H		Outcome of Long-Term Bisphosphonate Therapy in McCune-Albright Syndrome and Polyostotic Fibrous Dysplasia. Majoor BC1,2, Appelman-Dijkstra NM1,3, Fiocco M4,5, van de Sande MA1,2, Dijkstra PS1,2, Hamdy NA1,3. J Bone Miner Res. 2017 Feb;32(2):264-276. (n=41)
	FD: Bisphosphonates and denosumab	H		Pegvisomant for the treatment of gsp-mediated growth hormone excess in patients with McCune-Albright syndrome. Akintoye SO1, Kelly MH, Brillante B, Cherman N, Turner S, Butman JA, Robey PG, Collins MT. J Clin Endocrinol Metab. 2006 Aug;91(8):2960-6. (n=5)
	Role of bisphosphonates, particularly which one, in McCune Albright syndrome.	H		Bisphosphonate treatment of bone fibrous dysplasia in McCune-Albright syndrome. Lala R1, Matarazzo P, Andreo M, Marzari D, Bellone J, Corrias A, de Sanctis C; Study Group for Gs alpha Protein Related Diseases of the Italian Society for Pediatric Endocrinology and Diabetes. Pediatr Endocrinol Metab. 2006 May;19 Suppl 2:583-93. (n=14 5-18.7yr)
	Are bisphosphonates safe ?	P	F	Pamidronate treatment in bone fibrous dysplasia in children and adolescents with McCune-Albright syndrome. Matarazzo P1, Lala R, Masi G, Andreo M, Altare F, de Sanctis C. J Pediatr Endocrinol Metab. 2002;15 Suppl 3:929-37.
	The effectiveness of bisphosphonates treatment.	P	F	Long-term effects of intravenous pamidronate in fibrous dysplasia of bone. Chapurlat RD1, Delmas PD, Liens D, Meunier PJ. J Bone Miner Res. 1997 Oct;12(10):1746-52. (n=20)
	any treatments found to prevent new growth new treatments being used to prevent growth or re grow bone.	P	F	https://academic.oup.com/jcem/article/98/3/871/2536494
	It would be absolutely required to find a system helping to monitor and balance PTH vs P supplementation.	P	X	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3488185/
	Does human growth hormone help OI patients?	P	O	
	Is treatment with bisphosphonates in adults with FD and OI effective? Is it safe in the long-term?	H		
	How aggressively do we need to treat to change long-term outcomes?	H		
	Use of therapies long-term	H		
	For FD:Does denosumab differ from BPs? And why? Impact for long-term therapy?	H		
	optimal duration of treatment	H		
	FD&OI: How long should we treat FD with bisphosphonates? Is there an increase in the risk of osteonecrosis of the jaw or atypical femur fractures?	H		
	X:How long should we treat XLH with calcitriol in order to minimise harm?	H		
	OI: How long should we treat for?	H		
	FD: Which treatment, how often?	H		
	OI: Any long term benefits of bisphosphonates	H		
	Optimal use of bisphosphonates in Fibrous Dysplasia, particularly in younger patients.- risks of atypical fracture etc and alternatives such as denosumab (and problems with stopping this)	H		
	Optimal dosing strategy for phosphate replacement and avoidance of tertiary hyperparathyroidism	H		
	Do bisphosphonates work in OI How long should you treat for	H		
	Does bisphoshonate treatment prevent longer term complications	H		
	What happens to adults with OI who have had bisphosphonate treatment as a child. Are there any longer term impact	H		
	Does treatment for OI as adult with bisphosphonates have any impact on longer term outcomes	H		
	Have there been changes seen with those who took medical trials as children?	P	O	
	I have Oi but I am so wary about the things I do so that I can minimise the pain factor (fracture in spine!). I was on the meds but taken off them because 10 years was up. Told that after 10 years meds can cause facturers and work against you. So what happens now? At the minute I live on solaphine and get on with it!	P	O	
	Are there side effects re the drugs we take over the long term	P	O	
	what long term negative affects come from taking Tramadol daily?	P	F	
	What are the long term impacts of bisphosphonates?	P	O	
	Should patients continue with phosphate/vit D medication throughout life or does this cause other problems ie do the benefits outweigh the possible problems caused?	P	X	
	What proof is there that taking a bisphosphonate impacts on the teeth and the jaw. If I need impants or dental treatment in the future (either NHS or Private), what evidence can I show to prove there is no additional risk (or is there?)	P	O	
	What evidence is there so far into the long term benefit of bisphosphonates given in childhood?	P	O	
	I am concerned about the long-term effectiveness of the treatment which I currently receive (Bisphosphanates) Will the treatment continue to help, what are the long-term impacts on my general health	P	F	
	Side effects of childhood treatments	P	X	
	More readily available information about effects/ benefits of long term medication	P	X	
	Would lifelong physiotherapy/hydrotherapy be beneficial?	P	X	
	What happens with biphosphonates at long term?	P	O	
	What are the long term effects to experimental medications (eg Fosamax) taken during trials as a child, there is no follow up of monitoring afterwards by medical professionals.	P	O	
	effects of the drugs in the long term	P	X	
	Consequences of the drugs	P	X	
	What are the long term effects of taking Phosphate and Alpha on my body?	P	X	
	Does taking phosphate and alpha encourage additional bone to form in unwanted parts of by body?	P	X	
	What are the short- and long-term side effects of bisphsphonates? How effective are they?	P	O	
	How long will treatment need to continue once osteoporosis is diagnosed?	P	O	
	What are the side effects of long term use of injections and transfusions for the treatment of osteoporosis?	P	O	
	How long can I keep taking Zol?	P	F	
	Are there any side affects from years of taking medicines?	R	F	
	Issues into long term usage of Phosphate on body?	R	X	
	What are the long term effects of treatment? My niece is 10 and receives biphosponate treatment. What are the long term effects if she continues? Can it ultimately cause more harm than good? Is it really helping?	R	O	
	Various combinations of medication. Different ways to take medication and impact of taking them together etc?	R	X	
	Longterm side effects of medication	R	X	
	From an on going perspective, what are the known side effects if you stop treatment once 'growth' adolescent years are over? What is the prognosis for those who cease treatment at this time?	R	X	
	Long term effects of bisphosphonates	R	O	
	What is the long term effect on taking the current drugs for xlh and ADHR and do they help prevent osteomalacia, fractures/ breaks later in life?	T	X	
8. How do rare metabolic bone disorders progress as people grow older and how is this different from normal ageing?	What is the natural history of these conditions?	H		Monostotic fibrous dysplasia of the proximal femur: natural history and predisposing factors for disease progression. Han I1, Choi ES, Kim HS. Bone Joint J. 2014 May;96-B(5):673-6
	What is the natural history of Craniofacial FD lesions?	H		
	FD: What are the predictors of change in condition (i.e. symptoms or bone expansion)?	H		
	How can one tell if pain is caused by XLH or simple ageing?	P	X	
	At what age do the dental problems usually abate? Is there much research on dental problems continuing in adulthood?	P	X	
	What does this mean for the rest of my life? Does it mean I wont stop breaking.	P	O	
	Getting an overview of the long term prognosis for adults with Osteogenesis Imperfecta?	P	O	
	Why isn't pain considered a real side effect of xlh among adults?	P	X	
	Why is the impact on adult teeth not considered a risk by the profession?	P	X	
	How will my condition progress as I get older?	P	O	
	I am a woman of 57 years of age what might happen in a few years ? will my bones lose more density?	P	O	
	I think XLHers should be given advice on what to expect in their future	P	X	
	Any age-related deterioration in people with OI,	P	O	

	FD lesions in vertebrae thoracic and lumbar, what is likely to happen if these continue growing?	P	F	
	I would like there to be more insight / knowledge of how XLH in the long term effects your bones and muscles at older age can specific bone-/ muscle-effects in adults be identified as a result of XLH that are not a result of general old-age-issues such as artrose?	P	X	
	Impact on the inner ear is not well known and seems to lead to major impairment at various life stages according to the individual patient.	P	X	
	Is it possible for the body to regenerate itself and the bone structure? In other words is it possible for at patient to get better?	P	X	
	What might happen regarding OI as I get older?	P	X	
	Is there a key age where more problems occur?	P	X	
	How will this affect my life. How bad will the pain get	P	X	
	What is the long term prognosis	P	X	
	How will I feel when I'm older? Am I going to feel much worse?	P	X	
	What is the long term effect of XLH on the usual aging process?	P	X	
	Have you heard of FD becoming active again in late years?	P	F	
	Can Fibrous displasia continue to grow as adult?	P	F	
	what might happen	P	X	
	Can it [diagnosis] change after having it for years?	P	F	
	How does the disease progress as the patient gets older?	P	F	
	As i age can i expect my bone density to reduce and if so when should i expect this to happen and what can i do to slow down the reduction	P	O	
	The emphasis here is on studies with children. But what about us middle aged people? What's around the corner? No doctors can tell me what to expect.	P	O	
	But basically my joints seem to be affected, more and more. As well as OI, I've been told I also have osteoporosis. What else will happen / what should I expect. I have bad Tinitus and have been told it could be because of my health issues. What's next?	P	O	
	Aging effect on bones	P	O	
	I think as I get older, the pain will get worse	P	O	
	As I reached adulthood my breaks have been a lot less frequent and possibly no longer related to OI. Is there no way persons like myself who has been lucky enough to (grow out) of OI could be monitored or tested to see what makes our bones different to those who continue to suffer	P	O	
	Am I likely to go deaf in the future?	P	O	
	Will I be in this much pain for the rest of my life? Will it get worse?	P	O	
	Will my condition get worse.	P	F	
	Will my condition worsen with age	P	F	
	Long term prognosis	P	X	
	Future issues to watch out for	P	X	
	What does the future hold for me	P	O	
	Yes more info on long term affects on bones, low phosphate levels and benefits of different surgeries,	P	X	
	How can I determine which ailments are related to the disease and generally how is it all connected to my health?	P	X	
	I want to know what my mobility will be like when I'm older? As I worry about my flexibility and stiffness.	P	X	
	I have often been told that my symptoms are not connected to XLH but just another odd thing you have or when saying I have a physical problem that it is normal for you and nothing can be done or when reporting I cannot do a sport anymore because of swelling or pain that I have done well to play to the standard I have or for as long as I have. Eventually I feel I will have to stop sport altogether - what then? It is such a large part of my life, what can I do to play for as long as possible. I fear I have now gone past this point.	P	X	
	What are the degenerative effects of OI as a patient gets older? All information I can gather are for osteoporosis or assumed effects that should happen to adults with OI. What are the real life effects being seen in the adult population?	P	O	
	How does one differentiate between pain of getting older and pain of XLH?	P	X	
	What is the long term impact on my physical wellbeing?	P	X	
	What are the long term effects? Will I end up in a wheel chair? Need regular surgery like hip replacement?	P	X	
	I'm 36 and I worry about what future holds as far as my career, mobility etc	P	X	
	Will this worsen? (Pain and/Or will my lesions grow?)	P	F	
	What will happen as I grow older?	P	F	
	Can this disease impare me more.	P	F	
	What can I expect as I age?	P	X	
	It impacts my life and future yet even the specialist says little is currently know about its behaviour and progression	P	F	
	What is known about the impact of XLH and the ageing process? What is likely to go wrong and when is it likely to happen?	R	X	
	Generally, as one ages, bones seem to get weaker and more prone to fracture. Is there any evidence that this is worse for people with XLH?	R	X	
	and what to expect as I age. Aging with OI	R	O	
	what is the prognosis.	T	X	
9. How are other parts of the body affected by rare metabolic bone disorders to cause other symptoms?	Are hearing loss and migraines associated a xlh?	P	X	A study compared muscle size, density and fuction in 34 children and adults with XLH and 34 age and sex matched control subjects. The authors reported no difference in muscle size, but lower muscle density and muscle force and power in XLH (Veilleux LN et al, J Clin Endocrinol Metab 2012; 97: 1492-1498).
	Will all my tendons be affected by OI.	P	O	Patients with McCune-Albright syndrome have a broad spectrum of abnormalities in the gastrointestinal tract and pancreas.Wood LD, Noë M, Hackeng W, Brosens LA, Bhajee F, Debeljak M, Yu J, Suenaga M, Singhi AD, Zaheer A, Boyce A, Robinson C, Eshleman JR, Goggins MG, Hruban RH, Collins MT, Lennon AM, Montgomery EA. Virchows Arch. 2017 Apr;470(4):391-400.
