More detailed information to be shown on the JLA website for the questions discussed at the final workshop.

PSP Name	Total number of verified uncertainties identified by the PSP	Uncertainty (PICO formatted indicative uncertainty where possible. Advised minimum requirements are 'Population' and 'Intervention'. Not all submissions may be suitable for PICO structure, but they should be in a format that will ultimately be of value to the research community)	Explanatory note (a plain language summary of up to 150 words, explaining key points of the uncertainty and why it is important, for research funders to begin working on. PSPs may wish to include examples of the original survey submissions here)	Date of the priority setting workshop	Rank of the uncertainty at the final workshop. (If no rank was agreed, please indicate)	Evidence (reference, and weblink where available, to the most recent relevant systematic review identified by the PSP, plus a maximum of 2 other systematic reviews, including protocols for future systematic reviews, that the PSP considers relevant.)	Unanswered or partially answered?
		Could an understanding of the cellular and molecular processes in mitochondrial disease lead to new treatments?			1	https://www.ncbi.nlm.nih.gov/pubmed/30535772	**unanswered**
		Can the damage to cells caused by mitchondrial disease be repaired (e.g. to restore hearing, vision or repair the pancreas)?			2	No good evidence found	**unanswered**
		What are the biological mechanisms that cause mitochondrial disease to get worse over time?			3	No relevant evidence found (studies exist for secondary mithochondrial disorders, outside the scope of this PSP.	**unanswered**
		What biomarkers (biological markers that can be measured e.g. in blood samples) could be used to diagnose mitochondrial disease and to track its progress?			4	https://www.ncbi.nlm.nih.gov/pubmed/27794108 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5841218/ https://www.ncbi.nlm.nih.gov/pubmed/29735722	partially answered
		Could gene therapy help people with mitochondrial disease?			*5*		

		What are the psychological impacts of mitochondrial disease? What are the best ways to provide psychological support for people with mitochondrial disease and their families?			6	https://www.ncbi.nlm.nih.gov/pubmed/28476522 https://www.ncbi.nlm.nih.gov/pubmed/26856513 https://www.ncbi.nlm.nih.gov/pubmed/26391741 https://www.ncbi.nlm.nih.gov/pubmed/23529909 https://www.ncbi.nlm.nih.gov/pubmed/23398775 https://www.ncbi.nlm.nih.gov/pubmed/23430944 https://www.ncbi.nlm.nih.gov/pubmed/20573558 https://www.ncbi.nlm.nih.gov/pubmed/20304331 https://www.ncbi.nlm.nih.gov/pubmed/19397532	First part is partially answered. Second part is unanswered.
		What are the best ways to reduce the risk of stroke-like episodes in people with mitochondrial disease?			7		unanswered
		What factors could trigger the start of mitochondrial disease in people who have a genetic mutation?			8	https://www.ncbi.nlm.nih.gov/pubmed/19525327 https://www.ncbi.nlm.nih.gov/pubmed/26673666 https://www.ncbi.nlm.nih.gov/pubmed/21551238	partially answered
		Why are people with the same genetic mutation affected so differently in mitochondrial disease?			*9*	https://www.ncbi.nlm.nih.gov/pubmed/29880721 https://www.ncbi.nlm.nih.gov/pubmed/29560378 https://www.ncbi.nlm.nih.gov/pubmed/31253706 https://www.ncbi.nlm.nih.gov/pubmed/30393588	unanswered
		What are the most effective ways to treat and manage fatigue?			10	https://tinyurl.com/yxrxpk7m	unanswered
		What are the genetic mutations that cause mitochondrial disease and how do they cause it?			11	https://www.ncbi.nlm.nih.gov/pubmed/26404827 https://www.ncbi.nlm.nih.gov/pubmed/25652200 https://www.ncbi.nlm.nih.gov/pubmed/27659608 https://www.ncbi.nlm.nih.gov/pubmed/31171843 https://www.ncbi.nlm.nih.gov/pubmed/28415858 https://www.ncbi.nlm.nih.gov/pubmed/30374071	partially answered

		Could a specific diet and/or supplements benefit people with mitochondrial disease?			12	https://www.ncbi.nlm.nih.gov/pubmed/26782788 https://www.ncbi.nlm.nih.gov/pubmed/30024619 https://www.ncbi.nlm.nih.gov/pubmed/27444792 https://www.ncbi.nlm.nih.gov/pubmed/27665271 https://www.ncbi.nlm.nih.gov/pubmed/30407699	unanswered
		What can prevent mitochondrial disease from getting worse over time?			*13*		unanswered
		How do the different genetic mutations cause the symptoms people experience with mitochondrial disease?			14		unanswered
		Is there a way to predict who will become ill with mitochondrial disease, and whose symptoms will be worse?			15		partially answered
		What can prevent the start of mitochondrial disease in people with a genetic mutation?			16	https://www.ncbi.nlm.nih.gov/pubmed/31091381 https://www.ncbi.nlm.nih.gov/pubmed/30319102 https://www.ncbi.nlm.nih.gov/pubmed/29950320 https://www.ncbi.nlm.nih.gov/pubmed/28415858	unanswered
		What are the most effective ways to treat and manage problems with muscle weakness?			17	https://www.ncbi.nlm.nih.gov/pubmed/30710167 https://www.ncbi.nlm.nih.gov/pubmed/28179228 https://www.ncbi.nlm.nih.gov/pubmed/30406383	unanswered
		What aspects of their health should be monitored over time and how often in people with mitochondrial disease?			18	https://www.ncbi.nlm.nih.gov/pubmed/28749475 https://www.ncbi.nlm.nih.gov/pubmed/31138493 https://www.ncbi.nlm.nih.gov/pubmed/31083569 https://www.ncbi.nlm.nih.gov/pubmed/28647693	partially answered
		What are the most effective ways to treat and manage problems with memory, concentrating, learning and making decisions?			19		unanswered
		What are the most effective ways to treat and manage pain?			20		unanswered

		What are the most effective ways to treat and manage problems with balance and co-ordination?			*21*	https://www.ncbi.nlm.nih.gov/pubmed/29891055_ https://www.ncbi.nlm.nih.gov/pubmed/29307008 https://www.cochranelibrary.com/cdsr/doi/10.1002/14651858.CD005397.pub4/abstract	partially answered
		How does mitochondrial disease change over time as people get older?			22		unanswered
		Does exercise benefit people with mitochondrial disease? If yes, what is the best form of exercise?			23	https://www.ncbi.nlm.nih.gov/pubmed/23742928 https://www.ncbi.nlm.nih.gov/pubmed/25008908 https://www.ncbi.nlm.nih.gov/pubmed/23835682 https://www.ncbi.nlm.nih.gov/pubmed/31105594	partially answered
		What causes the genetic mutation in people with mitochondrial disease whose parents don't have the mutation?			24	https://www.ncbi.nlm.nih.gov/pubmed/27659608_ https://www.ncbi.nlm.nih.gov/pubmed/26315846_ https://www.ncbi.nlm.nih.gov/pubmed/26404827	partially answered

Data management spreadsheet for use by Priority Setting Partnerships for all questions received. Spreadsheet to be published on the JLA website at www.jla.nihr.ac.uk on completion of the PSP.

ID	Uncertainty (PICO formatted indicative uncertainty where possible. Advised minimum requirements are 'Population' and 'Intervention'. Not all submissions may be suitable for PICO structure, but they should be in a format that will ultimately be of value to the research community)	Original questions submitted to survey: these are listed in full on the sheet "Individual questions submitted".	Evidence (most relevant evidence found)	Source of Uncertainty (if there are multiple sources, a PSP may wish to show them e.g. 1 x patient, 19 x clinician, 4 x research recommendations)	Unanswered or partially answered?	Notes
1	Could an understanding of the cellular and molecular processes in mitochondrial disease lead to new treatments?	See cells D695-696	https://www.ncbi.nlm.nih.gov/pubmed/30535772	HCP	unanswered	
2	Can the damage to cells caused by mitchondrial disease be repaired (e.g. to restore hearing, vision or repair the pancreas)?	See cells D681-684	No good evidence found	2 x P, 1 x C, 1 x HCP	unanswered	
3	What are the biological mechanisms that cause mitochondrial disease to get worse over time?	See cells D19-24	No relevant evidence found (studies exist for secondary mithochondrial disorders, outside the scope of this PSP.	5 x HPC, 1 x P, 1 x C	unanswered	
4	What biomarkers (biological markers that can be measured e.g. in blood samples) could be used to diagnose mitochondrial disease and to track its progress?	See cells D25-32	https://www.ncbi.nlm.nih.gov/pubmed/27794108 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5841218/ https://www.ncbi.nlm.nih.gov/pubmed/29735722	6 s HPCs, 2 x C	partially answered	
5	Could gene therapy help people with mitochondrial disease?	See cells D640-652				
6	What are the psychological impacts of mitochondrial disease? What are the best ways to provide psychological support for people with mitochondrial disease and their families?	See cells D697-715	https://www.ncbi.nlm.nih.gov/pubmed/28476522 https://www.ncbi.nlm.nih.gov/pubmed/26856513 https://www.ncbi.nlm.nih.gov/pubmed/26391741 https://www.ncbi.nlm.nih.gov/pubmed/23529909 https://www.ncbi.nlm.nih.gov/pubmed/23398775 https://www.ncbi.nlm.nih.gov/pubmed/23430944 https://www.ncbi.nlm.nih.gov/pubmed/20573558 https://www.ncbi.nlm.nih.gov/pubmed/20304331 https://www.ncbi.nlm.nih.gov/pubmed/19397532	6 x P, 6 xC, 6 x H	First part is partially answered. Second part is unanswered.	
7	What are the best ways to reduce the risk of stroke-like episodes in people with mitochondrial disease?	See cell D553			unanswered	
8	What factors could trigger the start of mitochondrial disease in people who have a genetic mutation?	See cells D65-77	https://www.ncbi.nlm.nih.gov/pubmed/19525327 https://www.ncbi.nlm.nih.gov/pubmed/26673666 https://www.ncbi.nlm.nih.gov/pubmed/21551238	8 x C, 3 x P, 2 x HCPs	partially answered	
9	Why are people with the same genetic mutation affected so differently in mitochondrial disease?	See cells D8-18	https://www.ncbi.nlm.nih.gov/pubmed/29880721 https://www.ncbi.nlm.nih.gov/pubmed/29560378 https://www.ncbi.nlm.nih.gov/pubmed/31253706 https://www.ncbi.nlm.nih.gov/pubmed/30393588	6 x HCPs, 3 x C, 1 x P	unanswered	
10	What are the most effective ways to treat and manage fatigue?	See cells D595-609	https://tinyurl.com/yxrxpk7m	6 x P, 5 x HCPs, 1 x C	unanswered	

