Alex TLC consultation guidance

UK National Screening Committee

Newborn screening for metachromatic leukodystrophy

Section and / or page number in consultation that relates to info if you'd like to read about it	Online form question	What to expand on in your response and what criteria to refer to at the start of your response for each question.
	Please tell us if this condition has affected you, your family or your friends?	*We want to show how MLD can affect not only those that are affected themselves but also those who have a family member or loved one that is affected (spouse, sibling, parent, friend, community member, etc)* • *Explain how it has affected you or your loved one*
Page 8, para 3 Page 10, criterion 9 Page 13, para 3 Page 28 Q1 Page 46	**Do you have any comments on the evidence considered by the UK NSC in the review? For instance, was any important evidence missed?**	***Can state at the beginning of your response " this relates to Criterion 9 - Efficacy of treatment in the pre-symptomatic phase and Criterion 14 - Cost effectiveness of NBS for MLD"*** *We want to demonstrate how beneficial early diagnosis of MLD is to you and/or other family members, even if you/your loved one can't be treated at the moment.* *Please insert here your stories about:*

		• *How a lengthy diagnostic odyssey has affected you/ your family (including financially going to several doctors, hospital stays, etc)* • *How you felt when you received a diagnosis, was your anxiety lessened?* • *If you would have wanted to have your diagnosis from birth – what difference would this have made to you/your family?* • *Unwittingly passing on MLD to your children and how this makes you feel* • *Your experiences of pre-implantation genetic diagnosis (PGT) and the reasons why you chose this route* • *The cost of having MLD, both financially (medications, home adaptations, etc) and psychosocially the effect it has had on you, your loved one, family, children, etc. (burden of care, financial struggles, changes to work and school, impact on social life, etc)*
Page 10, criterion 9 Page 11, para 3 Page 58, para 5	**Do you have any comments on the evidence considered by the UK NSC in the review? For instance, was any important evidence missed?**	***Can state at the beginning of your response "this relates to Criterion 9 - Efficacy of treatment in the pre-symptomatic phase, Question 2, and Criterion 14 - Cost effectiveness of NBS for MLD"*** *We want to demonstrate that knowledge of an MLD diagnosis from birth offers significantly different outcomes than if MLD is diagnosed due to symptoms. And how that can also be cost effective vs all the costs involved in the care of an affected individual and the impact on their family* *Please insert here your stories about:*

		• *Transplants that have been successful because of early diagnosis identified through another family member* • *Transplants that were "just in time" due to clinical detection eg through your own persistence or any other reasons* • *Successful transplants where the patient is now an adult and how they are doing eg. in work, married, no further issues* • *Successful gene therapy (Libmeldy)* • *Transplants that have been unsuccessful as they were performed too late* • *A diagnosis that was too late for transplant and the consequences for you/your family* • *The cost of lifelong care, the impact it has on your and your loved one's lives*
Page 8, para 2 Page 13, para 1	**Do you have any comments on the discussion, conclusion or recommendation in the review?**	*We want to demonstrate that the incidence of MLD reported is higher than the number of patients that have been identified and that this is an unacceptable situation.* *We want to get across our worries that there are adults that are "lost" in the system as the condition initially causes behavioural changes that can mean relationship breakdown, isolation, homelessness, inability to access medical help.* *We also want to highlight the risks of not allowing reproductive choice for adults who do not know they have MLD.* *Please insert here your stories about:* • *How the behavioural problems caused by adult onset MLD caused relationship breakdown/isolation* • *Issues in accessing medical help*

		• *Issues in accessing a diagnosis* *Your feelings about unidentified adults with MLD passing on the gene to their children*
Page 14, para 1-4 Page 58, para 2	**Do you have any comments on the discussion, conclusion or recommendation in the review?**	***Can state at the beginning of your response "this relates to Criterion 9 - Efficacy of treatment in the pre-symptomatic phase"*** *We want to demonstrate the successes of treatments for MLD especially prior to symptom onset.* *You can state that:* • *BMT has been an option for MLD but gene therapy is now a good alternative option and it has been proven clinically effective thus being approved by NICE for treatment in the UK.* *Please insert here your stories about:* • *Childhood BMT* • *Adult BMT* • *Gene therapy – was your child unable to access a BMT as there wasn't a suitable donor and gene therapy was not available at the time* *Any other treatment you or your loved one have received for MLD that has been successful or helped*
Page 58	**Do you think screening should or should not be recommended? Why?**	*The NSC concluded that the volume and type of evidence related to newborn screening for MLD is currently insufficient to justify adding it to the NBS panel.* *Please insert here your thoughts on this decision – you might want to think about the following:*

		• *Benefits of early diagnosis* • *Screening is now happening in Illinois, US and Norway– how does this make you feel?* • *The impact it has made on your/your family's life because there was no screening when you/your loved one was born – has an MLD diagnosis affected your physical or mental health, ability to work, live in your home, education choices. How has the diagnosis affected siblings - have they experienced mental health issues such as survivor guilt, depression, has their education or personal life been affected?* • *The impact it would have made on your life if screening had been in place when you/your loved one was born*
	There could be many alternatives to a screening programme. How else do you think the NHS or the government could help people with the condition?	*We want to demonstrate that there is nothing else besides screening that would be able to help people with MLD as if they are not diagnosed before symptoms begin then they will be too late for treatment. Screening and thus early diagnosis for MLD is the only option to help people with the condition so they have a better chance of being eligible for treatment.*
	Do you have any other recommendations?	*If there is anything else you'd like to mention or address about newborn screening for MLD feel free to input that here.* *It could be a good place to put a final point saying that we believe MLD should be added to the newborn screening panel.*