	I think OI is affecting some of my organs is there any evidence? Effect on other parts of body e.g. kidneys. Liver etc	P	O	
	I seem to have a few extra things to deal with and am often told it is "nothing to do with XLH but just another odd thing you have" this seems strange to me as I cannot believe some things are not connected.	P	X	
	Is there a relation between XLH and ears (such as hearing loss at a young age, frequent ear infections (even in adulthood), congenital /inborn deafness, constant ear-pain, Meniere's disease) ? What is the relation between XHL and ear-problems ? How can those problems be explained from XLH?	P	X	
	Why is it many that go wrong with me seems to be unexplained/undiagnosed in spite of the usual tests being carried out? eg severe night sweats for many years, severe pain in ankles, sudden onset of migraines, dizziness, tinnitus. It always seems to be written off as XLH related. Is there an actual database of likely symptoms?	P	X	
	And, what else may go wrong or deteriorate?	P	X	
	Does throats also get larger and neck wider?	P	F	
	Why do I have kidney issues? And so many more health issues...	P	O	
	Is there any link to heart conditions and stomach perforations to oi	P	O	
	How is the heart affected and at what age is screening advisable?	P	O	
	What is causing my fatigue?	P	O	
	Why do I get inflammations (muscles) easier than others?	P	O	
	Are people with OI more likely to have balance problems and therefore fall more often? Perhaps because of calcium that is blocking the balance bit on the inside of the ear?	P	O	
	When I see a physio they give me exercises to help strengthen my muscles but they don't really understand how my condition affects my muscles/joints and even the smallest amount of exercise can bring on pain. Is it possible that this condition also affects ligaments/tendons?	P	O	
	Does XLH affect memory, difficulty finding words and speaking, cause headaches?	P	X	
	I thought this was a bone disease not muscular disease. I have recently learned it's both. Is it true our muscles can be very painful with XLH?	P	X	
	Does XLH affect the nervous system and how?	P	X	
	Are hearing, and balance issues related to XLH?	P	X	
	What other symptoms are linked to the condition?	P	X	
	heart problems	P	O	
	Is high blood pressure linked to XLH?	P	X	
	Is there specifics to be concerned about other then spine, lungs, and heart?	P	O	
	What frequency do people with XLH have hearing issues?	P	X	
	Could this be the reason I get exhausted from time to time?	P	F	
	Do many people with Fibrous Dysplasia have problems with their liver as well ?	P	F	
	ide effects of the disease in the skull. And tumor growth.	P	F	
	What is the connection between OI and problems such as stroke, aneurysm, etc.?	P	O	
	Condition not just about broken bones- feel all the other symptoms are overlooked.	P	O	
	Cardiovascular health (rapid heart beat, irregular heart beat).	P	O	
	Why is hearing of XLH patients affected? Is this linked to phosphates or calcification?	R	X	
10=. What are the best ways to prevent dental problems in people with rare metabolic bone disorders?	Is there any prophylactic treatment other than good oral hygiene to prevent dental infections?	P	X	A prospective study reported that dental abnormalities were common in XLH, unrelated to delayed medical treatment. The authors recommended regular oral examination (Souza MA et al, Clinics (Sao Paulo) 2010, 65: 1023-1026). Small case series suggest that duration of tretment with activated vitamin D supplements may be associated with a beneficial effect on dental health (Chaussain-Miller C et al, J Pediatr 2003; 142: 324-331, Connor J et al, J Clin Endocrinol Metab 2015; 100: 3625 3632).
	Looking after your bones? Looking after your teeth due to bone loss in the jaw	P	O	https://onlinelibrary.wiley.com/doi/full/10.1111/j.1754-4505.2008.00070.x

	Is there any preventive dental treatment advice?	P	X	
	Are there any procedures that a dentist might suggest to a non XLH person that would not be appropriate to the an XLH patient? eg deep cleaning, whitening?	P	X	
	What can be done to elite dental problems eg broken teeth.	P	O	
10=. How and why do people with rare metabolic bone disorders have different symptoms, even when they have the same genetic mutation?	What impacts upon the disease spectrum in terms of symptoms? Why are some patients significantly more symptomatic than others with similar levels of bone disease?	H		
	Why do I have OI and my identical twin doesn't?	P	O	
	Why does it sometimes take several years to diagnose e.g. why doesn't it "show up" sometimes until you're 12-18 mths of age?	P	X	
	I have never had someone tell me what strength my oi is	P	O	
	Are there actually different grades of xlh? Symptoms seem to vary widely.	P	X	
	Why do some people stop having fractures after puberty and others go on to continually fracture?	P	O	
	Why did I have no symtoms until I was in my 50s	P	F	
	Why are XLH patients so different from one another, even within the same families?	P	X	
	I have a mutation that has never been found according to the university of washington collagen database. How does that impact me in terms of the progression of OI vs other common mutations?	P	O	
12. Are there more effective, long-term treatments for pain (including non-opioid drugs and non-drug treatments)?	Gradation of effect on FD pain by comparing bishops regimes	H		A randomized, double blind, placebo-controlled trial of alendronate treatment for fibrous dysplasia of bone. Boyce AM1, Kelly MH, Brillante BA, Kushner H, Wientroub S, Riminucci M, Bianco P, Robey PG, Collins MT. J Clin Endocrinol Metab. 2014 Nov;99(11):4133-40. (n=24 adults)
	FD: What is the optimal way of treating pain in fibrous dysplasia?	H		Outcome of Long-Term Bisphosphonate Therapy in McCune-Albright Syndrome and Polyostotic Fibrous Dysplasia. Majoor BC1,2, Appelman-Dijkstra NM1,3, Fiocco M4,5, van de Sande MA1,2, Dijkstra PS1,2, Hamdy NA1,3. J Bone Miner Res. 2017 Feb;32(2):264-276. (n=41)
	How do you treat the pain associated with fibrous dysplasia	H		Total hip arthroplasty in patients with underlying fibrous dysplasia. Sierra RJ1, Cabanela ME. Orthopedics. 2009 May;32(5):320. (n=11)
	Pain relief besides those listed on the OI Website. Tried them all, nothing has helped!	P	O	Long-term effects of intravenous pamidronate in fibrous dysplasia of bone. Chapurlat RD1, Delmas PD, Liens D, Meunier PJ. J Bone Miner Res. 1997 Oct;12(10):1746-52. (n=20)
	I would like to know more about what alternative there are to taking pain drug on a regular basis. Cognitive behavioral therapy	P	O	Bisphosphonate treatment of bone fibrous dysplasia in McCune-Albright syndrome. Lala R1, Matarazzo P, Andreo M, Marzari D, Bellone J, Corrias A, de Sanctis C; Study Group for Gs alpha Protein Related Diseases of the Italian Society for Pediatric Endocrinology and Diabetes. Pediatr Endocrinol Metab. 2006 May;19 Suppl 2:583-93. (n=14 5-18.7yr)
	What are some appropriate pain relief options which don't involve opiods?	P	X	Pamidronate treatment in bone fibrous dysplasia in children and adolescents with McCune-Albright syndrome. Matarazzo P1, Lala R, Masi G, Andreo M, Altare F, de Sanctis C. J Pediatr Endocrinol Metab. 2002;15 Suppl 3:929-37.
	I have OI and over the past few years my pain in my back and hips has increased. I don't like taking pain Killers, but that's all my G.P tells me to take. I would love to try alternative medicines.	P	O	
	pain relief	P	O	
	What are the best drugs/pain relief to help with severe aches and pains - often described as stress/micro fractures?	P	O	
	I have constant pain in lower back and legs on pain relief but just about takes edge off it	P	O	
	Pain Management effect of pain medication on stomach	P	O	
	Is there anything better than Alendronic acid and pain relief I take tramadol and paracetamol but constantly in bone pain.	P	O	
	Pain relief. My pain is not managed. Alternative meds are not advised on and expensive if i go private	P	O	
	I have seen OT and Physio and Endocrine and while really nice and try to help they don't know how to and cant help with my pain. Doctor will only give pain relief while fractured	P	O	
	Defo pain relief. Getting no where with anyone on that Also on pamidronate or similar after adult hood..No one knows and endocrine even admit here they don't know	P	O	
	What is available as a long term pain relief	P	O	
	I have sever neck and spinal pain which is getting worse, none of my Doctors can offer any meaningful remedy for this sad side effect of OI. Can your work help me?	P	O and RA	
	Pain relief	P	X	
	Does acupuncture help with pain relief in people with OI meaning that strong drugs, which have a sedative effect, are no longer required?	P	O	
	or some kind of pain relief that can actually negate the pain caused by it.	P	O	
	pain relief	P	X	
	When will there be an available treatment for bone pain?	P	X	
	What kind of things can I do to prevent the constant aching bone pain? Is there any way to get relief from pain?	P	X	
	what can be done about pain	P	O	
	Given doctors are clamping down on prescribing opioids; are there any other prospects for pain relief on the horizon? Or are we really going to end up where we're headed for; people with osteogenesis getting nothing stronger than paracetamol for regular fractures and dislocations, never mind the routine bone and joint pain?	P	O	
	Would like more information on what pain relief works ?	P	F	
	As an adult, I'm having trouble with on going leg pain. My doctor thinks it's due to low phosphorus. I'm taking KPHOS but the pain is still there in my lower legs mostly. I try to walk some every day. Phosphorus is very expensive, even with insurance, so I can't take alot and I know it doesn't stay in the body long. How or when will something else be available to help with this? I m almost 65 now. My mobility is getting limited. I can walk. No running. I use cane sometimes.	P	X	
	are there any new pain controls drugs shown to help OI	P	O	
	Effective pain medication like CBD.	P	F	
	What medications are there for pain relief that are as effective as opiates but without the adverse side effects.	P	F	
	How do you treat pain in the long term as it varies from from painful to severe on a near daily basis	P	F	
	Is there any support for those who have pain in spine, hips, knees other than medication.	P	F	
	What pain medications are most effective in treating long term bone pain in OI?	P	O	
	What are the best pain relief options for people with OI?	P	O	
	Pain relief options for the long-term	P	O	
	Long-term pain management	P	O	
	Appropriate pain relief (inc alt. Meds).	P	O	
	What is the best pain medication?	P	O	
	What is the best strategies for different kinds of pain?	P	O	
	Has there been any research into medical cannabis/CBD drugs that are legal in the UK? Does it help joint and bone pain in people with OI?	P	O	
	What is best pain relief for someone with OI, Osteoporosis and Osteoarthritis?	P	O	
	What alternatives to analgesics are viable for management of the day to day pain associated with OI?	P	O	
	Pain relief	P	X	
	I am trying to keep working but the pain is now getting unbearable.	P	X	
	What is the best pain management without using narcotics?	P	X	
	What works for pain relief?	P	X	
	What is the best long term management for pain especially in today's environment where doctors are being very discouraged in prescribing narcotic pain medications?	P	X	
	I would like to get infusions but am being told to stay on tablets. I take pain medication every day and have been refused a consult with pain management	P	O	
	Is cannabis (CBD, specifically) a form of pain management?	P	X	
	Is there any pain relief to help with oi I have oi and eds alongside malignant hyperthermia and struggling big time	P	O	
	What alternative pain remedies exist for those not wanting to be labeled "drug addict behaviour" but are in pain. Fractures which sometimes shows up on x-rays or not at all, however they show on bone scans but only after weeks have gone by.	P	O	
	Better pain relief	P	O	
	What is the most effective pain relief	P	O	
	What can be done to help with tendon pain /damage this is as difficult for me as bone pain	P	O	
	Are there any treatments or medications to help with the chronic pain?	P	X	
	What are the options for effective pain relief without the risks of opiate addiction (or at least minimizing those risks)?	P	X	
	Alternative medicine for pain relief Long term pain options (that are non pain meds or non narcotic)	P	F	
	What can help me with pain?	P	F	
	I would like to see help with pain relief,	P	F	
	What is the most effective pain relief? Can these work along with other medicines?	R	F	
	Why cant the patient be offered better pain relief?	R	F	
	Different ways of coping with long-term pain.	R	X	
	Something needs to be done about the pain that XLH'ers go through on a daily basis. Long term...they shouldn't have to be in pain for years.	R	X	

13. How does menopause impact on women with rare metabolic bone disorders?	OI:What is the impact on fracture risk of menopause in women?	H		
	Is anyone collating statistics on falls, bone density post menopause?	P	O	
	How will the onset of the menopause affect my condition?	P	O	
	Menopausal deterioration of bones.	P	O	
	I'm only 48 and menopause will be kicking in, so I'm wary of coming higher risk.	P	O	
	Any age-related deterioration in people with OI particularly women around the menopause.	P	O	
	does the menopause affect FD pain?	P	F	
	What happens after menopause with bone quality?	P	X	
	what should I expect to happen after the menopause?	P	O	
	Why are female patients not listened to by their dr/s when they are having hormonal issues, I.e. menopause. The pain and regrowth can increase during this time?	P	F	
	Menopausal effect upon bones	P	O	
	For females is post-menopause likely to increase risk of stress fractures and bone softening in the way that osteoporosis becomes a greater risk?	P	X	
	What happen with the menopause in women with XLH	P	X	
	Will sO see any changes after menopause?	P	F	
	Also more info and support re menopause effects.	P	O	
	As a 44 year old woman who has been treated since birth (my dad had it), I wonder about the implications on osteoporosis.	P	X	
14. How does care and support for adults need to differ from care and support for children? What are the best ways to support people through that change?	Is there anything more than best supportive then palliative care we can offer	H		
	Changing needs through life - how do they change and what services are required to support them?	H		
	transition from child to adult services.	H		
	All: How can we ensure transition from paediatric to adult care?	H		
	Should you stop seeing a dr. when you become an adult?	P	X	
	My grandchildren see a geneticist and endroconologist on a 6 monthly basis but there is not the facility for adults and I have never been offered any support as an adult.	P	O	
	Why is most research focussed on the impact on children? There appears to be very little management of adults wirh xlh.	P	X	
	Will there be any help for OI suffurers in old age but some time i could of asked a 100 n 1 questions	P	O	
	Is there any handover process in place for for teens to adult care?	P	O	
	Why are adults abandoned and left to deal with OI alone?	P	O	
	Why does support significantly reduce when you leave child services	P	O	
	I haven't seen an OI specialist since i was 15 as i was transferred to adult services and then no one was able to help as didn't know about OI.	P	O	
	I have a 21 the old also with OI and now abandoned. from aged 5 loads of excellent care.. Transition at 15 and fell through the gaps.now seeing endocrine who admits knows nothing of OI. It's really frustrating and unfair. Our GP despite being great is at a loss. Our local hospital only help in times of fracture but then discharged and left in pain and with no support.	P	O	
	Will there be more support for adults n managing conditions	P	O	
	Is someone going to be there for me medically through out my life.	P	F	
	Why is it that when children become adults they no longer have a consultant and just have to go to A&E and see a different doctor every appointment.	P	O	
	Long term management should be equal to treatment that children receive. A yearly appointment would be sufficient for many adults seeing all specialists in one appointment.	R	O	
	More support needs to be given at diagnosis and throughout?	R	F	
	Are there any alternative options to 'maintain' on going health once formal adolescent treatment has been ceased?	R	X	
	Why is the treatment & care within the NHS with an OI adult different from what a child receives?	R	O	
15. Are people with rare metabolic bone disorders at risk of any other health conditions?	what other illnesses might i encounter later in life due to xlh? are they preventable?	P	X	A small study suggested that early onset of hypertension was a complication of XLH, particularly if secondary hyperparathyroidism was present (Nakamura Y el al, Endocr J 2017; 64: 283-289).
