Consultation Report

Topic details

Title of policy or policy statement:	Cholic acid and chenodeoxycholic acid for treating inborn errors of bile acid synthesis (all ages)
Programme of Care:	Women and Children
Clinical Reference Group:	Metabolic Disorders
URN:	1623/1696

1. Summary

This report summarises the outcome of a public consultation that was undertaken to test the policy proposal.

2. Background

Inborn errors of bile acid synthesis are a group of very rare conditions where the liver has difficulty making important substances called bile acids (such as cholic acid and chenodeoxycholic acid). Bile acids usually help to digest fats and aid vitamin absorption but, in people with inborn errors of bile acid synthesis, there is a problem with the proteins in the body that help with chemical reactions (enzymes needed for bile acid synthesis). Instead the liver makes too many unusual bile acids and substances known as metabolites (unfinished products of chemical reactions in the body that would usually be broken down). These substances can then build up in the liver and damage it. Inborn errors of bile acid synthesis can interfere with the body's ability to absorb enough of the fats and vitamins that it needs to be healthy, and can cause liver disease, cirrhosis (scarring of the liver), liver failure and death. In some cases, inborn errors of bile acid synthesis can cause progressive diseases of the central nervous system (affecting the brain and how people move).

There are many different types of inborn errors of bile acid synthesis. The timing of diagnosis, the symptoms, and outlook for the disease varies from person to person, and depends on the type that a patient has. These diseases are very rare; there are less than 65 patients in England known to be affected by them currently.

There are no curative treatments for these very debilitating conditions. Cholic acid and chenodeoxycholic acid, used singly and sometimes in combination are the only treatments for these diseases.

3. Publication of consultation

The policy was published and sign-posted on NHS England's website and was open to consultation feedback for a period of 30 days from 22nd February to 24th March 2019. Consultation comments have then been shared with the Policy Working Group to enable full consideration of feedback and to support a decision on whether any changes to the policy might be recommended.

Respondents were asked the following consultation questions:

- Has all the relevant evidence been taken into account?

- Does the impact assessment fairly reflect the likely activity, budget and service impact? If not, what is inaccurate?
- Does the policy proposition accurately describe the current patient pathway that patients experience? If not, what is different?
- Please provide any comments that you may have about the potential impact on equality and health inequalities which might arise as a result of the proposed changes that have been described?
- Are there any changes or additions you think need to made to this document, and why?

4. Results of consultation

There were five responses to the consultation. All were supportive of the policy. Only one change was proposed to the policy in relation to the stopping criteria. The policy working group considered that the policy stopping criteria are adequately defined and allow appropriate clinical judgement to be applied.

Responders commented:

'The medication is life changing and, in our case, has transformed his life.'

5. How have consultation responses been considered?

Responses have been carefully considered and noted in line with the following categories:

- Level 1: Incorporated into draft document immediately to improve accuracy or clarity
  There were no level 1 responses.
- Level 2: Issue has already been considered by the CRG in its development and therefore draft document requires no further change.
  There was one level 2 response .
- Level 3: Could result in a more substantial change, requiring further consideration by the CRG in its work programme and as part of the next iteration of the document
  There were no level 3 responses.
- Level 4: Falls outside of the scope of the specification and NHS England's direct commissioning responsibility
  There were no level 4 responses.

6. Has anything been changed in the policy as a result of the consultation?

No changes have been made to the policy proposition as a result of public consultation.

7. Are there any remaining concerns outstanding following the consultation that have not been resolved in the final policy proposal?

There are no remaining concerns outstanding following the consultation that have not been resolved in the final policy proposal.