11	What are the genetic mutations that cause mitochondrial disease and how do they cause it?	See cells D1-7	https://www.ncbi.nlm.nih.gov/pubmed/26404827 https://www.ncbi.nlm.nih.gov/pubmed/25652200 https://www.ncbi.nlm.nih.gov/pubmed/27659608 https://www.ncbi.nlm.nih.gov/pubmed/31171843 https://www.ncbi.nlm.nih.gov/pubmed/28415858 https://www.ncbi.nlm.nih.gov/pubmed/30374071	4xHCPs, 2xC, 1xP	partially answered	
12	Could a specific diet and/or supplements benefit people with mitochondrial disease?	See cells D188-233	https://www.ncbi.nlm.nih.gov/pubmed/26782788 https://www.ncbi.nlm.nih.gov/pubmed/30024619 https://www.ncbi.nlm.nih.gov/pubmed/27444792 https://www.ncbi.nlm.nih.gov/pubmed/27665271 https://www.ncbi.nlm.nih.gov/pubmed/30407699	22xP, 14xC, 10xHCPs	unanswered	
13	What can prevent mitochondrial disease from getting worse over time?	See cells D567-594		14xC, 7xHCPs, 7xP	unanswered	
14	How do the different genetic mutations cause the symptoms people experience with mitochondrial disease?	See cells D33-57		19xHCP, 6cC	unanswered	
15	Is there a way to predict who will become ill with mitochondrial disease, and whose symptoms will be worse?	See cells D533-538		3xHCP, 2xP, 1xC	partially answered	
16	What can prevent the start of mitochondrial disease in people with a genetic mutation?	See cells D557-566	https://www.ncbi.nlm.nih.gov/pubmed/31091381 https://www.ncbi.nlm.nih.gov/pubmed/30319102 https://www.ncbi.nlm.nih.gov/pubmed/29950320 https://www.ncbi.nlm.nih.gov/pubmed/28415858	6xHCP, 3xC, 1xP	unanswered	
17	What are the most effective ways to treat and manage problems with muscle weakness?	See cells D635-636	https://www.ncbi.nlm.nih.gov/pubmed/30710167 https://www.ncbi.nlm.nih.gov/pubmed/28179228 https://www.ncbi.nlm.nih.gov/pubmed/30406383	1xP, 1xH	unanswered	
18	What aspects of their health should be monitored over time and how often in people with mitochondrial disease?	See cells D127-147	https://www.ncbi.nlm.nih.gov/pubmed/28749475 https://www.ncbi.nlm.nih.gov/pubmed/31138493 https://www.ncbi.nlm.nih.gov/pubmed/31083569 https://www.ncbi.nlm.nih.gov/pubmed/28647693	10xP, 7xC, 4xHCP	partially answered	
19	What are the most effective ways to treat and manage problems with memory, concentrating, learning and making decisions?	See cells D618-620		1xP, 1xC, 1xHCP	unanswered	
20	What are the most effective ways to treat and manage pain?	See cells D610-617		4xHCP, 3xP, 1xC	unanswered	
21	What are the most effective ways to treat and manage problems with balance and co-ordination?	See cells D621-625	https://www.ncbi.nlm.nih.gov/pubmed/29891055 https://www.ncbi.nlm.nih.gov/pubmed/29307008 https://www.cochranelibrary.com/cdsr/doi/10.1002/14651858.CD005397.pub4/abstract	3xP, 1xH, 1xC	partially answered	
22	How does mitochondrial disease change over time as people get older?	See cells D479-532		30xP, 13xC, 11xHCP	unanswered	
23	Does exercise benefit people with mitochondrial disease? If yes, what is the best form of exercise?	See cells D171-187	https://www.ncbi.nlm.nih.gov/pubmed/23742928 https://www.ncbi.nlm.nih.gov/pubmed/25008908 https://www.ncbi.nlm.nih.gov/pubmed/23835682 https://www.ncbi.nlm.nih.gov/pubmed/31105594	8xP, 6xHCP, 3xC	partially answered	
24	What causes the genetic mutation in people with mitochondrial disease whose parents don't have the mutation?	See cells D58-64	https://www.ncbi.nlm.nih.gov/pubmed/27659608 https://www.ncbi.nlm.nih.gov/pubmed/26315846 https://www.ncbi.nlm.nih.gov/pubmed/26404827	5xP, 2xC	partially answered	
unranked	What is the risk of children inheriting mitochondrial disease from their parents?	See cells D545-547	https://www.ncbi.nlm.nih.gov/pubmed/31091381 https://www.ncbi.nlm.nih.gov/pubmed/30319102 https://www.ncbi.nlm.nih.gov/pubmed/29950320 https://www.ncbi.nlm.nih.gov/pubmed/28415858	2xHCP, 1xP	partially answered	

unranked	What are the most effective ways to treat and manage problems with eating and digestion?	See cells D626-634	https://www.ncbi.nlm.nih.gov/pubmed/22283595 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4101992/ https://www.ncbi.nlm.nih.gov/pubmed/28347206	5xC, 4xHCP	unanswered	
unranked	What lifestyle changes benefit people with mitochondrial disease (e.g. reducing stress, changing jobs, saunas)?	See cells D234-243			unanswered	
unranked	What can be learnt about managing the condition from people who successfully cope with mitochondrial disease?	See cells D244-252	https://www.ncbi.nlm.nih.gov/pubmed/30424784 https://www.ncbi.nlm.nih.gov/pubmed/26988355	4xP, 3xHCP, 2xC	unanswered	
unranked	Why do some treatments work well for some people but not others? Does treatment need to be tailored to specific genetic mutations in mitochondrial disease?	See cells D672-680	https://www.ncbi.nlm.nih.gov/pubmed/30459337	5xHCP, 3xC, 1xP	unanswered	
unranked	Should the treatment of common conditions (e.g. diabetes and heart disease) be different in people with mitochondrial disease?	See cells D685-694	https://www.ncbi.nlm.nih.gov/pubmed/30884027 https://www.ncbi.nlm.nih.gov/pubmed/30326976 https://www.ncbi.nlm.nih.gov/pubmed/26518446 https://www.ncbi.nlm.nih.gov/pubmed/25330715 https://www.ncbi.nlm.nih.gov/pubmed/22936362 https://www.ncbi.nlm.nih.gov/pubmed/27453452	5xHCP, 3xP, 2xC	partially answered	
unranked	Would cannabinoid oil benefit people with mitochondrial disease?	See cells D665-667		2xP, 1xC	unanswered	
unranked	How is life expectancy affected by mitochondrial disease?	See cells D541-542	https://www.ncbi.nlm.nih.gov/pubmed/23355809	1xP, 1xC, 1xHCP	partially answered	
unranked	What is the best end of life care for people with mitochondrial disease?	See cells D168-170	https://www.ncbi.nlm.nih.gov/pubmed/29161160	2xHCP, 1xC	unanswered	
unranked	Do alternative therapies benefit people with mitochondrial disease (e.g. acupuncture, massage, reflexology)?	See cells D653-664		8xP, 3x, 1xHCP	unanswered	
unranked	Would physiotherapy benefit people with mitochondrial disease?	See cells D668-671	https://www.ncbi.nlm.nih.gov/pubmed/29980632 https://www.ncbi.nlm.nih.gov/pubmed/25503498 https://www.ncbi.nlm.nih.gov/pubmed/23835682 https://www.ncbi.nlm.nih.gov/pubmed/25008908 https://www.ncbi.nlm.nih.gov/pubmed/17085458 https://www.ncbi.nlm.nih.gov/pubmed/11506394	2xP, 1xC, 1xHCP	partially answered	
unranked	Does deep brain stimulation benefit people with mitochondrial disease?	See cell D555	https://www.ncbi.nlm.nih.gov/pubmed/30713906 https://www.ncbi.nlm.nih.gov/pubmed/17960792	1xC	unanswered	This was a single-ask question but within scope
unranked	Is there a way to provide long-lasting treatment for mitochondrial disease that avoids having to take tablets (e.g. idebenone) daily?	See cell D639		1xH	unanswered	This was a single-ask question but within scope
unranked	Are males and females differently affected by mitochondrial disease?	See cells D543-544	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4032517/	1xC, 1xHCP	partially answered	
unranked	Are people with mitochondrial disease at greater risk of cancer?	See cells D539-540	https://www.ncbi.nlm.nih.gov/pubmed/25742477 https://www.ncbi.nlm.nih.gov/pubmed/24868266 https://www.ncbi.nlm.nih.gov/pubmed/27181047	1xP, 1xC	unanswered	
unranked	Are people with mitochondrial disease at greater risk from surgery and anaesthetic?	See cells D548-550		2xC, 1xP	unanswered	
unranked	How effective is levocarnitine as a treatment for people with carnitine deficiency?	See cell D637	https://www.ncbi.nlm.nih.gov/pubmed/234182	1xH	Unanswered for secondary carnitine deficiency in primary mitochondrial disease.	This was a single-ask question but within scope

unranked	What are the risks of starving overnight e.g. before surgery or if unwell?	See cells D551-552	https://www.ncbi.nlm.nih.gov/pubmed/27896927_ https://www.ncbi.nlm.nih.gov/pubmed/24741716_ https://www.ncbi.nlm.nih.gov/pubmed/23534340	2xC	partially answered	

1.06E+10	4	4.1	How does the genetic mutation lead to disease?		B1	H
1.06E+10	6	6.1	Where did 3243a^g originally come from?		B1	P
1.06E+10	28	28.1	Interested in the genetics of the condition,		B1	CS
1.06E+10	47	47.1	Ongoing research into genetic causes of mitochondrial ocular disease with/out syndromic features		B1	H
		85.8	What are all the genetic causes of mitochondrial disease?		B1	H
1.07E+10	121	121.1	The role that defective fatty acid synthesis plays as an underlying cause of mitochondrial disease		B1	CS
1.07E+10	131	131.1	Progress with identifying the genetic basis is particularly important for severe paediatric onset diseases with no effective treatment as it allows prenatal diagnosis (or PGD) to be offered.		B1	H
		13.2	How can family members with the same condition present at different ages with different symptoms?		B2	P
1.06E+10	22	22.1	Why do some members of a family get MELAS and others don't. I carry 20%, my son passed away at [age] from it but my sister and my daughter don't appear to have any down to 1% that can be tested.		B2	CS
1.06E+10	40	40.1	Why do similar mutations in mtDNA give rise to such a diversity of phenotypes?		B2	H
1.06E+10	56	56.1	What is the importance of heteroplasmy in the brain with respect to CNS phenotypes? Is there regional heteroplasmy within CNS tissue?		B2	H
1.06E+10	72	72.1	I would like more research into the effects on carriers of the disease.		B2	CS
1.06E+10	81	81.1	How does heteroplasmy vary between tissues and cells and what are the mechanisms involved in these changes over time?		B2	H
		96.2	Why does the heteroplasmy in some gene mutations end up being split between either high or low levels whilst middle levels are a lot more rare?		B2	CS
1.07E+10	97	97.1	Why one mutation can result in different symptoms in different individuals.		B2	H
1.07E+10	103	103.1	Genetic and epigenetic profiling.		B2	H
		113.2	Role of nuclear modifying genes on mitochondrial DNA		B2	H
1.07E+10	117	117.1	It would be really good to know a little more about what genes influence and modify mitochondrial DNA mutations, causing such huge variations in phenotype and progression between individuals with the same genetic cause.		B2	H
1.06E+10	70	70.1	What factors cause progression in 3243a)g mutation?		B3	P
		81.2	What are the mechanisms of disease progression?		B3	H
1.07E+10	106	106.1	what determines progression?		B3	H
		117.10	It would be really good to know a little more about what genes influence and modify mitochondrial DNA mutations, causing such huge variations in phenotype and progression between individuals with the same genetic cause.		B3	H
1.07E+10	127	127.1	We understand very little about the mechanisms of disease progression in patients with mitochondrial disease. This is a vital area of research because if we understood more about the factors involved in progression then we may be able to devise better treatments		B3	H

1.07E+10	129	129.1	To understand more clearly the role heteroplasmy in disease progression of mtDNA diseases.		B3	H
		10.5	A way to monitor progression.		B4	CS
		10.6	Maybe research could Show what the best blood tests would be needed to spot progression or health changes.		B4	CS
1.06E+10	58	58.1	Progression : identification of bio markers of progressive disease		B4	H
		100.3	Are there biomarkers (e.g. on retinal imaging) for progression of mitochondrial retinopathy?		B4	H
		111.3	Pathognomic features or biomerkers for early detection of the disease.		B4	H
		136.2	Research on diagnostic markers		B4	H
1.07E+10	137	137.1	Biomarkers.		B4	H
		139.2	studies designed to identify prognostic biomarkers and trial endpoints		B4	H
		4.3	Why do retinal ganglion cells die in Leber's hereditary optic neuropathy?		B5	H
		22.5	I carry only 20% and have been told I wouldn't suffer symptoms but I feel I do, muscle fatigue and tiredness, is this true ?		B5	CS
		31.4	Which other systems are most likely to be affected and how?		B5	CS
		37.2	Like to understand low thyroid T3 connection is there a thyroid and T3 connection to the disease		B5	CS
		40.6	How do genetic mutations in POLG cause epilepsy? What determines phenotypic presentation of mitochondrial disease?		B5	H
		56.6	Why does 3243 cause MELAS in some people and MIDD in others?		B5	H
		56.7	What is the role of common variants outside of mito genes in the phenotypic expression?		B5	H
1.06E+10	73	73.1	How the Percentage of mutation affects the chances of having severe side affects		B5	CS
1.06E+10	80	80.1	What causes dysphagia in mitochondrial disease? How does it alter in different genetic diagnosis?		B5	H
1.07E+10	85	85.1	Is mitochondrial disease caused by different genes different in character - ie is there any gene-phenotype correlation?		B5	H
		85.2	Why are some organs affected in some patients and other organs in others? ie what are the reasons for tissue specificity of mitochondrial disease?		B5	H
1.06E+10	98	98.1	Genotype phenotype correlation Twin studies		B5	H
1.07E+10	100	100.1	What determines the development of mitochondrial retinopathy? Which retinal cells primarily undergo degeneration?		B5	H
		100.5	What determines different phenotypes (not extent) of mitochondrial retinopathy?		B5	H
1.07E+10	105	105.1	Why are retinal ganglion cells preferentially affected by Leber's Hereditary Optic Neuropathy (LHON) when the mutation is present in every cell in the body?		B5	CS
		111.2	Phenotype and genotype correlation		B5	H
1.07E+10	113	113.1	Genotype/phenotype relationship		B5	H
		117.2	It would also be really helpful to be able to better understand more about genetic mutations and how they manifest, in order to predict disease progression		B5	H