	Is FD related to osteoporosis	P	F	
	I know that there's a link between Ehlers-Danlos Syndrome and endometriosis. I also know that EDS is a mutation of the col1a1 gene like OI. So is endometriosis connected to OI too? I know that around 10% of women have endo; is that rate of prevalence the same or higher (or even lower) in women with OI?	P	O	
	I also would like to know what conditions can be linked to XLH.	P	X	
	what kind of diseases or problems am i going to have because of the	P	X	
	Relation between arthritis and XLH	P	X	
	Also there's other things that go with it ie hyper mobility,ED, diabetics but this is never mentioned.	R	O	
16. Could a combination of self-management approaches reduce pain and prevent bone loss (e.g., exercise, diet, life-style changes, meditation, yoga)?	Does calcium + vitamin D actually help my condition.	P	O	http://www.nutritionjrnl.com/article/S0899-9007(11)00144-4/fulltext
	I would like to know more about what alternative there are to taking pain drug on a regular basis. Exercise Diet Life style changes Meditation	P	O	
	What's an appropriate daily calorie intake for an xlh adult?	P	X	
	also telling patients about exercise and diet.	P	O	
	Are there any vitamins for adults that could help ease joints ceasing up	P	O	
	Should I avoid caffeine or fizzy drinks	P	O	
	and diet	P	X	
	Is there a specific diet XLHers should follow?	P	X	
	I have always been advised to "have any problems treated when they occur" mainly when I have asked about what I could do as a preventative measure but I feel I have then reported things too late for anything to be done. I understand everyone is different but I think XLHers should be given advice on what to expect in their future and what they could do personally to help themselves.	P	X	
	Is there a diet recommendation related to medical treatment? Does any foods negatively effect medical treatment?	P	X	
	Exercise, diet,	P	X	
	What foods/vitamins are recommended for people with Osteogenesis Imperfecta?	P	O	
	Does any specific diet help minimise symptoms?	P	X	
	I also want to know if there are other dietary methods to improve my current condition.	P	X	
	Are there any foods we should eat more/less of?	P	O	
	how we can help ourselves with yoga and mindfulness	P	X	
	Also what diets could help.	P	F	
	Should there be diet advice given to OI adults and children?	P	O	
	Best diet for overall bone health?	P	O	
	can diet help improve any part of the condition?	P	O	
	Condition-specific nutrition (bone-building)	P	O	
	Weight loss assistance due to reduced activity.	P	O	
	How can I loose weight when I'm in a wheelchair?	P	O	
	Should I take other supplements than D-vitamin and calsium?	P	O	
	What levels of D-vitamin should I have?	P	O	
	I would like to know what kind of diet would be helpful to me (what should I eat/not eat)? I'd like to know the levels of impact that different foods can have e.g. coca cola = really bad, sparkling wine = bad, black tea = acceptable. Nobody wants to give up everything, but if I'd know what are the really 'bad' foods I could make a decision on what to kick out completely and what to only eat sometimes.	P	O	
	Is there research regarding the usefulness of hemp oil/seeds and brown millet?	P	O	
	Diet Exercise	P	X	
	Are there specialist exercise programmes more suitable for people with our condition.	P	O	
	What exercise can I do safely and who can help me with creating a manageable exercise plan?	P	O	
	diet.	P	X	
	What can I do to better improve my condition in terms of lifestyle, diet and exercise etc?	P	X	
	diet exercise	P	O	
	What sort of supplements and exercise can be used to live a more active and painfree life or increase the quality of life for people affected by XLH	P	X	
	exercise and treatment	P	X	
	How to use the diet in people with XLH	P	X	
	Is there anything I can do so I won't end up in a wheelchair one day?	P	X	
	What exercises can i do to help alleviate pain?	P	X	
	any lifestyle changes I can make to lessen the impact of XLH on day to day life.	P	X	
	Best diet for weight loss and weight lifting and exercise for bone health	P	X	
	What foods are beneficial for improved bone health once osteoporosis is diagnosed?	P	O	
	The effects of diet on bones, muscles (reduce cramping especially).	P	O	
	Are there foods more likely to keep tumour active?	R	F	
	I want to know if any real research is being conducted as far as healthy BMI for a person with XLH.	P	X	

17. What is the best way to link up and organise all the health professionals who care for a person with a rare metabolic bone disorder?	Have we embedded enough thinking within primary care around rare diseases? How can we embed genomics in mainstream medicine into primary care?	H		
	How to best coordinate a multidisciplinary approach to care? Who should be involved?	H		
	Which health and social care professionals are best placed to deliver ongoing care and support?	H		
	How can long term care and support for these conditions be organised on a share care basis between specialist centres and more local facilities?	H		
	I would like XLHers to be monitored as a "whole" as opposed to being referred to various different Drs and Clinics.	P	X	
	What kind of team of doctors should one see.	P	F	
	Why do conditions like RA, Lupus and other rheumatic conditions have a multidisciplinary approach but OI patients are left without or fumble around to get help?	P	O	
	Why is there no multidisciplinary approach for adults with OI?	P	O	
	can gps be told what bloods tests and urine tests to monitor and how often?	T	X	
18. Which treatments are safer and more effective for people with OI (osteogenesis imperfecta), treatments that promote bone-building (anabolic treatments) or treatments that reduce bone loss (antiresorptive treatments)? Does combining treatments make a difference?	Would anabolic treatment for OI be better than bisphosphonate therapy?	H		https://www.ncbi.nlm.nih.gov/pubmed/27760454 https://www.ncbi.nlm.nih.gov/pubmed/27482615 https://link.springer.com/article/10.1007%2Fs11845-013-0995-x https://link.springer.com/article/10.1007%2Fs00223-013-9770-2 https://link.springer.com/article/10.1007%2Fs00198-011-1658-2 https://onlinelibrary.wiley.com/doi/abs/10.1359/JBMR.050312 https://onlinelibrary.wiley.com/doi/abs/10.1359/jbmr.2003.18.1.126 https://link.springer.com/article/10.1007%2Fs00223-017-0236-9 https://link.springer.com/article/10.1007%2Fs00223-010-9383-y https://onlinelibrary.wiley.com/doi/full/10.1359/JBMR.051015 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3904621/ https://link.springer.com/article/10.1007%2Fs00223-001-1055-5
	For OI:Is anabolic therapy better than anti-resorptive?	H		
	For OI:Effect of combination therapy	H		
	OI: Are anabolic treatments such as teriparatide or romosozumab of any benefit?	H		
	OI: Should adults with OI get medical treatment (antiresorptive or anabolic)	H		
	OI: Are anabolic agents more effective than anti-resorptive treatments in the prevention of fractures in osteogenesis imperfecta?	H		
	Are bisphosphonates harmful or helpful in this disease?	H		
	Should all OI suffers take Ibandronic Acid for bone thining.	P	O	
	Which bisphosphonate is the best for OI?	P	O	
19. What is the best form of exercise for people with rare metabolic bone disorders?	How can we maintain muscle strength when walking becomes limited and sports equipment is oversized?	P	O	
	Maintenance of levels with age? What type of exercise...hiit, yoga, weights?	P	X	
	Is running a good exercise for OI sufferers or is it too traumatic on the hips ,knees ,ankles.	P	O	
	Should I go for short walks	P	O	
	The best exercises to do for each part of the body, how long you should spend exercising each day/week and potential benefits.	P	O	
	What types of exercise can help avoid operations? Is there any recommendations, one can give physiotherapists who don't know anyone with XLH?	P	X	
	Is any exercise okay? What exercises is the best?	P	X	
	What is the best, easy exercise to do for someone with OI?	P	X	
	what can be done about lack of mobility	P	O	
	Do any specific exercises help minimise symptoms?	P	X	
	Some adults who were diagnosed as children have not been allowed to do any sports and then have muscle issues later in life, why is this?	P	F	
	As an adult with type 3 OI who hasn't had a fracture in 24 years which i put down to my level of continues activity, has any research been done to see what impact over all levels of activity has in relation to bone density.	P	O	
	What type of exercise can be done safely when recovering from/suffering the long term effects of injuries?	P	O	
	Condition-specific exercise (with hypermobility, other issues)	P	O	
	Appropriate exercise that won't damage or excessive tire	P	O	
	What kind of exercise should I do when recovering from surgery to keep the other parts of my body strong?	P	O	
	What kind of sports are useful to me?	P	O	
	What exercises can be done when recovering from fractures in the legs and currently unable to walk?	P	O	
	Can a suggested exercise regime be tailored to suit each OI type?	P	O	
	Should certain excerise's be avoided and others recommended?	P	X	
	What can I do to help my mobility in the longer term?	P	X	
	sport over 45 years old	P	O	
	What exercise is most advantageous for XLHers?	P	X	
	Is there help with exercise for this condition?	P	O	
	Should I be exercising despite lots of pain in my joints?	P	X	
	What exercise regimens are safe and effective for XLH patients, given the impact of weight-bearing exercise on soft bones.	P	X	
	What exercise or sport do other XLH'ers take part in as a child, and as an adult?	P	X	
	Exercise- feel monitored exercise would be preferred to inappropriate exercise at the gym or very limited physiotherapy. Ligamentous laxity can make even basic exercise very difficult or can cause injury.	P	O	
	What specific exercises can I do to build up bone density ?	P	O	
	What form of exercise, if any, is specially helpful in improving bone health once osteoporosis has been diagnosed?	P	O	
	What exercises are best to help bone density, pain management, flexibility.	P	O	
	Is exercising a good idea because my daughter was never allowed as a child and now has muscle waste?	R	F	
	Can there be more information on safe exercises to prevent fractures.	R	F	
	What exercises should be avoided	R	X	
	Whether the rolling balance exercise used for some sportspeople and some severely disabled people could be safely adapted for proactive use to improve conscious and unconscious balance to help prevent falls and other injuries. Every break changes the body's balance but it is hard to learn a new approach when habits are years old.	R	O	
20. How does drug treatment need to change as people with rare metabolic bone disorders get older?	Is treatment with bisphosphonates in adults with FD and OI effective?	H		
	Broad efficacy across the age spectrum Do adults need same treatment as children?	H		
	For OI:Do bisphosphonates reduce fracture risk in adults?	H		
	OI: are there any benefits to treating adults with OI with bisphosphonates? If so, who should be treated?	H		
	Do adults require phosphate medication?	P	X	
	Will my treatment need adjusting as I get older?	P	O	
	What about treatment for older OI suffers	P	O	
	What is the chnages to me made to medications along ageing ?	P	X	
	Do i need to take medication as an adult or not?	P	X	
	Is there differing treatments for OI depending on mild/moderate/severity of OI at childhood and when older?	P	O	
	I would like to know if I should have stayed on the treatment regimen I was on up until my late teens.	P	X	
	Is there a consensus on whether it is effective for XLH patients to take phosphates at all ages? It seems that opinion varies.	R	X	
21. Is stem cell therapy an effective treatment for people with rare metabolic bone disorders?	Stem cell treatments	P	O	
	Can stem cells be used to repair damaged cartilage, are there any trials I can take part in as joint replacements are not suitable for me and I think this treatment would be of great benefit for people with OI.	P	O	
	Stem cell research in adults?	R	O	
22. Why are some health professionals unaware of rare metabolic bone disorders and how can this be improved?	How can delays in diagnosis be reduced?	H		
	Diagnostic delay is agreed. Why? We can guess but don't know. Coding is inadequate. We need data accuracy - we can create this through a network of involved clinicians	H		
	Osteogenesis imperfecta was only diagnosed 2 years ago after a life time of fractures could this have been picked up earlier and treated quicker?	P	O	
	How can quick diagnosis be improved and quicker medical treatment be established?	P	X	
	I am disappointed that I was only diagnosed at the age of 41. After complaining of pelvic pain for years. (Over 25 years)	P	F	
	Why does it take so long? Why is it misdiagnosed?	P	F	
	Now have great doctors that answer my questions, but I was misdiagnosed for the first 40 years of my life. How can the medical community help those doctors who know nothing and pretend that they know it all and give wrong diagnoses?	P	X	

