

RECORD OF CONSUMER HEARINGS

Consumer meeting with Metabolic Dietary Disorders Association (Item 7.05)

The meeting covered the upcoming PBAC consideration of sapropterin for the treatment of patients with hyperphenylalaninaemia (HPA) caused by phenylketonuria (PKU). The following points provide a summary of the perspectives presented by the Metabolic Dietary Disorders Association (MDDA) to PBAC representatives:

- PKU affects the quality of life of patients and their families, impacting on a patient's cognitive function, mental health and social inclusion. The patients and their carers outlined the complex dietary requirements which are particularly difficult to manage in social situations, at school and at work.
- The patient representatives outlined that protecting brain function is the primary objective of any PKU treatment. High Phe levels can lead to irreversible neurological damage in babies and children, and subtle reversible effects on neurological function in adults.
- The patient representatives acknowledged that not all patients with PKU would respond to sapropterin, and those who do respond would still need to adhere to a highly restricted diet (although more liberal than without sapropterin). The patient representatives outlined that, in responsive patients, sapropterin would:
 - lead to improvements in neurological function, which may reduce fluctuations in mood, reduce anxiety and improve concentration. The patient representatives outlined that this may enable patients to study, maintain stable employment and healthy relationships, and aid compliance to diet;
 - minimise the health impacts associated with inadequate dietary intake of natural protein and nutrients which include impaired growth, osteoporosis, diabetes and obesity (as low Phe foods are generally high in calories and sugar);
 - minimise the fluctuations in Phe levels during periods of protein consumption or intercurrent illness (e.g. common cold or hayfever), which may improve neuropsychological outcomes;
 - enable more flexibility in a patient's diet which would facilitate eating in social situations such as at childcare and school, on school trips and at restaurants. It would also reduce worry and anxiety about diet;
 - reduce the need for special formula and complex multi-vitamin regimens. The special formula needs to be consumed at fixed intervals which can be intrusive on daily life; and
 - help patients maintain their blood Phe closer to the levels recommended in guidelines.
- The patient representatives highlighted that PKU is particularly difficult to manage during pre-conception and pregnancy. Lower blood Phe levels must be maintained to reduce the risk of deformities or miscarriage; for some patients this means they can only consume synthesised foods during this time. The patient representatives also outlined that sapropterin may enable some babies with PKU to be breastfed.
- The patient representatives considered that all patients with HPA due to PKU should have the opportunity to be tested for sapropterin responsiveness, regardless of age or Phe levels, as all patients have potential to benefit from sapropterin therapy.

- The patient representatives outlined that there is no distinction between patients whose blood Phe levels are well-controlled and those whose levels are poorly-controlled, with most patients cycling between the groups.