		122.2	What are the non visual affects of inherited mitochondrial optic neuropathy e.g. sleep, mood etc.		B5	H
1.07E+10	124	124.1	What factors influence the phenotype presenting within a family? Useful for genetic counselling about risk within a family & how to screen		B5	H
1.07E+10	126	126.1	genotype-phenotype correlation.		B5	H
1.07E+10	128	128.1	I would like to gain further insights into the molecular mechanisms by which mitochondrial dysfunction contributes to selective retinal ganglion degeneration in heritable optic neuropathies. In particular, I would be keen to know how mitochondrial interorganelle dysfunction contributes to this.		B5	H
1.07E+10	135	135.1	Correlation between phenotype and genotype,		B5	H
		144.2	Why (in LHON) is the optic nerve the only area damaged when it is not the only part of the body with high energy demand?		B5	CS
additional 2	147	147.1	What is the mechanism for causing GI dysmotility in mitochondrial disease?		B5	H
1.06E+10	13	13.1	What causes the mutation? Can the mutation occur at any time or is it historical?		C1	P
1.06E+10	52	52.1	what causes sporadic DNA mutations? why do they happen? are you sure that they are sporadice? how do you know?		C1	P
1.06E+10	60	60.1	In [name]'s case her disease is spontaneous rather than genetic, more research into why it happened.		C1	CS
1.06E+10	76	76.1	Why are there so many different mutations of the disorder. What triggers the mutations? Why do the mitochondria mutate?		C1	P
1.07E+10	83	83.1	What would have caused my mothers gene to mutate to cause the disease.		C1	P
1.07E+10	90	90.1	Is the fault on the gene a spontaneous mutation?		C1	CS
1.07E+10	94	94.1	I didn’t inherit this disease. So what caused it?		C1	P
1.06E+10	10	10.1	What are the triggers?		C2	CS
1.06E+10	33	33.1	What caused the disease to start in my teens,		C2	P
1.06E+10	39	39.1	Why does it take many years for diseases such as Leber's and MELAS to present yet the gene defect is there from conception. Could lead to therapeutic approaches.		C2	H
1.07E+10	96	96.1	In a mitochondrial DNA mutation can carrying a very ill child (99%) change the mother’s DNA mutation? This feels like a silly question when I say it out loud but I have a 65% ATP6 mutation and I can’t help feeling physically worse since having (and losing) my daughter.		C2	CS
		96.3	Is vitamin B12 deficiency linked to mitochondrial disease? I have been diagnosed with this recently and asked about any experiences of this in the lily mito family group and this seems to be a common connection.		C2	CS
1.06E+10	99	99.1	In LHON: 1. Why is vision lost suddenly years or decades after carrying an asymptomatic mDNA mutation? 2. Why is the second eye affected just days/weeks/months later? 3. What are the epigenetic and environmental factors that explain the low penetrance of this condition? (in other words, why doesn't everyone with the mutation lose sight?)		C2	H
1.07E+10	104	104.1	Would like to know the triggers for lhon but know this is not conclusive at present		C2	CS

		105.3	What triggers the catastrophic sudden death of retinal ganglion cells?		C2	CS
1.07E+10	107	107.1	How the disease can start		C2	P
1.07E+10	141	141.1	Can other / 'unrelated' conditions trigger the onset of my disease? How/ why?		C2	P
1.07E+10	142	142.1	Why do some people appear to get LHON even though nobody has previously been diagnosed in their family?		C2	CS
1.07E+10	144	144.1	What activates the mutation to cause carriers to move to being affected? Is it an action on the mutation itself or was it previously activated but the mitochondrial damage reaches a "tipping point."		C2	CS
		145.2	What are the contributory factors?		C2	CS
1.06E+10	29	29.1	How fast can the diagnosis can be turned around?		D1	H
		6.2	Is it possible to have a personalised care sheet for A&E attendance to hospital stays etc	P&P	OOS	P
		7.2	What do generalist health professionals need to know/what support do they need to care for "once in a career" conditions	P&P	OOS	H
		10.2	Emergency protocols with the best way to treat. In general more health profesionals need info as not well educated enough.	P&P	OOS	CS
		11.5	Better information network for other medical professionals especially g.ps.	P&P	OOS	CS
		12.5	More effective methods of informing GPs and other health professionals about the disease. I find knowledge level varies considerably. Ways of improving access to GPs would be helpful	P&P	OOS	P
		13.3	Treatment sheet for gp if a patient has an infection Treatment sheet for any medical or allied health professional (physio, dietician OT) about the condition. Treatment sheet for A&E.	P&P	OOS	P
		14.2	To improve the knowledge of local GPs about mito disease. To ensure they and patients have copies of emergency information sheets.	P&P	OOS	P
		17.5	info sheets for who? family, friends, hospital doctors?	P&P	OOS	P
		22.2	It's imperative that some sort of care sheet/ info sheet is available as not even drs understand what to do or how to treat.	P&P	OOS	CS
		23.5	GPs and other doctors are ill informed.	P&P	OOS	P
		31.5	How can we help the professionals who monitor our daughter's sight, hearing and heart understand more about her condition and what to look for?	P&P	OOS	CS
		35.2	What to do in an emergency like days of endless vomiting cause going through a and e and explaining everything to several drs and telling them u simply need anti sickness and a drip they still want to further investigate even though I've been there several times before isn't there some kind of plan that can be put in place to avoid this	P&P	OOS	P
		36.2	Health care professionals such as physiotherapists being made more aware.	P&P	OOS	CS
		36.4	understanding in medical community of conditions even at GP level. Understanding of progression by external professionals not hospital care.	P&P	OOS	CS

		40.2	Are health professionals accessing available sources of information in addressing healthcare needs of mitochondrial disease patients?	P&P	OOS	H
		49.2	Being near to the [name] Hospital [a specialist mitochondrial centre] I have no urgent questions about care, However there are very few medics or dentists around, who know either what MELAS or Rare Mitochondrial Disease is.	P&P	OOS	P
		49.3	Since there is no treatment, I would obviously like a breakthrough here. Knowledge about connections between mitochondrial disease and nerve damage seems relatively woolly. For years I have been gradually going down with trigeminal neuralgia, I can now say, with facial pain especially in the teeth, but nobody asked about that area, and I though it was a dental problem. Prof. [name] recommended duloxetine, which is an excellent treatment that would have saved me much pain long ago.	P&P	OOS	P
		52.5	do local GPs have the knowledge to provide care locally? are GPs / general hospitals active in seeking advice from specialist centres?	P&P	OOS	P
		54.2	emergency info sheet would help, going to hospital terrifies me	P&P	OOS	P
		60.5	Also physiotherapy related to mito I feel the physios don't understand mito	P&P	OOS	CS
		60.6	also an emergency information sheet would be an excellent idea	P&P	OOS	CS
		62.2	Em info sheet sounds brill idea, we have annual heart checks	P&P	OOS	P
		65.2	i love a information sheet as a lot of gps and other that help with my son do not know what it is	P&P	OOS	CS
		67.2	For my local GP to understand it better	P&P	OOS	P
		69.3	Patient emergency and hospital sheet definetly	P&P	OOS	P
		70.3	Is there an NHS online alert system in place, when a mito patient is admitted to any UK hospital? Is there an info sheet for patients admitted for surgery? Are there training days or online seminars to update GP's?	P&P	OOS	P
1.06E+10	71	71.1	Sheets you can give to local hospital if I am needing care.	P&P	OOS	P
		73.2	How to explain to other health professionals who have no or very little knowledge about the condition	P&P	OOS	CS
1.06E+10	74	74.1	Diagnosis. So many GP's are unaware of symptoms relating to MD diseases. It took me 8 years to get a diagnosis, fobbed off with all kinds of excuses such as being unfit, depression and even told on one occasion, where an MRI showed some degree of muscle wastage, that it wasn't muscle wastage, it was me, moving while in the machine that affected the scan results. Three years after that, I decided to pay to see a private consultant, I was diagnosed within 3 months and sent for genetic screening to confirm it was Myofibrilar Myopathy. More medical training is needed for medical professionals to prevent others going through the stress of getting a diagnosis.	P&P	OOS	P

		74.2	I was admitted to A&E last year, following a fall. None of the medical team on duty would listen to me, I told them all about my condition, but they almost killed me, because they didn't seem to believe what I was telling them. I asked them to contact someone from my medial team, gave them the numbers, but not one member of staff called my medical contacts. Due to lack of wheelchairs in the department, I was told unless I could walk to the toilet, I would have to wear a nappy during my stay. I was denied a bipap machine and offered oxygen, which led to complications. There was no hoist to transfer to the x ray table, therefore I went without an x ray, despite the doctor suspecting id broken my back. Medical professionals need some training, or told to listen to us, we live with this every day, therefore we are the experts. Many cases iv heard, medical teams do not check our care plans or medical records on admittance to A&E, which puts our lives at risk.	P&P	OOS	P
		76.2	People with mito should be given a card with their specific issues on it to show to authorities and other professionals. It would be helpful to have appointment on the same day, to lessen disruption to patient lives. Also having more registrars involved in the care to lessen waiting times.	P&P	OOS	P
		78.2	In my personal experience there is a lack of care in the community in all aspects of care with the mitochondrial disease I have. The reply I get when asked what I have is "never heard of it"!	P&P	OOS	P
		90.3	How can knowledge and understanding of the disease be made more widely known about amongst other professionals, both health and care?	P&P	OOS	CS
		91.3	Emergency information sheets would be useful and widening the understanding of GP's, psychiatrists, physios and OT's about the disease.	P&P	OOS	P
		94.13	Having a digital resource, as well as something physical, to explain the disease, symptoms etc, for healthcare professionals, public sector workers, family and friends etc.	P&P	OOS	P
		95.2	Gp to be more aware of conditions	P&P	OOS	CS
		95.4	When I have spoken to gp re mitrchondrial disease they have very little knowledge and dismiss as making you feel like a hypochondriac	P&P	OOS	CS
		96.5	What are health professionals knowledge of mitochondrial disease in the more peripheral professions e.g. anaesthetists, MRI specialists. My daughter had to wait a very long time with nil by mouth on the day of her MRI scan. Although this was at the diagnostics stage so it was not known she had mito I think it was already suspected.	P&P	OOS	CS
		101.2	an issue that is frequently discussed on our adult support group is the lack of understanding of mitochondrial disease by some medical professionals, including GPs and doctors following emergency admission to hospital. However, I'm not sure if it is possible to improve understanding of mitochondrial disease amongst medical professionals through research?	P&P	OOS	CS

		106.2	creation of emergency alert card	P&P	OOS	H
		109.2	Information about acute events and management that a patient can carry with them would be very helpful, patient held management plans.	P&P	OOS	H
		109.3	Clear information for GPs about medication that may help , focussing on evidence of harm as patients able toaccept that it may not help but GPs do not want to cause harm by prescribing	P&P	OOS	H
		110.3	Clearer mandatory training about the condition, so that they can go to a dedicated information source with information sheets and relevant information	P&P	OOS	CC
		110.8	Clearer information with understanding! All to often specialist consultants with little understanding of rare conditions can authorise or advise treatments which cause injury or medications which cause health issue to worsen	P&P	OOS	CC
		121.2	More knowledge in the medical community about mitochondrial health	P&P	OOS	CS
		140.2	We have been to optometrists who'd never heard of ADOA. Maybe make information more available at high street shop level.	P&P	OOS	CS
		141.3	Yes, the provision of information sheet/ online faq’s and answers, but also dedicated contact with specialized knowledge of LHON?	P&P	OOS	P
		144.3	Why are health authorities disinterested in monitoring this condition once people become affected and effectively feel abandoned and ignored by the health service?	P&P	OOS	CS
		12.2	How my care can keep in line with the progression of the disease and take note of new developments		H2	P
		13.4	If epilepsy(or diabetes) is a known symptom of the condition-should base line investigations be carried out at diagnosis		H2	P
		13.8	When to have hearing/eye examinations.		H2	P
		15.2	how often should people have heart checks?		H2	CS
		16.2	Why isn't care more proactive rather than reactive. My son has been in emergency life threatening situations which have subsequently been managed by interventions such as tracheostomy & gastrostomy		H2	CS
		17.2	have annual heart checks		H2	P
		20.2	What do you do if you’re having a ‘mitochondrial episode’ more information on mitochondria- like checks for diabetes, kidney.		H2	P
		21.2	Would a checklist of procedures be helpful? E.g when to have blood tests and which ones, when is my cardiology check due etc		H2	P
		27.2	How often should be seen?		H2	P
		36.3	Longer term progressive medicine to try and keep up with the symptoms. Medical equipment keeping pace with disease.		H2	CS
		50.3	Is once a year enough for check up ? What other general checks yearly do they need ie bloods scans etc		H2	CS
		60.3	We have been very lucky with our doctors but as [name] is now [age] and it is about to go to adult care I'm feeling very apprehensive, I think there should be a checklist like heart scans, hearing tests blood tests etc , so I can then follow it up		H2	CS
		69.2	Heart checks and others.		H2	P