	It took a long time to get diagnosed with FD. But then I only underwent surgery to remove tumours on ribs and was left largely unmonitored other than yearly chest X-rays. Only a chance encounter with a brilliant endocrinologist put me in contact with RNOH which lead to Zol infusions etc.	P	F	
	My mum is nearly 70 and was only possiably diagnosed with it 5 years ago when I was admitted to hospital why does it take so long? Why was this not considered before for my mother.	R	O	
	Why is fibrous Dysplasia mistaken for bone cancer too often?	R	F	
	Why does diagnosis take so long?	R	F	
	As with most rare diseases, lack of awareness of the diagnoses hampers early diagnosis. Whilst primary care and many secondary care physicians cannot be expected to have detailed knowledge of such diagnoses, perhaps more work can be done to raise awareness of the possibility of rare causes of fractures, bone pain etc.	H		
	How do we counter general ignorance, of these rare conditions, within the medical/non-medical communities & disseminate clear information to receive better informed treatment & needs provision?	P	O	
	Why do doctors disbelieve you when you say you have xlh?	P	X	
	I feel that I don't get enough support for my OI and the doctors don't take me seriously.	P	O	
	Why, indeed, are there still many doctors and consultants who are unfamiliar with condition and therefore not in the best position to offer guidance/carry out treatment?	P	O	
	Why are medical staff (from nurses, GP's and even top consultants) still not fully trained or understand the complexity of the disease and how treatment needs to be adjusted to each individual patient as the disease is manifested in so many different forms?	P	O	
	Many GP and even some Hospital Rhumatologists do not know XLH at all and thus misdiagnosed and/or do not care helpin patients affected.	P	X	
	I have no doctor / consultant who seems to know anything about my condition	P	X	
	Why is there not a central database of information for local doctors, dentists, physios etc to access when I go to them and say I have XLH? It would be helpful to be able to direct them to information that they can read before they begin to treat me and possibly have no effect or worse, no damage.	P	X	
	There still seems to be a lack of knowledge of OI in the medical profession. Are rare conditions taught in teaching hospitals?	P	O	
	Why don't hospitals have an awareness on how to deal with patients with OI.	P	O	
	Are dentists going to be made aware of the effect this condition has on patients teeth?	P	X	
	Most information those who have OI get are from social media connections, this is how we get our answers because medical professionals don't seem to either talk to each other or share their information. How can this change? How to make more up to date stats and information available?	P	O	
	How to explain to a doctor that people with OI feel pain like everyone else, they have just been in physical pain for much of their lives and deal with it differently. On the other hand, as an adult I'm just so tired of "dealing with the pain" and want it to stop or at least diminish enough to have a life. But am told "some people just can't handle pain so they ask for pills" and I should just "deal with it" since I don't LOOK like I'm in pain.	P	O	
	Are new medical staff going to be taught more on rare conditions	P	O	
	Long time care on hospitals and consultants knowing all the infotabout XLH and how to tem rear I properly	P	X	
	symptoms we have in common so doctors will believe us	P	F	
	I have great doctors that answer my questions. But many other are not so lucky. How can the medical community help those doctors who know nothing and pretend that they know it all and give wrong treatment to patients of rare diseases?	P	X	
	I have great doctors that answer my questions. But many other are not so lucky. How can the medical community help those doctors who know nothing and pretend that they know it all and do not know how to monitor rare diseases?	P	X	
	Also when I was admitted into hospital with side pains both my mother and I told the doctors of her bone problems and that I had broken bones before without doing anything major but this was dismissed and X-rays weren't took. It took me a week of asking for an X-ray until they did one and realised I had three broken ribs. It's like they don't want to know if it isn't in a text book? That I feel oi isn't taken seriously.	R	O	
	Each case is different and different strengths. But if you don't tick all the boxes doctors seem confused.	R	O	
	Can gp's be more educated on these diseases as treatment and referral takes so long?	R	F	
	More training of spotting the diseases in medical schools or in house training sessions at hospital.	R	O	
	Doctors who don't know anything about XLH need to research and get in contact with the doctors who do know about it and learn everything about it to help their patients.	R	X	
23. If we have a better understanding of what causes rare metabolic bone disorders, will that help find new treatments?	What underlying biochemical abnormality can we target?	H		The vast majority of people with XLH have mutations in the PHEX gene (Capelli S et al, Bone 2015; 79: 143-149), leading to elevated FGF23 concentrations and hypophosphataemia (Imel EA et al, J Bone Miner Res 2007; 22: 520-526). This provides the ratiionale for treatment for XLH with the anti-FGF23 monoclonal antibody.
24. Would specialist services result in better care for people with rare metabolic bone disorders?	are there tertiary centre or consultants with special interest to refer those patients to	H		
	Why have we not designated specialist centres as clear referral routes for patients with these rare diseases?	H		
	How many centres should be providing care for these long-term rare conditions?	H		
	How can care be coordinated through specialist centres?	H		
	can there be a UK wide network of speciliasts who are available to be consulted on the conditions perhaps with the referring doctor, the patient and their carer being involved via a Teleclinic delivered by SKYPE?	H		
	It would be preferable for patients with these conditions to be looked after by one specialism, rather than be divided into paediatric and adult specialties.	H		
	There should be master classes in XLH and OI management to tiered groups of docs. We all do the same , then study it, measure effects, then change if not sufficient for prescribed outcome	H		
	proper pain management clinic that has consultants that know about all these medical conditions. And doctors that don't make you feel like a hypochondriac.	P	O	
	Are there specialist facilities available for older patients with my condition?	P	O	
	There should be specialised adult OI clinics GPS should be required to refer adult OI patients to specialist doctors	P	O	
	Shouldn't every child/adult with OI have allocated a specialist consultant to take over their case when they end up in hospital with fractures?	P	O	
	Orthopedic specialist for joint replacements?	P	X	
	Why are there no adult OI specialists in the UK? Seeing a rheumatologist is a waste of time.	P	O	
	My GP seemed/seems totally unaware and is not involved at all in my treatement. I feel I could have benefited greatly from an earlier referral to a specialist.	P	F	
	Should there be more specialist centres for rare diseases as smaller hospitals haven't seen these diseases before? Patient group has been more useful than the hospital.	R	F	
	Similarly for dental? Is it best to see a dental surgeon familiar with the condition?	R	X	
	Adults with OI still need regular check ups and this would be best placed in a centre of excellence, on a whole day to avoid multiple appointments requiring time away from work etc, why is this not done?	R	O	
25. Is life expectancy affected by rare metabolic bone disorders?	Will OI affect my life expectancy	P	O	
	How long will I live?	P	X	
	Will my life expectancy be diminished?	P	X	
	Is it true life expectancy is not affected with XLH? I feel like my health is poor and I'm 53. The pain is terrible and it makes moving hard to do.	P	X	
	Can this disease kill me	P	F	
	Can you live a normal healthy life expectancy?	T	X	
The remaining shortlisted questions (not in any order)				
Does pregnancy have an impact on women with rare metabolic bone disorders and their treatment?	Will I need to change my treatment if I become pregnant?	P	O	Pregnancy associated with polyostotic fibrous dysplasia. Med Ann Dist Columbia. 1948 Mar;17(3):157-9. NELSON RB
	I would like research into effects of pregnancy and/or breastfeeding upon bone density in OI patients and for this to create advice as to risk of pregnancy and breastfeeding to women and their bone density.	P	O	
	I would also like research into occurrence of spinal fractures in relation to pregnancy, for research to be done into why this happens in some and not others, if it can be prevented by treatment prior to or during pregnancy, and the risk of vertebral fractures with breastfeeding and in subsequent pregnancies and breastfeeding.	P	O	
	I would like to know if there is an association between OI and pregnancy induced osteoporosis as I am aware of several women who had serious spinal fractures in or immediately after pregnancy.	P	O	
	Is there any way of using medications to optimise bone density during and after pregnancy and breastfeeding?	P	O	
	What is the research being done on new lesion growth particularly after pregnancy?	P	F	
	OI women- more info and understanding re pregnancy and childbirth. Not just genetics.	P	O	
	My Autosomal Dominant type of Hypophosphatemic Rickets is what doctors call a symptomatic, however I showed signs of it during and after pregnancy in my blood tests and urine tests is this linked to hormone changes in the fgf23 gene?	T	X	

Would there be benefits to monitoring adults with rare metabolic bone disease? If so, what form should this take?	What is optimal follow-up and monitoring of these rare conditions?	H		
	monitoring during treatment-free periods	H		
	such rare conditions, there seems to be little info for any of them about ongoing support in terms of physio/orthopaedic opinion and monitoring.	H		
	How frequently should patients have screening with ECHO, hearing tests, etc	H		
	In oi what monitoring should be done ?	O		
	What monitoring should be done?	P	O	
	Is Parathyroid hormone monitored routinely in stable adults? Are DEXA scans done from time to time?	P	X	
	Why aren't adults monitored as closely as children?	P	X	
	Monitoring patients should be done by their needs.	P	O	
	We need heart control? What kind of control? A sonography? An electrocardiogram?	P	O	
	Can we be checked regularly by someone who knows more about condition as i feel as getting older bone pain and arthritis symptons getting worse and affecting my mobility	P	O	
	What monitoring should be done, particularly with scoliosis of the spine which is related to OI	P	O	
	Should diagnosed people be called for a check up every 6 months	P	O	
	We have little adult monitoring. I have been waiting a year for bone scan results so far. There is so little interest out with fractures when is when actually i feel the safest!	P	O	
	I receive no regular monitoring and find it too painful to travel far, I have always been led to believe I just have to put up with it and no help is available.	P	O	
	Will I continue to be monitored into old age?	P	X	
	Why is muscular monitoring not done? - that in theory strong mucles could help prevent bone softening. Reseach based recommendations about exercise and monitoring via hospital	P	X	
	How often should one have scans ? What type of scans ? I have only been monitored for short time when diagnosed: How regularly should one be monitored? How often should one be scanned ?	P	F	
	My aim would be to have regular scans to see change in fd	P	F	
	Monitoring the FD.	P	F	
	Why aren't a patients vitamin d levels checked more often?	P	F	
	Why can't the patient be given a isotope scan every five to ten years to check the bones, as other bones can be affected at a later stage.	P	F	
	How often should hearing tests be performed in adults and children with OI?	P	O	
	Is there standardised recommended advice for frequency of dexa scans and blood tests to monitor bone density throughout adulthood?	P	O	
	Long term monitoring ?	P	O	
	How often should I be monitored once I have been taken off the bisphosphonate treatment? E.g. Dxa scans	P	O	
	What health checks should be done for OI and how often?	P	O	
	What doctors or members of the medical community do you recommend that we should be seeing regularly?	P	O	
	Should I check my heart?	P	O	
	Who should follow up people with OI?	P	O	
	How often can I ask for a bone density scan? Or rather how often does it make sense to have one? Yearly?	P	O	
	Is there anything I should ask for the consultant or GP to have a look for?	P	O	
	If a person affected by osteogenisis imperfecta has tachycardia, do they require regular monitoring of their conditions?	P	O	
	Is there a monitoring programme for every child/adult with OI so that their condition and bone density is monitored and their needs are meet in terms of support with wheelchairs/equipment.	P	O	