		84.2	An agreed protocol of clinical monitoring.		H2	H
		87.3	National (and international) guidelines/recommendations for cardiac screening		H2	H
		88.2	Shoud heart checks be done on a regular basis as my daughters heart rate goes up and down ..		H2	CS
		94.4	Continued and frequent monitoring by consultants, such as the consultants and professors who work with mitochondrial disease, cardiologists, breathing and lung tests, neurologists, ophthalmologists etc. Continued testing and care to track the progression, and to ensure that anything else that may develop, will be caught quickly and managed.		H2	P
		114.6	screening for potential complications to treat them early		H2	H
		124.2	What screening is appropriate for particular patient groups? E.g. A1555G Should percentage mutation load influence what screening an individual has?		H2	H
		142.3	How regularly should eye tests be conducted after diagnosis i.e. intraocular pressures, health of the back of the eye etc? Should LHON sufferers have annual health checks/blood tests to check for the possible onset of secondary issues?		H2	CS
		143.3	Regular reviews for hearing, eyes, heart, fitness levels, muscle usage. Are there any regular monitoring processes as the disease progresses? For example, does the patient get regular assessed for muscle deterioration?		H2	P
		14.4	What measures could be put in place to help MERRF (EPILEPSY) patients recover from common illnesses?	P&P	OOS	P
		28.3	Care has been inconsistent, health professionals not easy to contact.. think reasesrch could focus on benefits of direct health care access compared to limited contact.	P&P	OOS	CS
		51.2	Why don't all mito diagnoses have an automatic referral to a specialist mito centre. My daughter is 5.5 yrs post diagnosis, now age []. We have annual neurology appointment and quarterly paediatric appointment.	P&P	OOS	CS
		54.3	if no cure, some sort of management plan with gp notified	P&P	OOS	P
		55.2	The doctors in [location] do not have a clue about my case they get medication wrong all the time which is dangerous to meet their needs to be more whereas with doctors and specialists	P&P	OOS	P
		68.2	I'd like to see emergency health care plans for all mito patients Including help for day to day care plans as well as emergency care	P&P	OOS	CS
		78.4	Managing my disease is hit and miss! I learn by errors! I have a PEG tube and the support service is practically non-existent!	P&P	OOS	P
		81.3	Having a mitochondrial specific care plan and a mitochondrial specific care pathway to have misconceptions and risk for these vulnerable population.	P&P	OOS	H
		83.4	What is the best use of available support to help.	P&P	OOS	P
		88.5	Should all pateints have a health care plan if hospitalized .	P&P	OOS	CS
		90.4	What are the support needs as people grow older and who understands this? E.g. The risk of people falling between physical and mental health services.	P&P	OOS	CS

		90.5	Better communication between the different involved professionals e.g. GP, local hospital consultant, [location - hospital] specialists, local psychiatric services etc.	P&P	OOS	CS
		91.9	Also good communication between the different professionals who might need to be involved.	P&P	OOS	P
		95.5	gp to provide more support when required	P&P	OOS	CS
		99.5	I would like to see research that promotes the development of better support services for patients affected by mitochondrial disease (practical, financial and psychological)	P&P	OOS	H
		121.6	how to care for patients with a mitochondrial disease.	P&P	OOS	CS
		145.4	Why is there no "lead" professional to coordinate cross specialty consultations denoting clear clinical responsibility? Is there a role for a key worker like in cancer where you have Clinical nurse specialists who can provide support?	P&P	OOS	CS
		145.5	More rapid referral to specialist who have knowledge of treatments that are available and support of a "nurse specialist" to aid decision making	P&P	OOS	CS
1.06E+10	54	54.1	I would like to know what disease I have, having no diagnosis means I cant access any sort of help from anybody		H4	P
		112.3	How can those without a difinitive diagnosis still be included and receive support		H4	CS
		68.4	A very difficult subject but more information on end of life care should be available when requested. Too many mito patients are pit through unnecessary suffering. As medical belief moves towards both saving lives and saving suffering I would like to see better end of life care plans		H5	CS
		85.3	Better supportive therapies. Better evidence for the use of supportive therapies. Better end of life planning.		H5	H
		98.2	Advanced care planning in mitochondrial disease		H5	H
		6.11	How much exercise is too much?		M1	P
		8.4	How does exercise impact thr disease		M1	CS
		13.5	Does excercise help or make it worse?		M1	P
		17.4	How 2 exercise? thinking of doing seated yoga? Have a DVD compuiled by double amputee.		M1	P
		23.2	I'd like to know more about the effects of exercise on my disease.		M1	P
		24.2	Can exercise be beneficial		M1	P
		27.7	How much exercise should I be doing?		M1	P
		50.5	the value of exercise does it exeacerbate progression?		M1	CS
		85.7	How much exercise and what forms of exercise should people with mitochondrial disease participate in? Should exercise be prescribed (like drugs)?		M1	H
		87.5	Role of exercise in treatment of mitochondrial cardiomyopathies		M1	H
		90.9	Understanding of appropriate levels of exercise, support of specialist physios and OT's.		M1	CS
		93.2	role of exercise in maintaining function.		M1	H
		93.5	exercise		M1	H
		94.12	Day to day gentle exercises that could be done at home?		M1	P
		117.12	Aerobic exercise and muscle strengthening in people with significant impairments such as cardiomyopathy or seizure.		M1	H

		117.4	Evidence for specific interventions including specific exercises for specific manifestations/phenotypes, including: fatigue, balance, tremor or ataxia, exercise for a range of genotypes (outside of 3243) and research about how best to support individuals to manage their conditions/how best to optimise participation in exercise in the longer term for example.		M1	H
		143.4	Exercise advice. What is the Positive impact from exercise?		M1	P
		6.12	Are supplements beneficial?		M2	P
		6.6	Does a gluten free, low carb or ketogenic diet help?		M2	P
		8.5	How does diet impact thr disease		M2	CS
		10.8	Best supplimennts to take		M2	CS
		13.6	eating and drinking makes me feel better- is this evidenced?		M2	P
		14.5	What foods should be included and would be benificial in the diet of MERRF patients?		M2	P
		16.3	We were told vitamins & coenzymes are useful anecdotally: is this the case ?		M2	CS
		16.4	Would a ketogenic diet be helpful?		M2	CS
		20.5	what diet can help with mitochondria, any information!		M2	P
		23.6	I'd like to know more about the effects of diet on my disease.		M2	P
		27.4	What is the best type of diet for me?		M2	P
		30.3	What is a healthy diet when you have Mitochondrial disease?		M2	P
		41.2	the Well-being of both our late son and daughter there is clearly linked with nutrition but how is unknown at present		M2	CS
		41.3	How effective are CoQ10 and Riboflavin as supplements? The ideal diet?		M2	CS
		42.3	Coenzyme Q 10		M2	H
		48.2	diet		M2	P
		52.9	how can i improve the daily quality of my life? how can i be sure that i am eating well? are there any specific dietary recommendations for my condition?		M2	P
		62.4	diet important, needs 2 be brain and heart food perhaps.		M2	P
		69.5	Definetly diet		M2	P
		70.11	Does a ketogenic or paleo diet help with energy production?		M2	P
		70.6	Can long term use of B vitamins cause breast cancer in a mito patient?		M2	P
		70.7	Does magnesium or other supplements help the mito body detox and reduce oxidative stress?		M2	P
		70.9	What supplements should I be taking?		M2	P
		71.3	What supliments or vitamins can help with mito.		M2	P
		72.3	Success of CoQ10 and other potential treatments of Alpers.		M2	CS
		72.4	Diet would be beneficial		M2	CS
		79.4	Which foods should be avoided if any ..		M2	P
		81.5	How diet adjustments would benefit the GI symptoms of patients?		M2	H
		84.3	Research of possible treatments, evidence of value of various vitamins and guideline of what mix to use in what condition.		M2	H
		84.4	Is diet important and in what way?		M2	H
		85.4	Is there an optimal diet for mitochondrial disease?		M2	H
		87.6	Role ofdiet in treatment of mitochondrial cardiomyopathies		M2	H
		90.7	Impact of diet - what might help or worsen things.		M2	CS

		91.5	What food is good or bad for my condition? e.g. is sugar good or bad?		M2	P
		92.4	as I have bowel issues a recommended diet would be helpful as currently I am receiving conflicting information.		M2	P
		93.4	research into effects of diet		M2	H
		94.11	Information on diet, as it is difficult to move as much as I'd like, but am trying to lose weight.		M2	P
		97.4	How effective is providing the body with a ketone supplement. Use of slow release carbohydrates such as Glycoside as an adjunct to a normal diet.		M2	H
		101.6	Some patients on the discussion group suggested fish oils could help but I don't know if there is any research to back this up?		M2	CS
		103.2	Impact / efficacy of nutrition / nutritional supplements on health outcomes.		M2	H
		110.6	Alternative diets		M2	CC
		115.5	What food and drinks could have made her condition better or worse.		M2	CS
		119.4	Diet to best support LHON?		M2	CS
		138.2	The utility of current non specific vitamin supplementation.		M2	H
		142.5	Is there any evidence that certain vitamins or foods may assist in stabilising LHON?		M2	CS
		143.2	Impact of nutrition for the specific disease. What is the Positive impact from nutrition		M2	P
		6.7	Does far infrared saunas help the MiTo body detox?		M3	P
		6.8	Are magnesium baths beneficial?		M3	P
		27.6	How many hours a week should I be working?		M3	P
		34.3	Can muscle be damaged by over doing day to day life because you won't give in.		M3	P
		70.10	Does infrared sauna help the mito body detox and enhance energy production?		M3	P
		100.4	Would any of the available treatments or life style recommendations have an effect on progresion of mitochondrial retinopathy?		M3	H
		115.8	Does stress and crying have an effect on the condition?		M3	CS
		115.9	Should certain treatments be limited as much as possible, e.g. tests which cause stress		M3	CS
		121.5	Better coordination amongst medical and nutritional elements to ensure optimal mitochondrial function		M3	CS
		141.5	What lifestyles help/ hinder ?are other treatments / therapies potentially of value?		M3	P
		7.4	What do patients and families find helps them to get on with life		M4	H
		15.4	what is ther best way of maintiaing good pschological health?		M4	CS
		31.11	How do people cope with the prognosis / uncertainty for the future?		M4	CS
		52.7	how can i improve my mental health when i know i have a progressive condition?		M4	P
		53.4	What impacts most in day to day coping		M4	P
		53.5	What one specific has helped to cope with mitochondrial		M4	P

		117.8	Evidence and information about specific strategies for optimising sleep, managing fatigue, managing stress and anxiety associated with diagnosis of mitochondrial disease - led by experience and strategies successfully used by people who have the diagnosis and impairments.		M4	H
		117.9	PPI to best understand experiences and priorities or strategies used by people with mitochondrial disease (eg - fatigue management)		M4	H
		118.5	How do I handle losing so many friends and kids of friends to Mitochondrial disease. How do I prepare my own family for this. How do I handle having my dreams i.e. House, job family of my own shattered. As a visual person I enjoy art, style, writing. Since sightloss I can no longer do this. I have lost my identity.		M4	P
		1.2	LHON: Does idebenone really work	INFO	OOS	H
1.06E+10	2	2.1	My son is taking IDEBENONE for lhon, are we any closer to any other treatments or medication in the future.	INFO	OOS	CS
		4.5	Why is there such slow progress in finding drug treatments?	BROAD	OOS	H
		5.4	What constitutes stress? Why is physical exercise not stress?	BROAD	OOS	CS
		6.3	Can MiTo patients donate organs?	INFO	OOS	P
		6.4	MiTo toxic drugs	BROAD	OOS	P
		8.3	What treatments might be of benefit	BROAD	OOS	CS
		9.2	what can happen to mito patients. The affects. Poss treatments. Drugs what can and can't be used	INFO	OOS	P
		9.3	When new medicines become available how quickly can I get them and is funding available. Also other treatments physiotherapy for balance issues etc more help	INFO	OOS	P
		9.4	Equipment available to help. Treatments, physiotherapy and big time psychological support needed but not always available	P&P	OOS	P
		9.5	More info available about psychological effects of mito.	INFO	OOS	P
		10.3	A list of medication that is safe to take and wont harm mitochondria more!	INFO	OOS	CS
		11.2	How care can take account of new developments and ho.w to be kept informed of these.	INFO	OOS	CS
		11.4	Information on effective alternative therapies and access to local psychological and social support	INFO	OOS	CS
		11.9	How can easier access to g.ps be ensured.	P&P	OOS	CS
		12.4	Updates on developments in effective diet, alternative therapies would be helpful.	INFO	OOS	P
		12.7	Local psychological and social support network	INFO	OOS	P
		13.7	Psychological support would have benefited me- about diagnosis, how I explain my condition, do I tell family members? How do I explain to strangers, that I am ill.	INFO	OOS	P
		13.9	What medications you should or shouldn't take?	INFO	OOS	P
1.06E+10	15	15.1	what are the causes of this disease?	BROAD	OOS	CS
		15.3	what is the best treatment?	BROAD	OOS	CS
1.06E+10	18	18.1	I'm not diagnosed with mitochondria disease but neuro specialist think so	BROAD	OOS	P
1.06E+10	19	19.1	How save my son ? If is alone with this mutation, who work for rechearch to save it 😩	BROAD	OOS	CS
		20.4	More access for physio,	P&P	OOS	P

		21.5	Are there any alternative therapies available?	INFO	OOS	P
		22.3	Are there any treatments that can help and when can that critical 1% be tested for my daughter and if she wants children??	INFO	OOS	CS
		23.3	I'd like to know more about different trials and vitamins and medicine I can take.	INFO	OOS	P
		23.4	I don't feel the social aspect is very well researched. I am often exhausted and struggle to do things but there is no research out there that tackles the issues facing patients and day to day life.	BROAD	OOS	P
1.06E+10	24	24.1	Greater research into Diabetes	BROAD	OOS	P
		24.3	I find it hard to explain to my wife what is happening to me and sometimes it causes conflict, I feel that sometimes it needs a third party to explain.	BROAD	OOS	P
1.06E+10	25	25.1	what treatment is there ? what research and where is it happening	INFO	OOS	CS
		25.3	also in [location] we have no help support network how can we get together with other families and raise the mito profile	INFO	OOS	CS
		27.5	Is there any psychological support available?	INFO	OOS	P
		27.8	Would it be possible to get more regular local support as travelling to Oxford is very tiring?	INFO	OOS	P
		28.4	Very useful to know treatment.. my daughter is profoundly deaf, had a contra-indicated drug before diagnosis, that caused hearing loss.	INFO	OOS	CS
		28.5	Would be good to know through research what to expect. We have had psychology assessments and diagnosis that we were told originally were not mitochondrial connected.	BROAD	OOS	CS
		29.2	Which treatment is safe and efficacious	BROAD	OOS	H
		30.2	It was my understanding there was no current treatment for Mitochondrial disease - true or false?	INFO	OOS	P
		31.2	How could it affect other family members (we don't yet have a genetic diagnosis)?	INFO	OOS	CS
		31.3	Our daughter has a clinical diagnosis - are there any circumstances where it could be ruled out? In cases where mitochondrial disease is misdiagnosed what does the alternative explanation turn out to be?	INFO	OOS	CS
		31.8	Which research into other conditions could potentially be relevant to mito patients?	BROAD	OOS	CS
		33.3	What can I do to help myself. What should I avoid	BROAD	OOS	P
		33.4	I am in pain and this is ignored despite asking for treatment. Can you provide a care plan	P&P	OOS	P
		33.5	Information on trials	INFO	OOS	P
		33.6	What psychological help can I access why do appointments take months to obtain	INFO/ P&P	OOS	P
		33.7	I struggle with psychological issues only with my health why can I not access treatment or help	P&P	OOS	P
		34.2	How to cope with symptoms.	BROAD	OOS	P
		34.3	Having had no treatment since diagnosis 20 years ago except Q10s. Is there any other treatment available.	INFO	OOS	P
		35.3	Anything other than treating symptoms as they arise?	INFO	OOS	P
		35.4	More information about 3parent baby treatment	INFO	OOS	P

		36.6	Without hospice support would not have coped. External professionals had little understanding of Leighs syndrome.	P&P	OOS	CS
1.06E+10	38	38.1	Hello, My brother had it. He also had' the wolfram syndrome'. He died with it in [year] kind regards	BROAD	OOS	CS
		39.2	Can we fund treatments that actually reach their primary endpoint in the trials not those treatments that are a triumph of marketing over efficacy?	BROAD	OOS	H
		40.3	I would like to assess the efficacy of several different compounds in clinical trials.	BROAD	OOS	H
		40.4	What is the impact of mitochondrial disease on your family? Financial, educational, practical?	BROAD	OOS	H
1.06E+10	43	43.1	the genetics of my condition have been explained to me by medical proffesionals including the possibilities of my children inheriting the condition	BROAD	OOS	P
		43.3	In discussion with the neurologist we have discussed if and what medication might aleviate some of the negative aspects of the condition but there appears to be nothing other than strong pain killers avaialabi.At the moment any benefit that might be gained from medication seems to be outweighed by the side effects. It was suggested that I try co enzime q 10 but these seemed to make little any difference	BROAD	OOS	P
1.06E+10	44	44.1	What tests should be done to determine who carries defective genes?	D1	OOS	CS
		44.2	There is no care of Alpers Disease that helps	BROAD	OOS	CS
		44.5	Alpers is so final, there are too many questions!	BROAD	OOS	CS
1.06E+10	45	45.1	Any cure	BROAD	OOS	CS
		47.2	General patient education leaflet re mitochondrial eye diseases.	INFO	OOS	H
		47.5	Research + education regarding donated mitochondria for conception.	BROAD	OOS	H
1.06E+10	48	48.1	underlying causes and why	BROAD	OOS	P
		48.4	support groups in your local area	INFO	OOS	P
1.06E+10	50	50.1	Cause	BROAD	OOS	CS
		50.4	General treatment for symptoms	BROAD	OOS	CS
1.06E+10	51	51.1	None. Daughter is polg1 Alpers We understand cause, genetics. Palliative support just needs to be specialised.	BROAD	OOS	CS
		51.3	I would like to have further comprehensive information on cbd oil, financial support, practical support. Strengths, dose introduction, effective ways to administer.	INFO	OOS	CS
		51.5	Is there a national framework in place to guide professionals team in support of sufferers.	P&P	OOS	CS
		52.10	what social care / support is available ?	BROAD	OOS	P
		52.2	are there any tests that are diagnostic that do not involve invasive procedures?	INFO	OOS	P
		52.4	do you have specific factsheets for each type of disease? who provides the care when you do not live near the centre of excellence? who is responsible for providing the care? is there any financial support available? what support groups are available? how do you assess daily living activities amd ability to carry out daily activity?	BROAD	OOS	P

		52.6	when will there be a cure? when will there be an effective treatment ?	BROAD	OOS	P
		52.8	which medications should be avoided , including OTC?	INFO	OOS	P
1.06E+10	53	53.1	What progression has been made to help quicker diagnosis	D1	OOS	P
		53.2	More information on hydration	INFO	OOS	P
		53.3	More awareness to patients suspected of having mitochondrial	BROAD	OOS	P
		54.4	tech and equipment, I struggled for years with visual impairment before someone put me in touch with rnib who give me things to try, previously I had to pay for things that turned out to be no use, lists of things that have proven benehit would have saved me a fortune and given me a better quality of life	INFO	OOS	P
		55.3	I wasn't told that mitochondrial can cause diabetes I would have liked to have known so I could maybe prepare for the future	INFO	OOS	P
		55.4	Getting equipment from ot is very difficult you have to pay for everything when it's not necessary we need these things to rely on	P&P	OOS	P
		55.5	Thank god we have boys for the disease cannot be passed on through female	BROAD	OOS	P
		56.4	How can we best assess who with mitochondrial diseases can safely drive?	BROAD	OOS	H
1.06E+10	57	57.1	Are there more potential ways to diagnose Mitrochondrial or even a specific type of the disease?	D1	OOS	P
		58.2	CAUSES: aimed at potential therapy interventions	BROAD	OOS	H
1.06E+10	59	59.1	none, its happened, we've moved on, we need a cure now	BROAD	OOS	CS
		59.2	Mitochondrial donation was great for the headlines and a knighthood, but what about the people here and now who need a cure.	BROAD	OOS	CS
		62.5	Yoga or pilates.....seated of course	BROAD	OOS	P
		62.6	Facebook group, Lily foundation r only support	BROAD	OOS	P
		62.7	Surely therapies and diet can help MS, Parkinsons so can perhaps have a support group 4 these. in fact ALL question 6	BROAD	OOS	P
		65.4	help with getting my son out of his home he finds it hard to mix with his frineds the same age	INFO	OOS	CS
1.06E+10	66	66.1	To give more to people that got mitochondrial disease and more research and more awareness	BROAD	OOS	P
		66.2	Mitochondrial disease	BROAD	OOS	P
		66.3	more treatment needs to be done	BROAD	OOS	P
		66.4	More equipment	BROAD	OOS	P
		67.4	Questions about all the pints you mention above	BROAD	OOS	P
1.06E+10	68	68.1	How can those drugs that are at experimental stage be used to help those who are at end of life, where their experience could help in research	BROAD	OOS	CS
		68.3	More compassionate use of drugs for those who have no other options. So drugs that are in early research being used on compassionate grounds, to aid further research into that drug	BROAD	OOS	CS
		69.4	I have to admit I'm well informed	BROAD	OOS	P
		69.6	access to physiotherapy and other therapies available	P&P	OOS	P

		70.12	Would local area Mitochondrial Disease Reps or information Co-ordinators in the UK, be beneficial with updated information sharing. Maybe willing able patients or a nominated local health professional?	P&P	OOS	P
		70.4	Can a mito patient be an organ donor?	INFO	OOS	P
		71.2	What medicine can be available for certain symptoms I have relating to mito	BROAD	OOS	P
		71.4	Help in regards to information for pregnancy and mito	INFO	OOS	P
		71.5	Symptoms that can occur due to mito, more research, a ‘place to look’ for more understanding of mito and how it can effect different people in different ways.	INFO	OOS	P
		72.2	Best practise	BROAD	OOS	CS
		74.3	Make medicinal cannabis available to patients with MD related diseases, or at least decriminalise use for medicinal use.	P&P	OOS	P
		74.4	technology is out there, for example, robotic arm supports, wheelchairs that do steps, wheelchairs with seat risers, core muscle supports, but this equipment is out of our price range, not available to us due to financial pressures of living off PIP (those lucky enough to get it), if we had access to this kind of technology, many of us could stay in work longer, live more independently in our own homes for longer, use less hours of PA support, and avoid accidents that cost the NHS money.	P&P	OOS	P
		74.5	PIP assessments need to be put back into the hands of our medical teams. Better access to social care, training for medical teams working with us, and will we miss out on essential research once the UK leaves the EU?	BROAD	OOS	P
1.06E+10	75	75.1	WHAT caused the mito disease	BROAD	OOS	P
		76.3	More research into cures/treatments is needed.	BROAD	OOS	P
		76.4	Prevention is also key, may an in vitro test that could be performed, or genetic testing prior to pregnancy.	P&P	OOS	P
1.06E+10	77	77.1	I need to be kept up to date with research if not for me, might help those that follow.	BROAD	OOS	P
		77.3	What support is out there for patient. I have,'t found any!	INFO	OOS	P
		77.4	I was informed there is no treatment and no cure.	BROAD	OOS	P
		77.5	Day to day life is difficult, fatigue is a major factor and dietary needs as have a peg tube. Group support would help tremendously. Never met anyone with this before!	INFO	OOS	P
1.06E+10	78	78.1	more information about disease seems to be so little to help me deal with the condition I have.	INFO	OOS	P
		78.3	I understand there is no treatment for my condition but would like to know of any potential treatment in the future.	INFO	OOS	P
		78.5	How do I contact people with same disease as myself-specifically mitochondrial myopathy.	INFO	OOS	P
		79.2	What treatments are there for the disease	INFO	OOS	P
		79.3	Why can’t I get coq 10 and riboflavin through my consultant or gp ..	P&P	OOS	P
		80.2	How can pts access support for swallowing and communication difficulties	AUDIT	OOS	H
		82.2	How can we improve outcomes of patients with mitochondrial disease ?	BROAD	OOS	H