	I feel as adults we should be monitored by specialists in this field every 6-12 months as when you age your body has to compensate	P	O	
	I wish I had been told what I should be monitoring at various stages of my life and hope I can be advised for my future health.	P	X	
	How often should adults have check ups	P	X	
	How often should calcium be checked and by whom, GP or hospital clinic, when known hyperparathyroidism?	P	X	
	How often should adults with XLH be monitored to prevent fractures and bone deterioration? What form should the monitoring take.	P	X	
	How often should i have medication review. What checks should be done?	P	X	
	Should I see an endocrinologist more often than yearly for management?	P	X	
	Who do I go to for.montiring and meds if I decide to do therapy as an adult?	P	X	
	Multidisciplinary approach whereby we do not go off the radar.	P	O	
	Is a 3 year review adequate?	P	O	
	I'm having very little monitoring No dexa scan for several years I haven't had my heart monitored for over 15 years. Maybe a more consistent approach across the country.	P	O	
	What kind of care should I have now that I am stable?	P	X	
	Monitoring for bone density and cardiovascular health	P	O	
	Alkaline Phosp seems to be the only convenient metric/marker other than 2 yearly CT scan and 5 yearly Technetium bone scan. Is there anything better planned for FD treatment & monitoring?	P	F	
	If you are on pain relief could it be monitored more frequently i.e regular blood tests or medicine checks.	R	F	
	How often should patients have bloods and scans?	R	X	
	Why can't the patient have more frequent bone scans to check other bones later in life?	R	F	
	Scans? Recommended and if so why? If not why? X-rays? As above Blood or Urine samples? As above.Any other form of recommendations of monitoring?	R	X	
	It's hard to treat patients with ADHR and xlh as there isn't a lot of information available for them or gps and it feels like you have to jump through hoops to get routine adult check ups done can anything be done to change this?	T	X	
How are deformities in the skull (basilar invagination) best detected and treated?	OI:Whom and how often to screen/monitor for basilar invagination?	H		
	There should be monitoring of Basilar invagination for all OI patients	P	O	
	Has there been an increase in deaths through Basilar Invagination? Is there any records of deaths by Basilar Invagination?	P	O	
	Is there treatment for Basilar Invagination?	P	O	
	What is the best treatment for basilar invagination?	P	O	
Would women with rare metabolic bone disorders benefit from monitoring and treatment during menopause?	What kind of treatments are better for woman in the menopause?	P	O	https://www.ncbi.nlm.nih.gov/pubmed/6727948
	What are the best ways of coping with the condition as a female patient who is in her mid-forties and finding fractures are occurring more frequently? Is there a way of managing the condition as you approach old age?	P	O	
	Eg. I am in the menopausal stage of life, what should I be doing to help myself?	P	X	
	What medications are available for people with Osteogenesis Imperfecta after the menopause?	P	O	
	What are the most effective ways of monitoring and treating bones and fragility during and after menopause?	P	O	
	Can surgery and alternative medicines be the same as before menopause and be as effective?	P	O	
	What help is there for adults with OI especially menopausal women	P	O	
	As many of the osteoporosis drugs used for OI seem to be ineffective, should HRT now be considered a better alternative for ladies after the menopause?	P	O	
	Should post menopausal women with OI be treated with biphosphates and which one?	P	O	
	There should be more information on treatment of post menopausal women	P	O	
	Is there any particular management that should be adopted in preparation / during / after an OI woman goes through the menopause?	P	O	
	Should I be tested for osteoporosis? MENOPAUSE	P	O	
	I am now menopausal, what signs should I be looking out for in regard to XLH?	P	X	
	Am I going to need extra monitoring when I'm going through menopause?	P	X	
	My bone density has dropped further following the menopause can anything be done to build the bones back up \?	P	O	
	Can female patients be checked for changes to bones and hormones during menopause?	R	F	
How could current devices used to treat people with rare metabolic bone disorders be improved?	Devices: Are they adequate? Can they be improved?	H		
	There must be things to help make the bone heal easier and rather than plaster casts, maybe breathable casts	P	O	
Are complementary therapies safe and effective for adults with rare metabolic bone disorders?	What about alternative medicine?	O		
	Is chiropractic care appropriate?	P	X	
	Is it okay to see a chiropractor?	P	X	
	Is there any alternative medicin that can help also?	P	X	
	More information on alternative medications?	P	F	
	Alternative meds	P	X	

	Alternative medication	P	X	
	alternative medicines	P	X	
	Is there any natural remedies or medication that can strengthen are bones besides doing the bisphosphonates treatments? Could the medical community research in to alternative natural treatments?	P	O	
	What alternative medicines are there?	P	X	
	Alternative medicines with a scientific point of view	P	X	
	Are there any alternative medicines I can try?	P	F	
	Are there all natural medical treatments?	P	X	
	Alternative medicine like acupuncture and massage for pain. Link to chiropractic stroke (I had one at age 29 from a neck adjustment causing an vad).	P	X	
	Would like to know about alternative medicines	R	O	
	Alternative treatments?	R	X	
	Is massage safe? Other alternative therapies?	R	O	
Is it possible to predict which patients with rare metabolic bone disorders will respond to a particular treatment, or might not need treatment?	what is the impact of not taking medicine.	P	X	Outcome of Long-Term Bisphosphonate Therapy in McCune-Albright Syndrome and Polyostotic Fibrous Dysplasia. Majoor BC1,2, Appelman-Dijkstra NM1,3, Fiocco M4,5, van de Sande MA1,2, Dijkstra PS1,2, Hamdy NA1,3. J Bone Miner Res. 2017 Feb;32(2):264-276.
	Long term risks with suboptimal biochemical reponse in X linked hypophos rickets	H		Prognostic Factors From an Epidemiologic Evaluation of Fibrous Dysplasia of Bone in a Modern Cohort: The FRANCEDYS Study. Benhamou J1, Gensburger D1, Messiaen C2, Chapurlat R1. J Bone Miner Res. 2016 Dec;31(12):2167-2172.
	For OI:Better understanding of responses related to underlying genetic defect	H		A retrospective study on craniofacial fibrous dysplasia: preoperative serum alkaline phosphatase as a prognostic marker? Ma J1, Liang L, Gu B, Zhang H, Wen W, Liu H. J Craniomaxillofac Surg. 2013 Oct;41(7):644-7
	FD: which patients benefit from bisphosphonate treatment?	H		An instrument to measure skeletal burden and predict functional outcome in fibrous dysplasia of bone. Collins MT1, Kushner H, Reynolds JC, Chebli C, Kelly MH, Gupta A, Brillante B, Leet AI, Riminucci M, Robey PG, Bianco P, Wientroub S, Chen CC. J Bone Miner Res. 2005 Feb;20(2):219-26.
	OI: If they should, who will benefit?	H		Pain in fibrous dysplasia of bone: age-related changes and the anatomical distribution of skeletal lesions.Kelly MH1, Brillante B, Collins MT. Osteoporos Int. 2008 Jan;19(1):57-63.
	OI: What are indications for medical treatment	H		Disease severity and functional factors associated with walking performance in polyostotic fibrous dysplasia. Paul SM1, Gabor LR2, Rudzinski S3, Giovanni D4, Boyce AM5, Kelly MR6, Collins MT7. Bone. 2014 Mar;60:41-7.
	Less severe cases of Osteogenesis Imperfecta presenting in adults need better identification, so that treatment can be more specific	H		Fracture incidence in polyostotic fibrous dysplasia and the McCune-Albright syndrome. Leet AI1, Chebli C, Kushner H, Chen CC, Kelly MH, Brillante BA, Robey PG, Bianco P, Wientroub S, Collins MT. J Bone Miner Res. 2004 Apr;19(4):571-7. Epub 2003 Dec 22.
	I was never prescribed any drugs or treatment (other than general treatment for broken bones) Why is there such a broad spectrum of treatments for OI	P	O	
	Does a person with osteogenisis imperfecta have to undergo treatment in order to sustain a normal life?	P	O	
	Can genome testing be done to help find the gene fault so other family members can be matched and diagnosed quicker as well as offering better support for the exact condition.	P	O	
What is the best way to manage craniofacial fibrous dysplasia?	What types of surgery are best for the face?	H		Long-term outcome of optic nerve encasement and optic nerve decompression in patients with fibrous dysplasia: risk factors for blindness and safety of observation. Cutler CM, Lee JS, Butman JA, FitzGibbon EJ, Kelly MH, Brillante BA, Feuillan P, Robey PG, DuFresne CR, Collins MT. Neurosurgery. 2006 Nov;59(5):1011-7; discussion 1017-8.
	What is the optimal treatment strategy for patients with CF FD?	H		Orbitocranial Fibrous Dysplasia: Outcome of Radical Resection and Immediate Reconstruction With Titanium Mesh and Pericranial Flap. Fadle KN1, Hassanein AG, Kasim AK. J Craniofac Surg. 2016 Nov;27(8) (n=22)
	Surgical treatment for craniofacial FD.	P	F	Three-Dimensional Printing of Reduction Template in the Contouring of Craniofacial Fibrous Dysplasia. Wang R1, Li G, Liu C, Jia C, Han Y. J Craniofac Surg. 2016 Oct;27(7):1792-1794
	Are there advances in treating facial symmetry issues?	R	F	Surgical Management of Polyostotic Craniofacial Fibrous Dysplasia: Long-Term Outcomes and Predictors for Postoperative Regrowth. Boyce AM1, Burke A, Cutler Peck C, DuFresne CR, Lee JS, Collins MT. Plast Reconstr Surg. 2016 Jun;137(6):1833-9.
				Optic nerve compression in craniofacial fibrous dysplasia: the role and indications for decompression.Tan YC, Yu CC, Chang CN, Ma L, Chen YR. Plast Reconstr Surg. 2007 Dec;120(7):1957-62.
				Strategies for the Optimal Individualized Surgical Management of Craniofacial Fibrous Dysplasia. Denadai R1, Raposo-Amaral CA, Marques FF, Ghizoni E, Buzzo CL, Raposo-Amaral CE. Ann Plast Surg. 2016 Aug;77(2):195-200
				Treatment of fibrous dysplasia orbital deformities with digital imaging guidance.An G1, Gui L, Liu J, Niu F, Chen Y, Wang M. J Craniofac Surg. 2015 Mar;26(2):449-51
				Craniomaxillofacial fibrous dysplasia: conservative treatment or radical surgery? A retrospective study on 68 patients. Valentini V1, Cassoni A, Marianetti TM, Terenzi V, Fadda MT, Iannetti G. Plast Reconstr Surg. 2009 Feb;123(2):653-60 (n=68)
				Optic nerve compression in craniofacial fibrous dysplasia: the role and indications for decompression. Tan YC1, Yu CC, Chang CN, Ma L, Chen YR. Plast Reconstr Surg. 2007 Dec;120(7):1957-62.
				Optical Coherence Tomography in the Management of Skull Base Fibrous Dysplasia with Optic Nerve Involvement. Loewenstern J1, Hernandez CM2, Chadwick C2, Doshi A3, Banik R4, Sarkiss CA2, Bederson J2, Shrivastava RK5 World Neurosurg. 2018 Jan;109
Shortlisted single questions (asked only once by a single respondent)				
What is the best treatment for osteoporosis in someone with XLH?	What treatment is recommended for a diagnosis of osteoporosis with XLH?	P	X	
What are the best kinds of shoes to provide ankle support for people with rare metabolic bone disorders?	Research based recommendation about ancles and shoes	P	X	
Are there any drugs or alternative medicines that can help preserve the collagen in skin in someone with OI?	We all know there is medication to help strengthen the bones but my skin has become more fragile with age and prone to pressure sores. Are there any drugs or alternative medicines that can help preserve the collagen in an O.I. skin?	P	O	
Do statins lead to muscle weakness in people with OI?	Could statins have greater risk of muscle weakness in patients with OI?	R	O	
Are rare metabolic bone disorders more severe in men or women, or are they the same?	With regards to severity, are there differences between men and women?	P	X	Although XLH is twice as common in females as males (Whyte MP et al, J Clin Endocrinol Metab 1996; 4075-4080), there is little evidence of a gender difference in severity, although there was a trend (p=0.07) for increased severity in males in one small study (Beck-Nielsen SS et al, Calcif Tissue Int 2010; 87: 108-119).
How does having a rare metabolic bone disorder impact on pregnancy?	Long term effects on life - pregnancy	R	O	
Questions already answered by previous research				
Could diagnosis in adults be made faster and more accurate e.g. through use of genetic or other biomedical tests?	Improved diagnostic tests would be useful, and hopefully more detailed genetic information may come from projects such as the 100,000 gene own project.	H		Although ideally the diagnosis of XLH would be made by identifying the mutation in the PHEX or other associated genes, together with an elevated FGF23 concentration, a small study suggests the use of high FGF23 and low concentrations, compared with age-dependet reference ranges (Endo I et al, Bone 2008; 42: 1235-1239).
	OI&X: is there any role for genetic testing in adults for making a diagnosis?	H		GNAS mutation detection is related to disease severity in girls with McCune-Albright syndrome and precocious puberty. Wagoner HA1, Steinmetz R, Bethin KE, Eugster EA, Pescovitz OH, Hannon TS. Pediatr Endocrinol Rev. 2007 Aug;4 Suppl 4:395-400.
	diagnostic testing methods short of bone biopsy and where these tests can be accessed	H		Searching for somatic mutations in McCune-Albright syndrome: a comparative study of the peptidic nucleic acid versus the nested PCR method based on 148 DNA samples.Kalfa N1, Philibert P, Audran F, Ecochard A, Hannon T, Lumbroso S, Sultan C. Eur J Endocrinol. 2006 Dec;155(6):839-43
	being diagnosed quickly	P	F	
	Identification of a more definitive test (other than genetic testing) to aid in the diagnosis of new patients eg role of FGF levels or other biochemical markers	P	X	
	Can genome testing be done to help find the gene fault so other family members can be matched and diagnosed quicker as well as offering better support for the exact condition.	P	O	
	How accurate is diagnosis from imaging from MRI's, CT, Xray and bone scan? I was originally diagnosed with an aneurysmal bone cyst which was operated on 6 years prior to the new diagnosis, because the lesion had returned	P	F	
	Is there a diagnosis pathway for people will all types of OI?	P	O	

	Was diagnosed pre-birth with type 3 OI. However looking at other OI peoples pictures, and only after having taken Fosamax during childhood, do I now look more like a type 4. However my body still "acts" like a type 3 (bones break just as much as they did in childhood). For example breaking ribs when sneezing. How can someone switch types (if at all) or look a different type but bones still respond to original diagnosis. How to explain to medical professionals who "look" and see a certain type but it's only on the outside. Inside (bones, ligaments etc) still act like the previous type originally diagnosed.	P	O	
	My first question relates to the diagnosis with the bone scan and MRI ,it seems that they only mark the largest area and neglect to add smaller lesions that also contribute to painful areas.	P	F	
Single question	For FD: What is optimal imaging technique?	H		CT and MRI in the evaluation of craniospinal involvement with polyostotic fibrous dysplasia in McCune-Albright syndrome. Bulakbaşı N1, Bozlar U, Karademir I, Kocaoğlu M, Somuncu I. Diagn Interv Radiol. 2008 Dec;14(4):177-81. The role of radionuclide bone scintigraphy in fibrous dysplasia of bone. Zhibin Y1, Quanyong L, Libo C, Jun Z, Hankui L, Jifang Z, Ruisen Z. Clin Nucl Med. 2004 Mar;29(3):177-80.
Out of scope questions				
Questions about quality and access to services	what are the gold standard treatment or any pathway existing	H		
	The lack of specialist adult services for individuals with rare bone disease.	H		
	OI - now that gene testing is much less expensive, she would be planning gene testing for all?	H		
	What does good practice and long-term care look like?	H		
	Criteria for treatment initiation - drug policies Criteria for treatment discontinuation - drug policies Who funds the drugs? Who gate keeps the drugs?	H		
	H: How effective are we at case finding in parents and adult relatives of children with HPP?	H		
	FD: How well do oncologists liaise with specialist metabolic bone services, when cases are identified?	H		
	OI: How do we ensure adequate orthopaedic review	H		
	How can patients get to the right specialist more quickly?	H		
	As for all progressive disabling disease in childhood which stretches into adulthood, how will home care be provided when parents are no longer able to cope?	H		
	What are the best strategies to get the patient to the adequate specialist?	H		
	What are the most effective diagnostic pathways?	H		
	Best treatment pathway?? Again, A clearly defined pathway	H		
	Access to genetic testing	H		
	Better referral pathways and networks-there are too many obstacles stopping patients getting to the right doctor	H		
	Would it be possible for GPs to appreciate that bone profiles are abnormal without them getting worried if results are stable? Maybe a letter on file from hospital? Would save GPs time and money.	P	X	
	Education packages for employers. How can they help keep us in the workforce	P	O	
	Are their any consultants willing to take on OI adults? Is there anyone willing to manage and monitor adults with OI?	P	O	
	How come occupational health is less funded in adult servces	P	O	
	I just think it would be good if services where more linked. I hear about other people my age seeing specialist while I've had one scan and no results til this sept..Scan was in January!	P	O	
	I get a bone density scan every two years but the hospital doesn't go into anything about it. It's like they are going through the motions and that's it. Going through my medical records ...When I was 6 they thought I had OI, considered inconclusive and dropped, so there aren't enough medics aware of it. Only thing the hospital has said 'whatever it is you are doing, keep doing it'. I do nothing! Consultant told me 13 years ago my back would fracture again at any time. I told recall the level of pain I had then. I couldn't go through that again.	P	O	
	When I have a pain, my first thought is fracture. On occasion I have rang the doctor asking they would refer me to A&e for X-ray just to be sure. Instead they make me attend the clinic, and they end up referring me anyway. What a waste of their time. By knocking out the fracture factor, I'll cope with the rest. I think anyone with OI should be able to check in with A&e without a whole load of hassle. Only want the X-ray and for someone to tell me that there is no fracture.	P	O	
	Is there opportunity to have beneficial treatment such as the Alexander Technique provided for on the NHS? I have found this extremely beneficial for my OI but at £68 a session I cannot afford to continue on a regular basis.	P	O	
	Why does people with conditions like OI have to be grouped in with others when assessed for equipment,my GP referred me for a power chair as I can walk very little and it causes extreme pain and fractures yet as I can transfer I don't qualify,OI is rare and it's unfair I am housebound because I don't fit into a box	P	O	
	finding the right doctors	P	F	
	Licensing of the new xlh treatments asap in england. Currently on trial	P	X	
	Long term guidance on whether adults should be treated	P	X	
	Unless there is a break or a fracture why is no other treatment offered?	P	F	
	Is there a standardised pain management plan for patients with long term pain with OI?	P	O	
	But also, where can I get help? In Germany it is so rare, most of the doctors and professionals have never seen an OI sufferer.	P	O	
	Can dental treatment for ol sufferers be exempt from charges	P	O	
	Is there a check-list in hospitals for babies born with suspected OI?	P	O	
	Is there a criteria for treatment in adults?	P	O	
	Long-term plans for quality of life	P	O	
	Better attitudes from medical professionals, more knowledge around pain relief and its effectiveness	P	O	
	Difficulties with the benefits system DWP. Stress in dealing with them.	P	O	
	Why is there nobody I can get help from. No GP is aware of the condition. No dentist can help. I get passed from person to person yet get no treatment.	P	X	
	Why isn't growth hormone considered a standard of care for kids with XLH and therefore covered by insurance?	P	X	
	What is the standard of care for newborns born to a mom with XLH?	P	X	
	Why aren't all patients with this condition offered physiotherapist appointments?	P	X	
	Only one ortho when I was a child, now as an adult the discipline is specific but no one wants to be responsible for care.	P	O	
	Why do patients with rare conditions still come under a wide criteria fr many conditions,such as refusing power chair if u can transfer	P	O	
	Should there be routine testing of brothers/ sisters and children irrespective of whether they have fractured	P	O	
	I take co-codamol for pain relief why wouldnt the doctor give me something stronger	P	O	
	What is the standard of care for XLH? (At present, there's no official standard of care for either kids or adults.)	P	X	
	I think adult manifestations are relatively undocumented, and lots of misinformation exists, therefore adult care appears to be inconsistent, even in the UK.	P	X	
	Why does the treatment vary so much between patient, medical professional, and country?	P	X	
	What are the first indicators that a GP should recognise before referring to a specialist?	P	X	
	Why do some doctors do surgery o.j. the tumor and some recommend against it? Why won't my doctors take it out and yet i have seen other patients have it removed.	P	F	
	Why is EDS and other similar conditions given more support?	P	O	
	Wheelchair services- why are such inappropriate chairs given to OI patients?	P	O	
	Why are OI patients often dismissed at A&E if fracture not visible on x-ray when many are able to describe a very specific "break pain" that often shows they were correct after the event.	P	O	
	Alternative Medicine- refused by NHS due to having a chronic condition.	P	O	
	Why is OI disregarded by the medical profession in the main- unless it's an ortho job or a severe OI case then management is very limited.	P	O	
	Why is there so little awareness or recognition of the connective tissue element of OI and its debilitating effects? why do medics focus on the broken bone element of OI almost to the exclusion of everything else?	P	O	
	Why are orthopaedic consultants so unwilling to refer adults with OI to the only adult OI specialist in the UK when they have problems that the orthopods claim to be unable to fix?	P	O	
	Living rurally shouldn't mean that you receive less help but that has been my experience.	P	O	
	I have found hydrotherapy to be beneficial but six weeks is not sufficient for lifelong condition.	P	O	
	Nhs is better at treating acute problems and I'm sure more could be done for complex chronic conditions.	P	O	
	How can universities teach doctors to be humans and listen to patients? When we say we are in pain, we are in pain. When we see bowing in our kids legs we see bowing. Rejecting patients feelings and symptoms does not help in the healing process.	P	X	
	Will the hospital keep the patient on their books or will they discharge them as they get older. After surgery once, my daughter was discharged and had to be re-referred?	R	F	
	Why does it take so long for the diagnosis or a hospital referral from gp? Are the dr's looking for other diseases as well?	R	F	

	Lack of adequate understanding of OI or of risk assessment for routine care of OI patients while hospitalised or in physiotherapy for OI and for non-OI conditions. Failure to provide electrically assisted bed led to re-opening of surgical wound, twice;physiotherapy injury; injury while being moved by wheelchair without foot rests.	R	O	
	There is clearly not enough training in medical school or specialist orthopaedic training to cover specific conditions and this creates a divide in treatment depending on where a person lives. What is being done to rectify this?	R	O	
	my two boys are routinely checked brilliantly and I have easy contacts numbers straight to their specialists endocrinology nurses and consultants can this be done for adults with rare diseases so they can get bloods checked more routinely or do you need to get gp referrals?	T	X	
Questions asking for information and advice	what treatments are available if any is there any ongoing clinical trials for treatment we can refer patient to be included in	H		
	Is treatment of XLH with the antibody to FGF-23 effective and safe?	H		
	X: does the antibody to FGF-23 improve the healing of pseudofractures? Is it both safe and effective?	H		
	How to approach new scientific developments (CRISPR-Cas9 ect.)?	H		
	Which diagnostic means are reimbursed?	H		
	Teriparatide in OI	H		
	Oi related: in Italy we use nerixia, is there another drug that can be used? If so what is it?	O		
	if the oi gene is not diagnosed yet, is it correct to start with bifosfonates?	O		
	How do you find a surgeon / cosultant competent in FB Do you see a rheumatologist? Endocrinologist?	P		
	Are there any possible ways to get help acquire resources in an urban area to assist OI families.	P		
	How do I know that I am getting all the support and treatment available?	P		
	Can anything be done for deteriorating hearing after double stapedectomy?	P		
	Can I help your research in any way?	P		
	How can one get good treatment for dental problems as an adult with XLH within the NHS?	P		
	How to find out what treatments are available for adult sufferers, the outcomes of treatments, i.e.How successful? Accessing new treatments?	P		
	Will I be informed of the development of new treatments as time goes on?	P		
	Biphosphonates: time? what happened after 8 doses of zolendronate?	P		
	What about BPS-804 of Mereo Biopharma?	P		
	What treatments are available to help manage the condition and reduce the risk of fractures. Particularly for those that have had the condition for a number of years and not under any specialist care.	P		
	Is there a test can be done before pregnancy to see if gene there as i have 2 boys and 1 is getting married they dont have O..I.	P		
	When are the new clinical trials for injection for brittle bones diesease coming into action	P		
	what spinal operations are available for a OI sufferer with Degerative Disc Disease.	P		
	What medications are any adults taking?	P		
	What pain relief is available?	P		
	What operations are available?	P		
	What pain relief/alternative medicines should I use to prevent/reduce joint swelling?	P		
	My left ankle is particularly bad, can this be improved by an operation?	P		
	Side effects of drugs and any contraindications.	P		
	Efficacy and method of action of drugs.	P		
	Risks with operations, along with recovery periods.	P		
	The best time of day/frequency to take drugs to receive the optimum benefit.	P		
	if there are any more drug alternatives for xlh?	P		
	Is their an international recommendation for medical treatment. (medicin, dosis formula, number of times for phosphat etc)?	P		
	How come the fetus don't show physical symptoms before after being born?	P		
	Is there any research on operations? What type of operations do people who suffer from XLH typically get?	P		
	Has there been any change on the Residronate seemingly not working on adults front?	P		
	What is actually done with bone samples taken via operation?	P		
	What is the best way to access local physiotherapy?	P		
	What is the best way to determine whether I have OI- genetic or clinical?	P		
	Family planning issues- what can be done if I wanted to have a family?	P		
	and I am confused as to if any treatment would help with the pain	P		
	I am unsure as to what treatment I should be receiving	P		
	When will the new KRN23 be approved for patients? Will KRN23 be available soon enough for the course of my life to change?	P		
	If my kidney was transplanted into another healthy person would it give them XLH?	P		