		82.3	How can we improve treatments for patients with mitochondrial disease ?	BROAD	OOS	H
		82.4	How can we improve quality of life of patients with mitochondrial disease ?	BROAD	OOS	H
		83.2	What would be the best use of available treatments to help living with the disease day to day.	INFO	OOS	P
1.07E+10	84	84.1	Faster turnaround of the diagnostic process, agreed pathway of investigation nationally.	D1	OOS	H
		84.5	Prospective monitoring of a newborn diagnosed due to family history.	D3	OOS	H
		85.5	Discovery of new treatments. More evidence about existing 'treatments'. Effective clinical trials.	BROAD	OOS	H
		85.6	How to increase awareness of mitochondrial disease. How to leverage more research funding for this area.	P&P	OOS	H
		85.9	What are the causes in patients who do not currently have a diagnosis, despite extensive investigation?	BROAD	OOS	H
1.07E+10	86	86.1	I would like to see a reliable testing system for mitochondrial disorders. It is frustrating to "know it is mitochondrial" and struggle to get a genetic result or not be able to get a genetic result at all.	D1	OOS	H
		87.4	Novel therapies for mitochondrial cardiac disease	BROAD	OOS	H
1.07E+10	88	88.1	Why only one child out of the 2 has mito, we know what caused it and the diagnosis but not the genetics ie exacat mutation .	INFO	OOS	CS
		88.3	When a new treatment becomes available we should be told straight away if it would benifit . q10 has never been offered yet lots of people with mito talk about it . Should it be offered to everyone.	INFO	OOS	CS
		88.4	Pyshio it is always said it helps yet its hard to get . It would be nice to hear about anything that would help manage day to day life , and be offered them .	INFO	OOS	CS
		91.4	Research into treatments that might help.	BROAD	OOS	P
		91.8	If you overload your mind can it cause a stroke?	BROAD	OOS	P
		92.2	I would like to have an information sheet in laymans language covering care that could assist me in the future.	INFO	OOS	P
		92.3	we are all waiting for any form of treatment that would alleviate our symptoms.	BROAD	OOS	P
		93.3	developing technologies to assist with progressive disability.	BROAD	OOS	H
		94.7	Equipment for the house, such as a second bannister for the stairs, bottle openers, handles in the bathroom etc.	INFO	OOS	P
		94.8	When would possible treatments and a digital and physical resource letting people know that I have mitochondrial disease, symptoms etc become available? Are these things that could happen in my lifetime? How do I do more to help aid the research into mitochondrial disease?	INFO	OOS	P
		94.9	Empathy from healthcare professionals, and patience.	P&P	OOS	P
		95.3	Support and information	INFO	OOS	CS
		99.2	How can we speed up the diagnostic process and get these patients to specialist services faster?	D1	OOS	H
		100.2	How to diagnose and confirm the diagnosis of mitochondrial retinopathy?	D1	OOS	H

1.07E+10	102	102.1	Medicines	BROAD	OOS	CC
		102.2	Therapies	BROAD	OOS	CC
		103.7	Disease classifications need updating based on new genetic profiling capabilities.	D2	OOS	H
		104.3	What is becoming available	INFO	OOS	CS
		104.4	What mental health resources are available for people losing sight	INFO	OOS	CS
		105.5	Quicker diagnosis and genetic testing is required for LHON. Typically it takes many months to receive a diagnosis and mis-diagnosis is common.	D1	OOS	CS
		106.4	Do you have access to new technology to improve participation in day to day activities?	P&P	OOS	H
		106.5	what are patient priorities in mitochondrial disease?	BROAD	OOS	H
		108.2	And what treatments are avalable	INFO	OOS	P
		108.3	scans shud be disgusted and offered	P&P	OOS	P
		108.4	they should cover any forms of treatment s	P&P	OOS	P
		108.5	there should be more support out there and patients should be offered to attend groups to use equipment	P&P	OOS	P
1.07E+10	109	109.1	AS a GP we see rare conditions rarely but often are involved with whole affected families. It is often difficult to find the expert advice patients need. Easily searchable professional and patient advice is the first step. The next thin we need is EBM treatments and research across similar rare diseases could allow sharing of information of treatments that may help even if the patient numbers for very rare diseases in samples are small. Good genetic understanding of diseases is vital for families.	INFO	OOS	H
		109.4	Diet, technology.	BROAD	OOS	H
		109.5	options for patients to electronically consult, really helpful. also for Gps to have clear options to seek advice out of area where appropriate	P&P	OOS	H
1.07E+10	110	110.1	Clearer explanation in layman's terms of genetic causing condition	INFO	OOS	CC
		110.5	Psychological support must be paramount without fear from reporting to outside agencies!	P&P	OOS	CC
1.07E+10	111	111.1	New genes associated with the disease	BROAD	OOS	H
		111.4	Leaflet on ongoing trials (natural history or treatment trials) A database of associations and meetings dedicated to mitochondrial diseases around the world not only locally.	INFO	OOS	H
		111.5	Side effects; mechanism how drug works; latest trials updates.	INFO	OOS	H
1.07E+10	112	112.1	Diagnosis and what to expect	BROAD	OOS	CS
1.07E+10	114	114.1	confirmation of genetic diagnosis	BROAD	OOS	H
		114.2	most reliable data on natural history to enable genetic counselling	INFO	OOS	H
		114.4	best available medications and other treatment options, knowledge about the results of clinical trials	INFO	OOS	H
		114.5	lists of suggested medicines, medications to avoid, help with physiotherapy, OT	INFO	OOS	H

1.07E+10	115	115.1	We are still going through testing for the genetic causes for our daughter's condition - there is no guarantee we will be given a genetic diagnosis - this is the most important question we want answering, in terms of how it was inherited.	INFO	OOS	CS
		115.2	How would it be best to explain the condition to all health professionals as quickly and efficiently as possible?	P&P	OOS	CS
		115.3	What medicines could have made the condition better/worse (she has now died).	INFO	OOS	CS
1.07E+10	116	116.1	Genetics: Can you tell if other family members who are not affected at the moment may have this condition.	INFO	OOS	P
		116.2	Many years ago I was advised by a consultant at Moorfields that stem cell would cure this condition and I would be able to drive again. What is the update on this research?.	INFO	OOS	P
		118.2	Is there a card you can give out to people explains what Mito is.	INFO	OOS	P
		118.4	How can I maintain independence, travelling on my own, making food and drinks, know who I'm talking to, how do I cope knowing the future.	INFO	OOS	P
1.07E+10	119	119.1	What causes LHON?	BROAD	OOS	CS
		119.2	How can affected LHON carriers get information on meeting other affected people in uk? Support groups etc	INFO	OOS	CS
		119.3	What can be taken to improve eyesight?	INFO	OOS	CS
1.07E+10	120	120.1	are there any specific genes/hla markers that can be identified in patients with LHON that might prove useful in pre-natal testing	diagnosis	OOS	H
		120.2	A national register of patients held by central government would be helpful in monitoring and managing resources for this condition	P&P	OOS	H
1.07E+10	123	123.1	causes, genetics and progression	BROAD	OOS	H
		123.2	care	BROAD	OOS	H
		123.4	treatment	BROAD	OOS	H
		123.5	managing day-to-day life	BROAD	OOS	H
		123.6	reproduction	BROAD	OOS	H
1.07E+10	125	125.1	Being able to rapidly identify the genetic basis of suspected mitochondrial disease is critical. Work flows around improving the currently fragmented testing pathways in the UK via the NCG services, as well as expanding the repertoire of nuclear gene candidates considered to be disease causing, are both important.	D1	OOS	H
		125.2	Improving standard structure of disease surveillance.	P&P	OOS	H
		126.2	Evidence based guidelines for management.	P&P	OOS	H
		126.3	Treatments for which there is evidence and guidance on which treatments there is no evidence	BROAD	OOS	H
		127.2	I think we need more disease guidelines specifically related to the different forms of mitochondrial disease. Ultimately it would be excellent if these were evidence based but this is a challenge with small numbers of patients	P&P	OOS	H
		127.3	There is a desperate need for treatment to alter the course of mitochondrial disease. A major focus of research should be finding new treatments and then evaluating them with high quality clinical trials	BROAD	OOS	H
		127.5	improved communication aids for those with sight or speaking difficulties	BROAD	OOS	H

		128.2	I would like to know whether novel social programmes have been implemented for patients with mitochondrial eye disease.	BROAD	OOS	H
		128.4	I would like to know whether support groups between patients with similar conditions such as sight loss have been developed and whether these are effective in improving quality of life.	BROAD	OOS	H
1.07E+10	130	130.1	Cognitive outcomes with robust neuropsychological measures	BROAD	OOS	H
		130.3	The impact on quality of life; the impact on education; the impact on social opportunities; the impact on adaptive functioning	BROAD	OOS	H
		131.2	Case series remain useful to document the incidence of various complications (& so the need for differing monitoring), particularly now that these can be specific to particular genetic disorders, though the heterogeneity within any one mitochondrial disorder somewhat limits the value.	BROAD	OOS	H
		131.3	Obviously, it would be nice to develop effective treatment but this is a long way off for many diseases. Controlled trials with clinical endpoints are important.	BROAD	OOS	H
		131.4	Families are good at exchanging ideas about this through social media etc but objective evidence concerning alternative therapies is good. In my experience, the biggest dietary issue is often getting enough into young handicapped mitochondrial patients & the best treatment is often a gastrostomy!	BROAD	OOS	H
1.07E+10	132	132.1	Frontline testing in suspected cases -functional or genomic?	D1	OOS	H
		132.2	Are there any better biomarkers for mitochondrial disease on the horizon that are based on major manufacturer's platforms and not specialist tertiary service labs?	INFO	OOS	H
		134.3	access to trial info	INFO	OOS	H
		135.2	Access to family screening information, when children should be screening i.e. at what age? Do they need to be followed up in Metabolic Clinics/Genetic clinics if asymptomatic?	D3	OOS	H
		135.3	Should they reveal their genotype status when applying for life insurance/mortgage?	P&P	OOS	H
		135.6	availability and effectiveness of three-person family.	INFO	OOS	H
		135.7	The management of lactate of 8mmol/L in mito patients- does it require sodium bicarbonate if renal function and ABG are normal and patients are asymptomatic?	BROAD	OOS	H
1.07E+10	136	136.1	How can these diseases be diagnosed in timely manner especially antenatally?	P&P	OOS	H
		136.3	An information sheet with all the health surveillance requirements for all systems involved with alerts for clinical signs that need urgent attention by the specialist centre	P&P	OOS	H
1.07E+10	138	138.1	Better framework for genetic investigation and establishment of genotypic/ phenotypic specific national databases to enable proper natural history collection.	P&P	OOS	H
		138.3	Utility of systemic screening.	P&P	OOS	H
		140.3	We were told to contact RNIB. That's it. Professional Opthalmologist said sorry I can't help anymore.	BROAD	OOS	CS
		141.4	Availability/costs - if appropriate and options. Relating to drugs and other possible medical procedures. *Risk factors explained/ quantified.	INFO	OOS	P