	And finally, I am very eager to know if it will be possible to get the KRN23 drug and how soon it will be readily available to me as an Irish citizen living in France.	P		
	My physical condition has declined greatly in the last 10 years, but even more so in the last 5. I am again experiencing joint and bone pain daily, my spine has shrunk and curved and I seem to be developing bone spurs and solidification of my tendons. What needs to be done? What questions should I ask my GP here?	P		
	Is there a register that we could sign up to to ensure we receive relevant updates about research and treatments available?	P		
	Do we need formal diagnoses to access benefits now or in the future? Although I clearly have OI I have never been formally diagnosed and would not know how to go about it.	P		
	What tests do you do for diagnosis of Fibrous displasia / McCune Albright syndrome?	P		
	I am 3 foot tall and weigh just under 4 stone. Is it safe for me to take an adult dose of any drug?	P		
	What are the real dangers of having had many many X-rays. Should I try to avoid them as much as possible to reduce health risks?	P		
	Do operations make me grow more	P		
	How does one get this diagnosis? MRIs, biopsy, etc?	P		
	Is there any patient advocate organisastions for FD in europe?	P		
	I would like to know what treatments are available for me.	P		
	More needs to be explained to the patient about the hormone part of the disease (MAS)?	P		
	How long does the recovery take after operations?	P		
	What help can I get with my FD?	P		
	A leaflet on what to do next after being diagnosed with FD.	P		
	Wishing people understand what FD is all about.	P		
	More social meeting groups.	P		
	What research is currently underway for drugs that act differently to bisphosphonates and how can we be involved? (Ie teriparatide)	P		
	Is there a list of unrelated medications OI patients should not take long term due to them lowering bone density?(eg PPI's)	P		
	are there alternate medications other than bisphosphonates?	P		
	Is it possible for me to wear contact lenses or get laser eye surgery?	P		
	With all the current osteoporosis drugs on the market, which one has shown the best results for Adults with OI, and which ones have proved ineffective?	P		
	Whether to try and keep walking with aids or to become v. Active wheelchair user.	P		
	OI and the workplace. More OT practical input to help stay in work or enter work. What is out there? Same with personal care, ILF, canine partners. Need info on all the services and options available?	P		
	Aids and adaptations to assist.	P		
	Personal assistance help to be able to work- help with information re this.	P		
	Is there treatment for hearing loss? Opinions seem to vary regarding surgery (stapedectomy) with a general blanket ban across the NHS yet available privately.	P		
	Is surgery available for hearing loss in Osteogenesis Imperfecta?	P		
	What treatments are there for adults apart from oral meds like risodronate?	P		
	Can trigger point injections be a good therapy for muscle knots/inflammations?	P		
	When should I check my hearing?	P		
	Where can I find a dentist who knows OI-teeth?	P		
	How should the doctor measure lung capacity?	P		
	What is the difference between a clinical diagnose and genetic?	P		
	How can scoliosis give stomach problems? Is my obstipation problems from taking tramadol or are they from scoliosis?	P		
	My question is more regarding the diagnosis if I would have children. How soon will it be possible to find out whether my child would have OI and which level it might have?	P		
	Is scuba diving bad for my bones?	P		
	My last break was my foot (5th metatarsal and ankle) it's over seven months ago and I still sometimes have pain and I haven't regained full flexibility, my physio finished my treatment and said I can go to get the fracture checked again a full year after I broke my ankle. But I'm worried that they missed something, what can I do to find out if my pain is just normal or related to OI?	P		
	What are the risks and benefits of biphosphate treatment?	P		
	What are the most effective forms of pain relief (non-drowsy) for joint and bone pain?	P		
	What are the benefits of genetic testing? What would they be able to find out? How is it done?	P		
	Is it too late to get assessed for treatment when your in your 50's?	P		

	If a person with osteogenisis imperfecta has bowed bones and has also suffered trauma/injury to that bone, how do they confirm if it is caused by osteogenisis imperfecta or if it is a result of the trauma/injury?	P		
	What monitoring should I have.	P		
	What medication should I have.	P		
	What pain relief can I have.	P		
	What are the side effects to all of the treatments available to me?	P		
	What treatment will benefit me as someone who doesn't fracture that often, but struggles with mobility etc?	P		
	Information and help for dental issues/ implants etc	P		
	up to date lists of recommended orthopaedic surgeon s and dental professionals and more information about help with dental issues; I now have had to pay thousands of pounds for dental work, particularly teeth implants with no help financially when the weakness and loss of my teeth is down to my XLH and med s overdoses as a small child.	P		
	id like to hear of examples of people doing genetic testing when having kids, about their experience in having kids	P		
	What is the standard of care when first diagnosed with XLH? Ex: medications, what type of physician specialists should be seen, what age should treatment start?	P		
	Does the diagnosis of XLH qualify for disability if symptoms are appropriate?	P		
	Is there any coverage or funding available for the frequent transportation needs, parking for appointments or hospital visits, food for family while a person with XLH is hospitalized?	P		
	How can I help those who see their GP or nurse to recognise those early signs? What are the obvious signs that the first level of patient care could identify?	P		
	Is there a risk of cancer due to taking pain meds over long periods of time?	P		
	Krn 23, waiting for it to become available to me	P		
	How often do I do blood tests ?	P		
	How soon will KRN 23 be available and how much will it cost ?	P		
	What can be done to help insurance companies cover long term physical therapy?	P		
	Has there been any less dental issues for XLH patients with the new advancements in treatmemts?	P		
	Should I continue to take Alendronic Acid and for how long? How effective is this treatment through an intravenous drip?	P		
	I have swelling post exercise in my ankle, knees - what is the best pain relief/treatment for that?	P		
	How many people are their known in the UK with XLH	P		
	Risks and benefits of phosphate and alfacalcidol treatment in adulthood.	P		
	ho do go to when a rod (rodding in tibias and femurs) has move out of its place and the ortho's for leg and knees can't seem to figure out who should take charge?	P		
	Where to get information on research being done for OI. Since we as patients need to find them and bring to our doctors and hospital visits. Up to date, not in a textbook that is a decade old. Accurate information available to the public, or willing to pay a fee if it's the case, just to get information.	P		
	Is it necessary to avoid OTC medication, supplements etc due to their Vit.D content if on prescribed analogue?	P		
	On OTC medication, eg antacids, where hypophosphataemia is indicated as a contraindication is it necessary to adhere to this?	P		
	If osteoporosis shows on DEXA scan is that really osteoporosis or osteomalacia?	P		
	For old injuries I still suffer a great amount of pain and discomfort with, can physiotherapy be arranged? Can long term physio be arranged for injuries that clearly haven't healed fully or properly?	P		
	Why am I not entitled to financial help in the UK for this condition? Medication and transport costs are very expensive especially on a monthly basis. Will help be available financially in the future?	P		
	What happens when I can no longer have zoledronic acid infusions and HRT	P		
	Why wont any doctor do anything about the rods protruding out of both my Tibias	P		
	What complications or risks could occur from pregnancy when on zoledronic acid?	P		
	Will the much talked about 'wonder treatment' KRN23 be effective for adult bone pain where the cause is unclear and may be related to arthritis also?	P		
	What is the likely prescribing scenario for KRN23 ie what level of problems will be considered - only those presenting with continual pain or fractures?	P		
	What treatments are out there	P		
	Is there options for support days where patients with these rare conditions can get together yearly to support one another, raise money, awareness, talk to experts in a less formal environment and where families can be together to discuss the issues we face daily?	P		
	How can I talk to my doctor about pain relief without them thinking I'm a drug addict?	P		
	What can I do to find a doctor who knows about XLH and how can I find a doctor who will treat it properly?	P		
	When the new treatment will become freely available and if it will be funded on the NHS this is my main concern.	P		
	Also the use of medications in adulthood	P		
	Yes. I have done some research on things to strengthen bones. One of the things I discovered was oral oxandrolone (anavar). My findings is that it can be a replacement for HGH. It increases osteolytic bone resorption. Would this be beneficial for those with XLH?	P		
	Where do i go to find a Dr that has experience treating adults with this disease.	P		
	Should i see an endocrinologist even if i am not on any treatments?	P		
	Who do I see and how do I deal with the chronic pain aspect of this disease as an adult?	P		
	What are the pros and cons of treatment for adult patients?	P		
	What are the pros and cons of genetic testing as compared to diagnosis based on x-rays and lab tests?	P		
	What frequency do people with XLH have mobility issues?	P		
	What could a parent have done better for an earlier diagnosis?	P		
	I would like to know more about adult life, and end of life care for XLH.	P		
	How many diagnosed have lesions in multiple bones?	P		
	How many have lesion growth after puberty?	P		
	Are rods used for supporting the humorous?	P		
	What mg of bisphosphonates is recommended for monostoic Fibrous Dysplasia?	P		
	What will happen if it worsens?	P		
	How do biophosphate treatments workz?	P		
	I would like a list of professional doctors and research groups to help me. My current Drs are doing the best they can with the resources they have.	P		
	What team of doctors should i see for fd in the skull and tumor growth	P		
	What pain management medications have been helpful for chronic pain sufferers without causing too many side effects?	P		
	What new experimental drug therapy is out there,	P		
	OI and work-types to avoid and occ health advice for work and at home.	P		
	I take calcium and vit D, but recently developed tendonitis in my arm because a lump of calcium had formed in my shoulder joint, do I infact have too much calcium?	P		
	I was diagnosed with -2.2 osteoporosis what does this mean? What is the scale for osteoporosis severity measurement and what is it's interpretation?	P		
	To much worry about the benefits system.	P		
	When will KRN23 be done testing?	P		
	just wondering how long the studies for KRN23 will take before available to all.	P		
	I have Intravenous Zoledronate every 6 months but its unclear how effective this is in terms of slowing down FD tumour growth. Are better drugs/treatments being developed?	P		
	Who makes the decision whether I require more surgery?	P		
	What other help / treatment is availllable for FD?	P		
	At what point do doctors stop operating on the patients?	R		
	Is there any treatments for oi? Ie drug related I've been given vitamin d, calcium tablets which effected me and my mother giving us side effects as we both have been told we have it. But nothing else has been given or mentioned	R		
	Can the disease turn cancerous?	R		
	There is some confusion about inheriting this disease can it be made clear?	R		
	Known health issues, such as prone to fractures etc?	R		
	is there a resource which might be available (or prepared) which might be made available to non-specialist staff which outlines the symptoms and effects of XLH and whether XLH patients need to be given special treatment? For example, is there a fact sheet which might be given to (or be made available to) dentists, one for GPs or one for general hospital admissions?	R		
	Is it possible to safely diagnose OI and/or likely severity while in the womb?	R		
	How to help with pain management.	R		
	How to help him to be independent.	R		
	How to support MY SON when he's on a medication low while he is at school.	R		
	Do we need to have yearly reviews with Sch. Currently only have them at our local hospital.	R		
	when will kphos be easily available? I have to fight for it as opposed to Phosphate Sandoz	R		
	Changes to diagnosis and medication treatment over the years?	R		

	If my daughters have been treated with sandoz phosphate for their early childhood and new treatments such as the krn23 drug become widely available would it be better to move to an alternative treatment in late teens early adult hood. What would the implications of this change be?	R		
	What types of over the counter pain relief is acceptable to give on an on going basis to relieve pain? (for sporadic episodes only)	R		
	Whl left there are paediatric endocrinologists who are familiar with xlh and have experience wit it it is very difficult to fine similar for adults. Is there a register of suitably qualified and experienced UK / Ireland based adult doctors?	R		
	Is there a register of these [DENTISTS] to support with preventative on going care?	R		
	What alternative medications are available, and what are their side effects?	R		
	Are there any clinical trials taking place to look at collegen which is the deficiency in OI?	R		
	Can people with XLH be put on disability?	R		
	Why are insurance companies saying that the medicine K-Phos Neautral is a supplement when in my daughters case who has XLH, she literally HAS to be on it because of the phosphorus loss.	R		
	Are there any long term effects from years of xrays?	R		
	What is the incidence? of McCune Albright Syndrome.	R		
	What treatments are available, how effective are they,	T		
	Also, what support exists to support those afflicted.	T		
	The new drug x23 on trial in the US for xlh will it be available to try or trial in the uk and will it be used to treat Autosomal Dominant Hypophosphatemic rickets.	T		
	How often should you get bloods and urine checked when taking phosphate sandoz, alfacacidol and Theical D3. Should you be getting an ultra sound on your kidneys yearly to check for cacifcation even if your blood tests are fine??.	T		