		142.4	Does Idebenone conflict with any other medications e.g. painkillers, heart medications etc?	INFO	OOS	CS
		143.5	Which medicines should be avoided. Are there any medicines which can help.	INFO	OOS	P
		144.4	We know that companies struggle to justify investment in treatment options for rare diseases. Why is there so little publicly funded research in this area, given that we know that there could be wide ranging benefits in other conditions that could result from breakthroughs in these mitochondrial conditions?	P&P	OOS	CS
		144.5	Similar to my question about treatments (lack of public funding) but even more so when it comes to health supplements, diet etc, when there is little commercial motivation for such work. Where it does exist (eg study on effect of ketogenic diet for mito disease) it should not be hidden behind paywalls and therefore inaccessible to the vast majority of potential beneficiaries (what is the point of that)?	P&P	OOS	CS
1.07E+10	145	145.1	Better knowledge understanding of doctors of these rare diseases may speed up diagnosis .	D1	OOS	CS
		145.7	Why does it take so long to bring all the actions together e.g. Family member diagnosed, siblings and other family need genetic counselling and so on .	P&P	OOS	CS
		146.2	I would like to feel that GPs though interested have more knowledge to help	P&P	OOS	P
		146.3	Basic help with energy giving medicines would help	BROAD	OOS	P
		146.4	Simple information about what information about basic equipment	INFO	OOS	P
		147.2	Are healthcare professionals able to recognise the significance of GI dysmotility in this group of patients?	P&P	OOS	H
1.06E+10	7	7.1	Good evidence around prognosis for different conditions and the factors that may influence prognosis		P1	H
1.06E+10	8	8.1	Better prediction of prognosis		P1	CS
1.06E+10	9	9.1	How it affects everything especially the brain and how that can progress		P1	P
1.06E+10	11	11.1	Progression with aging.		P1	CS
1.06E+10	12	12.1	What progression to expect as time goes on		P1	P
1.06E+10	14	14.1	How will/ or will my ataxia develop over time?		P1	P
1.06E+10	16	16.1	My son has a SURF 1 mutation (leigh's disease) I'd like to know about other people with the same mutation as his & how rare it is to have survived into adulthood		P1	CS
1.06E+10	17	17.1	why do i bruise easily? is it my age or related 2 mito?		P1	P
1.06E+10	20	20.1	Information on what this condition has on people as you get older		P1	P
1.06E+10	23	23.1	I would like a more complete picture of what I may be affected with later in life. Doctors are always very vague and talk about "maybe this, maybe that". I never feel like I'm getting the full picture. I want to know exactly what ailments I'll have in the future.		P1	P
1.06E+10	27	27.1	I would like to know what the progression of my particular type of mitochondrial disease is likely to be?		P1	P

1.06E+10	30	30.1	What progression of the disease might I see over the next five to ten years? - for example, will I still be walking in ten years?		P1	P
1.06E+10	31	31.1	How can long it be managed without further progression (best case scenario)?		P1	CS
		33.2	what's the prognosis.		P1	P
1.06E+10	34	34.1	Does it get worse with age.		P1	P
1.06E+10	35	35.1	I have melas they don't really go into detail about it and what to expect as I get older or life expectancies and just tell you to look on the internet which doesn't really tell you much about it and can come up with really disturbing search results		P1	P
		40.7	What is the natural history of (various) mitochondrial diseases		P1	H
		43.2	I continue to see the consultant neurologist on an annual basis. she has suggested that I have an ECG at the same frequency. If there appears to be any significant changes further investigations are arranged ie ultrasoundI have also been provided with an information sheet for a hospital should I be admitted on an emergency basis. My main concern is about how my condition will develop.At the moment symptoms seem to be quite stable, There does however seem to be little information about how the condition might develop and at what rate.		P1	P
1.06E+10	49	49.1	The only answer I have had about progression is that it likely to continue at the same rate as it has, which does not help much when I need to plan to the end of my life.		P1	P
		49.4	It is just about the progress of the disease. Am I likely to need certain care in 10 years time and what sort when I have 14 disorders? Will I be dead? Should I retire or marry? Which limitations should I accept?		P1	P
		52.3	how do you quantify progression of disease? how would i know what life might be like in X years time? does the disease ever stop progressing?		P1	P
		54.5	the long trem effects and prognosis for strokelike episodes, also if I had a brain tumour it could be biopsied, a reasonably safe way to test brain tissue would be good, yes I will volunteer for trials as my brain is already fucked, but not if its done at [location]		P1	P
1.06E+10	55	55.1	Would like to know how this is going and pan out with mitochondrial I've had it since I was a baby I'm [age] nearly and it is affecting my life more now		P1	P
		60.2	Also her "code" has never been seen before so progression wise I Would like to know more about what's in front of her.		P1	CS
		61.2	Longer term outlooks		P1	CS
1.06E+10	62	62.1	am nearly [age], son and one daughter has inherited it, but not the other daughter.their main issue was hearing loss which we thought was the problem so bal poor. VERY worried about future		P1	P
1.06E+10	63	63.1	No information about long term impact of condition just told wait and see		P1	P
1.06E+10	64	64.1	What we can expect the progression of the disease to involve? Life expectancy? General health as they age?		P1	CS
1.06E+10	65	65.1	i like to know more on the progression of it		P1	CS

1.06E+10	67	67.1	How it will progress over time as I get older?		P1	P
1.06E+10	69	69.1	Mostly the effects over time. Original research concentrated on eyes and gearing but none on the effects on muscles and other organs.		P1	P
		77.2	Basic information on what to expect based on experience.		P1	P
1.07E+10	82	82.1	How can we better predict outcomes of patients with mitochondrial disease ?		P1	H
		82.5	How can we predict which organs will be involved ?		P1	H
		87.2	Natural history of conduction disease and cardiomyopathy		P1	H
1.07E+10	89	89.1	The prevalence of Dysphagia within mitochondrial disease and whether it is progressive.		P1	H
		90.2	What is the prognosis in terms of progression?		P1	CS
		91.10	and rate of progression		P1	P
1.07E+10	92	92.1	concerned about the progression as currently my eyes and balance are affected and would like to know what to expect long term.		P1	P
		94.3	How will the progression affect me in the future?		P1	P
1.07E+10	95	95.1	Related Health conditions deterioation of health		P1	CS
1.07E+10	101	101.1	something that comes up frequently on our online adult support group discussion is the unpredictable nature of mitochondrial disease and how it is difficult to manage symptoms that may be changing on a regular basis. The progressive nature of the disease is also hard to deal with, and patients/families often say they are afraid of the unknown. Is there anyway that research could address this, perhaps in the form of more natural history studies for specific mitochondrial conditions? The data could then be used to provide advice/guidance to those who have concerns.		P1	CS
		103.3	Immunological, cardio, nephro and pre-diabetic risks		P1	H
1.07E+10	108	108.1	i would like to no what courses the disease how bad it can get		P1	P
		110.2	True answer on degeneration		P1	CC
1.07E+10	118	118.1	How long will I have my sight for (my optic nerve is damaged)		P1	P
1.07E+10	133	133.1	How does this effect diabetes		P1	P
		134.2	long term outcome data		P1	H
		135.8	long-term outcomes, disease progression and prognosis.		P1	H
1.07E+10	139	139.1	Understanding of progression to allow understanding of disease course and what might influence this.		P1	H
		139.3	Good natural history studies		P1	H
		142.2	After a certain time is it likely that the condition will become stable and there will be no further deterioration in sight? Or is it still possible for a further loss of remaining vision?		P1	CS
1.07E+10	143	143.1	How does the disease progress over time - changes. Can the severity of the disease be explained. Impact on the body.		P1	P
additional 1	146	146.1	I would like to understand about progression. I know how difficult this is with the great variety of symptoms.		P1	P
1.06E+10	1	1.1	LHON - how can we predict who will get clinical disease		P2	H
		6.10	What are the chances of symptom progression depending on % of mutation?		P2	P
1.06E+10	21	21.1	Is there a way to predict the severity of the disease in infants of parents with Mitochondrial Disease?		P2	P

		105.4	Can a test be developed that predicts the chances of a carrier going blind?		P2	CS
		124.3	Is there a benefit to establishing percentage mutation load in a particular patient?		P2	H
		139.4	Understanding of predictive factors		P2	H
1.06E+10	41	41.1	Whether the stomach cancer ur son developed in his early 40s was connected to his unnamed mito condition that also affects his surviving older sister		P3	CS
		70.5	Is cancer more prevalent in mito patients?		P3	P
		90.10	What is the life expectancy?		P4	CS
1.07E+10	91	91.1	Research into life expectancy of different mitochondrial disorders (e.g. I have POLG1)		P4	P
		98.6	Differences between males and females		P5	H
		105.2	Why are males usually affected more than females?		P5	CS
		94.2	How do I know what chance there is of passing it onto children, if I have any?		P6	P
		129.2	To be able to give better informed and more accurate reproductive risks in mtDNA diseases.		P6	H
1.07E+10	134	134.1	association between mitochondrial mutation load and recurrence risk in offspring.		P6	H
		57.2	Why is general anaesthetic more of a risk to patients with Mitrochondrial?		T1	P
		115.4	Should certain treatments be limited as much as possible, e.g. surgery		T1	CS
		115.7	Does surgery have an effect?		T1	CS
		31.9	How long is it possible to go without food or drink overnight/when unwell without risking decompensation?		T10	CS
		115.10	Does starvation for surgery have an effect?		T10	CS
		5.2	How to care the patient so to minimize incidents of SLEs.		Q	CS
		56.2	Do people who adhere maximally to guidelines have the best outcomes?		M4	H
		121.3	Exploration of deep brain stimulation as a treatment for patients with a mitochondrial disease,		Q	CS
		130.2	The appropriate education provision for children	BROAD	OOS	H
		1.3	how to prevent clinical disease onset		R1	H
		1.6	New treatments to prevent clinical disease from developing in those at risk		R1	H
		4.4	Does early diagnosis and treatment with new drugs prevent blindness in patients who are carriers of mitochondrial mutations predisposing to LHON?		R1	H
		99.6	How can we prevent first eye involvement?		R1	H
		104.2	What to avoid [TRIGGERS FOR LHON]		R1	CS
		105.7	Can triggers be avoided? Is it possible to avoid triggering the condition?		R1	CS
		114.3	treatment options, information on disease progression, screening for potential complications to treat them early, options to prevent disease		R1	H
1.07E+10	122	122.1	To what extent do non genetic components influence disease phenotype and to what extent can these be influenced.		R1	H
		141.2	Prevention/ control of these factors ?! [TRIGGERS]		R1	P

		145.3	Are there known risk factors that could have prevented this disease?		R1	CS
		1.4	treatments to prevent progression/ 2nd eye involvement.		R2	H
1.06E+10	3	3.1	Progression as it's the most under researched		R2	P
		4.6	What is the role of exercise in preventing decline in vision in patients affected by vision loss with LHON?		R2	H
1.06E+10	5	5.1	What speeds up or slows down progression		R2	CS
		6.9	Can adult onset MiTo disease (after some progression) improve and symptoms disappear with good management and care?		R2	P
		8.2	What can We do to slow progression		R2	CS
		22.4	Anything to limit the condition or improve any longevity would be of interest		R2	CS
		28.2	Also how it has progressed and whether anything can be done to slow progression.		R2	CS
		31.7	Which other treatments could potentially prevent or slow progression?		R2	CS
1.06E+10	36	36.1	Nutrition given before diagnosis affecting long term outcome and life span.		R2	CS
1.06E+10	42	42.1	The value of coenzyme Q 10 to stop prgression		R2	H
		44.3	How to prolong life of Alpers Child.		R2	CS
		50.2	delaying progression		R2	CS
		61.4	or trying to slow down or stop hearing loss occurring.		R2	CS
		64.2	A cure, treatment that would prevent further mitochondria dying		R2	CS
		65.3	it would be good if they found something to slow it down my sons is growing fast		R2	CS
		70.2	Does a clean diet help prevent progression? Does toxic overload in body exacerbate progression?		R2	P
		73.3	Is there any medication to prevent symptoms getting worst or slow down the symptoms		R2	CS
1.06E+10	79	79.1	How to slow it down		R2	P
		90.6	As much research as possible into treatments that may improve the condition or slow progress of illness. Dare we ever hope for a cure?		R2	CS
		91.2	and how to slow it down.		R2	P
1.07E+10	93	93.1	any drugs/ supplements to slow deterioration.		R2	H
		97.3	Medications to stop disease progression.		R2	H
		99.3	In LHON: How can we prevent second eye involvement?		R2	H
		105.6	Can a treatment be developed that protects carriers from going blind?		R2	CS
		117.6	to reduce or slow rate of disease progression		R2	H
		118.3	What treatment would help the optic nerve stop dying and strengthen		R2	P
		141.7	Ditto the progression of the condition.		R2	P
		11.7	Alleviating symptoms such as fatigue		S1	
		12.9	Alleviating fatigue issues I have		S1	
		20.3	Mitochondria disease is untreatable but more information on treating symptoms- more energy		S1	
		27.3	Is there likely to be a treatment to help with energy levels?		S1	P
		32.2	What are best forms of Strategies for fatigue?		S1	H