	How often should blood tests and urine test be done when taking alficalcidol and phosphate sandoz and if your taking Theical D3 for osteopenia should you be getting checked more often?. when should you get your kidneys checked for calcification and should you be getting them checked even if your blood tests are coming back ok? Should you be getting other organs etc checked also for calcification when being prescribed the above drugs.	T		
	how often should babies/ toddlers be getting these tests done does it depend on age?	T		
	When getting a bone density scan done this year my t score in my hips was -2.278 and similar in my spine (osteopenia) should I be worried long term about breaks or fractures and will I get osteoporosis/osteomalacia even though I'm currently on treatment for almost a year, given that I'm only 36 I'm very concerned as my mother who is 67 is now wheelchair boundfrom having ADHR.	T		
	What treatments do you have for people with aggressive ADHR wheelchair bound, very crumbly bones and many fractures through the body. can the normal phosphate sandoz, alficalcidol dose help or is other supplements needed?	T		
Questions that are too broad or off-topic or unclear	? asfotase alfa treatment in adult hypophosphatasia (is there any evidence or RCTs ongoing or planned?)	H		
	Adult hypophosphatasia is a particular worry, particularly if they are offered antiresorptive therapies for low BMD in adult life.	H		
	Does asfotase alfa have a role in treating adults with Hypophosphatasia?	H		
	What is the difference in composition of XLH patient's teeth/enamel when compared with a non XLH person's teeth?	P	X	
	XLH patients seem to have bad teeth. Is this due to issues with the enamel? And does this arise from the same mechanism which affects bone formation?	R	X	
	It seems that younger XLH patients have poor bone formation. This would, intuitively, suggest that something is going wrong with calcium. If calcium is not processed correctly in younger patients, why is it that older XLH patients seem to suffer from calcification as they mature? In particular, it seems that there is calcification of ligaments and growth of spurs. As a non-medic, it seems that after calcification is not happening properly in younger XLH patients, it seems to switch in adulthood and calcification seems to work too well.	R	X	
	In general (but having in mind XLH patients), is it fully understood what links phosphates to bone formation? Why does lack of take-up of phosphates affect bone formation?	R	X	
	Is it fully understood what blocks the take-up of phosphates by XLH patients? Is there a specific process or mechanism which is not working properly?	R	X	
	Questions related to prevention and cure.	H		
	Lack of treatments in some diseases areas	H		
	Need more treatment options.	H		
	Not clear antifracture efficacy for new treatments in OI	H		
	How can we design trials for rare diseases?	H		
	No Treatment for Fibrous Dysplasia Ossificans Progressiva	H		
	Treatment of adults with X linked hypophosphataemia	H		
	Role of bisphosphonates in Osteogenesis Imperfecta in children (particularly in relation to dosage and effect of adult height)	H		
	How to improve access to genetic testing and genetic counselling?	H		
	Pharmacological interventions: Which drugs? For whome? Which dose? How long? Sequential drug interventions.	H		
	How can diagnosis making be improved in the future in situations where means of diagnosing are lacking today?	H		
	In FD there is no meaningful information on treatment	H		
	We do not have enough information about this as we do not have robust data on the prevalence of these diseases and so do not really have good evidence on how robust the diagnosis was	H		
	Optimal medications for patients with adult X linked Hypophosphataemic rickets	H		
	How did this disease occur?	P	F	
	Ongoing pain relief methods, ongoing research on adults.	P	O	
	Can they now detect OI in the womb. Can unborn babies be treated for OI in the womb.	P	O	
	All the above	P	O	
	Think the long tearm plan should be support the young ones n follow some till old age	P	O	
	I was diagnosed as a 4 year old but if in family the doctors should be made aware of it in pregnancy	P	O	
	Was diagnosed as a child	P	O	
	We are currently undergoing genetics testing. I was diagnosed due to my mother, grandmother and other relations' having it, but was only diagnosed after many bones being broken. I have a daughter who is 18 months and suspected ti have OI, so far no breaks, but we are hoping to get answers sooner rather than later so we can do the most possible to help her if she does have it....or let her lead a normal life if she doesn't.	P	O	
	Was diagnosed at birth	P	O	
	None as knew before i was born.	P	O	
	None was diagnosed at birth	P	O	
	My mother always said there was something wrong with me. Always sore, badly bruised, and hated and complained about walking. They paid 42 years ago to get me seen. And as I have said - inconclusive. I had a hateful pregnancy. Painful all the time. Son born by C:section in Jan 04. Agony. The day after I couldn't move. By Apr I was in constant pain with hospital saying it would settle down. Doctor finally sent me for X-ray. Result was fracture in spine. Doctor wanted me to see consultant (huge waiting list) I paid privately. Within 5 mins of sitting with him, he gave me my diagnosis and followed it up with genetics which confirmed it. 50/50 chance son would have it. I fought to get him tested. He came back clear. I was almost 36 when confirmed.	P	O	
	Does it work	P	O	
	Non had it since a baby .. so I live with it..	P	O	
	No questions. I have an endocrinologist that follows my meds and labs.	P	X	
	None. This has been in my family since my grandmother.	P	X	
	I have always exercised by playing sports, mainly hand to eye coordination ones like badminton, tennis and now table tennis as I have got older and have found it too painful to continue the first two as competitive sports.	P	X	
	I understand that XLH cannot be confirmed until you are about age 1 year old then via a blood test.	P	X	
	It took 14 years before my diagnosis, I had a CT and I then had a biopsy. Will there be more advanced ways of testing for FD in future?	P	F	
	It is a very obscure point for XLH regarding drugs to be maintained in the young and older adults.	P	X	
	How much closer are we getting to having access to a drug which can actually directly counteract the negative effects of the condition?	P	O	
	How likely is it that we could either see a full on cure to the condition in the future,	P	O	
	Diagnosed within a couple weeks of birth, I have no questions regarding this.	P	O	
	Would a more active medication regime help? Rather than checking my bloods and recording my symptoms?	P	X	
	Will this disease be worse if my children have it too?	P	X	
	what future treatments will be available	P	O	
	I was diagnosed as a child. My two children also have x-linked rickets. One male, one female. Also, one grandson.	P	X	
	new treatments investigacions about the treatments	P	X	
	Not applicable was diagnosed at birth	P	O	
	Skull injections that block nerves and pain relief gabapentin	P	F	
	Nothing not diagnosed yet	P	F	
	What is the best treatment for long term	P	F	

	Nothing. Been under the care of consultant for years, everything are very stable.	P	X	
	Been diagnosed since age 3.	P	X	
	I'm not entirely sure what it is for the main part except what I've been told by the clinical chemistry department	P	F	
	Why are my joints so bad too? My knees have been bad since I was about 25. Why did my finger joint tear, as well as my finger breaking... I only knocked it on a cupboard... I'm used to all the breaks, that's not new, but the joints getting bad is...Why am I constantly in pain? Why do I have kidney issues? And so many more health issues... I'm scared of what else is around the corner... would be good to know what to expect. Some days I can hardly move for pain, even though I have been taking 70 - 80 mg of Oxycodon, 1200 - 1800 mg Ibuprofen, every day since 2009. I took part in an experimental therapy over 4 years I recieved an IV of Aredia every 3 months, because from the age of about 25 my OI flared up again, and I started to break bones again regularly. That seemed to help with the breaking bones... but I have so many more health issues. Sometimes I just want everything to be over, I'm tired of it all. It's hard and painful and depressing.	P	O	
	Restricted lung function is common among those with scoliosis. Are there any exercises that can improve this?	P	O	
	New drug treatments	P	O	
	Diagnosis for children to definitively know if they have OI or not?	P	O	
	I had my rods fitted and are both causing great pain. The hospitals cant/wont do anything about it.	P	O	
	I was diagnosed at the age of 4 OR 5 more than 30 years ago. I am sure and hopeful that diagnosis is done a lot more sympathetic and with dignity now than it was then.	P	O	
	Do I have a long term obstipation that is not discovered?	P	O	
	Can you be diagnosed without having any fractures?	P	O	
	Are their future treatments that may help.	P	F	
	As Q3	P	O	
	If a person during pregnancy has a blood screening test performed at 9 - 12 weeks gestation and the test result shows a very low Pappa A level and a very high free beta BCG level, does this result mean the feotus has a chromosome abnormality such as Osteogenisis Imperfecta?	P	O	
	Why was it missed until I was 5	P	X	
	What care should I be receiving	P	X	
	What age should children be tested for XLH?	P	X	
	what might future treatments for me be?	P	O	
	I ultimately make most decisions upon my long term care by my diet, exercise and deciding whether or not to take any meds which I base on the little info out there which i know is slowly improving and ultimately everybody's treatment has to be so varied since XLH affects everybody to varying degrees and differently dependent upon the individuals other genetics (eg I have other bone conditions as well) but I do think I have to make most decisions on the little knowledge I have and make my own judgement s?	P	X	
	Mine was discovered when I was 3 years old so I now know there is a test but not sure how anyone would recognise signs in their child as little info about it advertised but doctors since to be more aware of it in last few years, dentists still struggling.	P	X	
	Im doing the trial with AXLES, Im interested to see if this is a short term or long term solution	P	X	
	How no one else in my known family history had XLH	P	X	
	If a baby is born with high phosphorus and alkaline phos. levels is that enough to diagnose XLH? (Mom has XLH).	P	X	
	Is there any hope of being able to identify eggs with/without XLH so women could chose to have an egg/s implanted to assure a baby without XLH?	P	X	
	If a mother is breastfeeding, how does any medication or treatment affect the breastmilk? Is it even beneficial to the baby to receive the breastmilk? Has breastmilk from an XLH affected mother ever been tested?	P	X	
	What about sclerostine in Mereo Laboratories?	P	O	
	menopause	P	O	
	I was told in my 20s I had OI as investigations went on with my small child who fractured her cheek at 6-7 months	P	O	
	If a mother (with xlh) has an amniotic fluid extraction and it's determined that the baby is a carrier, can treatment be started then?	P	X	
	None, I was diagnosed as a toddler and now have two daughters, one has it, one does not. Both were monitored and diagnosed positively or negatively by age one (although I could tell before then).	P	X	
	When will the trail drug be aviable to all? Is it as benifical to adults as children?	P	X	
	Why do we not have an alternative to Sandoz Phosphate as the side effects are quite severe.	P	X	
	Alternative medical treatments for patients with a poor response to 1st line phosphate and alfacalcidol treatment.	P	X	
	How to improve our quality of life	P	X	
	Chances of genetic screening preimplantation for XLH before conception	P	X	
	Been through diagnosis so no questions	P	X	
	How can we better address the delays in diagnosis for spontaneous cases of XLH, which often take until age 3 before being diagnosed.	P	X	
	Can it make my symptoms worse?	P	F	
	I have taken alternative medication for over twenty years , there's not enough knowledge of the disease. I am know not able to exercise or receive treatment other than swimming.	P	F	
	I am currently taking Fentanayl lyrica gaberpentin, colanapen, tamazapan, OxyContin. I have been on these medication for over two years. I fear my liver and kidneys may fail. I am waiting on medical canabis . I also received fosamax and stated loosing my teeth. I also need prescription medication to go to the bathroom.	P	F	
	I wish I had been diagnosed earlier in life. I would like one stop bone scan to list all effected areas. I have needed surgery for Fibrous areas in my uterus and in my breast . I was only diagnosed in my right hand and arm until my mammograms showed masses and tumors. I hemorrhaged for years from my Fibrous issue in my uterus.	P	F	
	Is there a way to NOT pass on XLH to children? Is there any treatment possible if is it just a coin flip (for women with XLH)	P	X	
	Drugs- fine balance between still being functional and taking the pain away.	P	O	
	If its known to be in the family , how it can be avoided in future generations	P	O	
	I was diagnosed when I was a baby. Not sure what to say here, guess it's something that I've lived with for a while.	P	O	
	None, I was diagnosed at birth because me dad had it.	P	X	
	Will there be other treatment options in the future?	P	X	
	Can an alternative tablet be produced the dissolvable tablet is just not practical? What happened to the other tablet that was available years ago? I used to take 6 tablets 4 times a day it was a lot but more practical then dissolving a tablet 4 times a day.	P	X	
	How is it done? osteogenisisimperfecta	R	O Type 5	
	What help can a patient in a less developed country get to sustain the operations lifestyle?	R	O	
	What causes the disease?	R	O	
	How we can manage it for our next generation	R	O	
	Will research hurt? Why hasn't there been any new drugs?	R	F	
	Will there ever be a cure or research for a cure?	R	F	
	Is there anything new and better? My understanding is that current treatment is ineffective	R	O	
	Getting my teeth fixed	R	O	
	My first daughter was diagnosed at 2.5 yrs as a spontaneous case. And after being speaking with a genetic counsellor we were told the chances of it happening again were in their 10000's. Our 2nd child doesnt have it and as a result we didn't test our 3rd child early for it (which we did on 2nd) .. and it turns out she has xlh also... following further genetic tests on me (mother) I don't have it in genes / blood/ urine howevery the tell me I 'must have it' in my gonodal reproduction cells.... no one ever mentions this as a possibility on the xlh network but if I have it 'silently' my fear is that my middle daughter may also have it the same way. While not an issue kow it may become so for her when she chose to have children herself. This is something that parents od spontaneous cases children should be made aware of	R	X	
	When to seek help	R	O	
	Why do we not have effective disease registries for these rare conditions?	H		