		41.6	How to manage the increasing exhaustion that has come with age.		S1	CS
		67.3	Cure for tiredness!		S1	P
		76.5	Treatment to help increase energy levels, fatigue is a big issue for mito patients.		S1	P
		93.6	fatigue management		S1	H
		94.15	Will there be any treatments, in any form, to help with fatigue?		S1	P
		97.2	How much and what type of energy dense foods are most effective in managing fatigue.		S1	H
		98.4	CoQ10 placebo controlled double blind crossover study for fatigue		S1	H
		101.4	our adult support group discussions show that by far, the most common symptom that has the biggest impact on many adult patients with mitochondrial disease is fatigue/low energy. This can affect every part of life, including relationships, employment, friendships etc, and can be very isolating. Research into treatments that could improve energy levels would be a priority for many.		S1	CS
		112.2	How to help him with energy levels		S1	CS
		117.5	Ongoing research into medications to manage symptoms (fatigue for example) physical treatments		S1	H
		7.3	What are the best ways to treat the symptoms related to mitochondrial disease (including pain		S2	H
		11.3	Developments in pain relief.		S2	CS
		12.3	Appropriate pain relief and developments in this area.		S2	P
1.06E+10	26	26.1	Are there any pre disposing factors that make symptoms worse or better? For example hot or cold temperatures can make my pain fluctuate		S2	P
1.06E+10	32	32.1	What are best forms of analgesia for muscle pain?		S2	H
		94.14	Will there be any treatments, in any form, to help with pain?		S2	P
		117.11	Ongoing research into medications to manage symptoms (myalgia for example) physical treatments		S2	H
		135.5	Pain management,		S2	H
		7.6	What are the best ways to treat the symptoms related to mitochondrial disease (including cognitive decline)		S3	H
		94.16	Will there be any treatments, in any form, to help with cognitive function?		S3	P
		101.5	Another question would be if there is any treatment that could help with brain/cognitive function? Some patients get very tired very quickly and find it difficult to concentrate for periods of time, and miss doing things like reading.		S3	CS
		11.8	Alleviating symptoms such as balance issues.		S4	CS
		12.6	Alleviating balance issues I have		S4	P
		20.6	Mitochondria disease is untreatable but more information on treating symptoms- the shaking		S4	P
		94.17	Will there be any treatments, in any form, to help with coordination?		S4	P
		117.7	optimising function in people with cerebellar ataxia, balance problems, dystonia and/or tremor		S4	H

		10.7	Is there a certain feed a g peg fed person should be fed to help maintain healthy brain etc.		S5	CS
		36.5	Digestive research into what can be done before eating is lost.		S5	CS
		41.4	Bowel motility and treatments.		S5	CS
		41.5	The ideal PEG regimen and feed? Our daughter cannot tolerate most feeds, also needs a slow speed of delivery.		S5	CS
		80.3	Do any treatments help dysphagia?		S5	H
		80.4	Which pts needs PEG's? Are there particular foods that are consistently difficult to swallow		S5	H
		96.4	Are there any less invasive ways of providing feeding support to young babies with mito? My daughter took a significant deterioration of health each time she needed a new nasogastric tube fitted.		S5	CS
		98.3	Gastrostomy support and quality of life		S5	H
		147.3	As constipation due to GI dysmotility is common and can lead to significant deterioration in condition and potentially life-threatening pseudo obstruction, what treatment options are available to manage this?		S5	H
		94.5	Will there be any treatments, in any form, to help with muscle weakness?		S6	P
		138.4	The best management of myopathy		S6	H
		103.4	Efficacy of levocarnitine in primary and secondary carnitine deficiency.		Q	H
		103.5	Efficacy of Levocarnitine in prediabetes.	BROAD	OOS	H
		120.3	Apart from daily oral medications are there are long-term formulations that can be used to give a depot injection of idebenone?		Q	H
		1.5	Role of gene therapy		T2	H
		5.3	I'd like to see treatment that effectively shift heteroplasmy level for mtDNA patients		T2	CS
		11.6	Possibilities of genetic solutions in near future		T2	CS
		12.8	Is there any possibility of genetic solutions in the near future		T2	P
		25.2	what treatment is there and also we were told of gene removal for going forward if we had p any more plans		T2	CS
		47.3	Development of current treatment options (eg idebenone), other drug treatments, and genetic therapy possibilities.		T2	H
		83.3	What are the best medicines available. Will gene therapy ever be an option.		T2	P
		121.4	how enzyme replacement therapies, ASO, and gene therapies can help mito patients.		T2	CS
		125.3	There is no coordinated approach to developing new treatments, and more importantly in developing robust evidence for efficacy. If the NHS is to fund any new treatments this evidence base is required. Disease-modifying and disease-correcting therapies are required, and focus on gene therapy/ gene correction treatments should be a priority.		T2	H

		128.3	I would like to know what novel treatments can be used to treat irreversible vision loss in inherited optic neuropathies. In particular, what drugs and gene therapy strategies are being developed and what novel strategies can be developed towards this.		T2	H
1.07E+10	140	140.1	Can the missing or mutated genes be replaced? Can OPA gene be taken from a relative? Would replacing a mutated gene fix the damage caused to an optic nerve? Could gene replacement work if a child is a carrier, but not yet showing symptoms, and possibly reverse the mutation?		T2	CS
		141.8	Viability of gene therapies, and similar?		T2	P
		142.6	Is it likely in the future that gene therapy may be an option for those suffering with LHON in excess of 10 years?		T2	CS
		13.10	Is there any alternative medication that helps the symptoms?		T3	P
		17.6	Does acupuncture help.		T3	P
		43.4	It would be interesting to know if there is any evedence that any form of alternative therapy is proved to be beneficial. Uncertainty about how the condition might progress remains the greatest question. I am of an age where there are possible unrelated health issues developing but it is difficult know what affects what.		T3	P
		52.12	are there any alternative thereapies that are helpful? are there any alternative therapies should avoid?		T3	P
		70.8	Does oxygen therapy help a mito patient?		T3	P
		87.7	Role of alternative therapies in treatment of mitochondrial cardiomyopathies		T3	H
		91.6	Any alternative therapies that might be known about in the world to be helpful.		T3	P
		94.10	Alternative therapies to help with muscle pain, and other pain, would be good. Someone who knows a bit about mitochondrial disease, and who can offer tailored massage, or reflexology etc.		T3	P
		110.4	Outside the normal medication routes, possible alternative therapies, how these could possibly aid.		T3	CC
		110.7	possible natural remedies where conventional medicines are not helping as expected or causing other health issues		T3	CC
		115.6	Are there any alternative therapies out there that could help?		T3	CS
		143.6	Any Natural remedy treatments?		T3	P
		6.5	Can CBD oil be prescribed for specific mutations?		T4	P
		21.3	Would medicines such as CBD oil be effective to manage some of the symptoms of Mitochondrial disease?		T4	P
		44.4	Would Cannabis Oil have prolonged the life of my grandchildren?		T4	CS
		10.4	How much physio is safe?		T5	CS
		14.3	Would routine physiothearapy help with mobility and the ability to tolerate exercise?		T5	P
		24.4	Can physio be beneficial		T5	P
		127.6	methods to keep patients mobile are all good examples		T5	H
		17.3	CO 10 helps some but not others. why?		T6	P
		31.6	Are my daughter's cells absorbing enough of the medicines she takes (N1, N2, Q10) to make a difference? Can anything be done to improve this?		T6	CS

1.06E+10	37	37.1	Do different gene polymorphism require different treatment. Would different people require different treatments		T6	CS
		42.2	The availability of compounded medication		T6	H
		61.3	Development of generic and more specific treatments for the different genetic mutations and type of disease.		T6	CS
		98.5	Idebenone for non-Leber's patients		T6	H
		103.6	Detailed nutritional monitoring, diagnostics integrated with EHR and genetic and epigentic profile via personalised digit health analytics platform.		T6	H
		117.3	and phenotype so that care and support can be appropriately directed, and care needs anticipated.		T6	H
		122.3	what causes the variable response to idebenone and other free-radical scavengers in treating mictochondrial disease		T6	H
		31.1	Is it possible to repair damage to motor pathways caused by mitochondrial disease?		T7	CS
		76.6	Development of nano tech to identify specific areas being affected by mito and to fix or replace damaged cells. E.g hearing, repairing cells in the ears. Diabetes caused by mito, identify how to repair/replace damaged pancreas.		T7	P
		99.4	If no effective treatment is possible, then can we develop techniques (eg using stem cells) to restore RGCs/vision once optic atrophy has occurred?		T7	H
		141.6	Viability of stem cell therapies, and similar?		T7	P
		20.7	What do you do if you're having a 'mitochondrial episode' more information on mitochondria- like checks for diabetes, kidney.		T8	P
		21.4	Are pancreatic transplants available to treat the diabetes associated with Mitochondrial disease?		T8	P
1.06E+10	46	46.1	Should the cardiac manifestations of mt disease be managed like non-mt causes of the same pathology (eg HCM)		T8	H
		56.3	Does aggressive epilepsy treatment prevent cortical volume loss and slow the rate of dementia in 3243/MELAS? What is the optimum epilepsy treatment in POLG? Is there a role for vagus nerve stimulation in the treatment of mitochondrial epilepsy? Who with mito are safe to take valproate?		T8	H
1.06E+10	61	61.1	Development of treatments and to target specific elements like improving kidney function		T8	CS
		62.3	long-term effect of some drugs, eg advised i could take statins 2 protect my heart, but they made my muscles so weak. Better 2 keep moving, potter about indoors but use scooter when out		T8	P
1.07E+10	87	87.1	Characterisation of heart muscle disease		T8	H
		101.3	Another discussion that has come up on several occasions is the best way to control diabetes associated with mitochondrial disease. Could research address this and provide advice/guidance to patients?		T8	CS
		106.3	efficacy and use of different anti-seizure medications		T8	H
		135.4	Is any treatment available for any mitochondrial diseases? Does anybody apply arginine i.v. to a known mitochondrial patients with CVA and how fast they are able to get hold of it in a DGH setting? How useful is it in treating metabolic stroke?		T8	H

		4.2	Can we develop new drugs that target fundamental defects across a range of mitochondrial diseases?		T9	H
		81.4	What kind if treatments could interfere with the basic mechanisms of cell dysfunction and disease progression over time to delay, stop or reverse tissue dysfunction?		T9	H
		7.5	What are the best ways to treat the symptoms related to mitochondrial disease (including distress,		Y1	H
		17.7	PSYCHOLOGICAL help is completely non-existent but would be great help		Y1	P
		40.5	What is the impact of mitochondrial disease on your family? emotional, psychological?		Y1	H
		47.4	Counselling / psychological support for patients & their families especially if more than one member affected or if planning for children. Working alongside charities/groups supporting those living with visual impairment.		Y1	H
		48.3	psychological support		Y1	P
		51.4	Can parents of mito children have a go to advocate to expedite access to technology and equipment. We are currently seeking private psychiatric consultation because we have been begging for antidepressants for 4 yrs. My daughter describes herself as broken.		Y1	CS
		52.11	which psychological therapies are effective ?		Y1	P
		56.5	How can we best support people with depression?		Y1	H
		60.4	I would like to see more psychological support, I find that's a big barrier for [name] yes physically it's hard for her but her mental state is holding her back to do more in life and i find that harder than the physical obstacle.		Y1	CS
		69.7	How it can affect us mentally. As I really feel this is not addressed more		Y1	P
		90.8	Counselling services by people with a real understanding of the disease, to help people come to terms with what's happened to them.		Y1	CS
		91.7	Can psychological support help me cope? There doesn't seem to be anything much to help counselling wise, but if there were it would need to be by a counsellor trained in understanding mitochondrial disorders.		Y1	P
		93.7	psychological support.		Y1	H
		94.6	Psychological support would be much appreciated, as it is difficult to live with this disease, and remain positive all of the time.		Y1	P
		101.7	a frequent discussion on our online adult support group is the lack of psychological support for mitochondrial patients/families given that the condition can dramatically impact on daily life and leaves those affected feeling very isolated. If research could be done to show the extent of this problem in mito patients, could the data be used to improve access to support services and/or increase funding?		Y1	CS
		105.8	Many people diagnosed with LHON feel isolated and require professional help to come to terms with this life changing event.		Y1	CS

		127.4	There is so much to answer in this area but is very disease or patient specific. for example psychological support for young adults and families - does this improve quality of life; improved communication aids for those with sight or speaking difficulties; methods to keep patients mobile are all good examples		Y1	H
		145.6	How does one access appropriate psychological support pertinent to the disease? How can Families affect support? Can there be an MDR approach to the treatment options and psychological sopport that will be required to enable an individual to adapt to different life?		Y1	CS
		147.4	Anxiety & depression are common when coming to terms with a long term progressive condition (mainly adults) with no treatments or cures. Are psychological support methods or CBT helpful to this patient cohort?		Y1	H