What is an Inherited Metabolic Disorder?

This toolkit aims to explain what an Inherited Metabolic Disorder (IMD) is and explore their underlying causes. This toolkit also aims to establish a connection to support groups for individuals living with or those who have family members living with these conditions. Information has also been made available in this toolkit for families striving for the most current valid treatment options.

What are Inherited Metabolic Disorders?

Inherited Metabolic Disorders are characterised by the inability to utilise or store energy. We have created this video which helps to explain what these conditions are and the effects they have on the body: https://www.metabolicsupportuk.org/inherited-metabolic-disorder/

How is an IMD inherited?

Primarily, IMDs are inherited and passed on through your parents’ genes; these genes carry mutations that code for enzymes which break down fats, proteins and carbohydrates. These genes may also code for transporter proteins which transport substances into cells of the body. Enzyme deficiency or inactivity may lead to a build-up of the enzyme’s by products or metabolites which may potentially be harmful to the body. There are multiple ways in which IMDs can be inherited which are represented below.

For further information on inheritance patterns, see the link below: https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns
Main centres of expertise within the United Kingdom
Finding a local specialist metabolic centre close to you may be a daunting task, therefore, Metabolic Support UK has compiled a list of centres! This list may help if you need a referral from your GP [see link below].
https://www.metabolicsupportuk.org/metabolic-centres/

Access to support groups
For some of the conditions we cover, there is an established patient organisation/support group available. We have developed good relations with these, and they are well-respected within the rare disease community. You can find a list of organisations here: https://www.metabolicsupportuk.org/where-to-get-support/ If there is a patient organisation available for your condition, we highly recommend getting in touch with them. Living with a rare condition can be isolating and information can be difficult to find. It is often said that the best experts are the patients and families themselves as they have experience of how the condition affects their day to day lives. We recommend building up a support network of relevant organisations, healthcare professionals and healthcare/social services to assist you. If you need some help to do this, please don’t hesitate to get in touch. Patient organisations/support groups will often have resources and events dedicated to your condition, and so they are ideal to compliment the support offered by Metabolic Support UK. You may also wish to connect with other people who are affected by the same condition as you via our Metabolic Connect service. You can learn more about this here: https://www.metabolicsupportuk.org/metabolic-connect/

Basic Treatment options
It is important to note that Inherited Metabolic Disorders affect individuals differently, even within the same family. Every condition has treatments that are aimed to manage individual symptoms and treatment plans that are tailored to you as an individual. For more advice on treatments for your specific condition, you should contact your metabolic consultant or your specialist.
There is no cure for the vast majority of IMDs. Some conditions can be managed by diet, others may require the avoidance of factors which can trigger symptoms, for example, with Medium-Chain acyl-CoA Dehydrogenase Deficiency [MCADD], it is advised to avoid prolonged periods of fasting as the body cannot break down fats for energy when glucose levels are low. Therefore, with conditions that effect fat metabolism this rule may apply. Other more severe conditions may require a multidisciplinary team of specialists and therapists who will assist in building a care plan and provide symptomatic and supportive care.
Emergency treatment regimens:
The British Inherited Metabolic Disease Group (BIMDG) provides emergency regimen guidelines for the IMD’s which require immediate treatment in cases of illness or other triggers. Your specialist will provide you with these guidelines and explain them to you when you are diagnosed and you will need to follow them in case of illness or fever, or other triggers, or if a child refuses food or is unable to keep food down due to vomiting for example. In some cases, you can follow your guidelines at home and symptoms will improve. If they do not, you will need to seek medical attention from your nearest hospital. The guidelines will include instructions on making up intravenous solutions and oral solutions in emergencies and aim to assist medical staff in your treatment. Your specific regimen will also have the contact details of your specialist and a direct line to the IMD department. These guidelines are applicable to most patients but there will be occasions when alternative management is appropriate.

Enzyme replacement therapy:
Enzyme replacement therapy [ERT] may be administered as a treatment option for some IMDs, primarily the Lysosomal Storage Disorders. This specialised therapy aims to replace the absent or deficient enzyme in the body via intravenous infusions. Once criteria for treatment has been met and this has been agreed as a valid treatment option, ERT can be administered at home with the aid of a multi-disciplinary team or in one of the local infusion centres. Each IMD has a specialised infusion centre, this would be identified with the patient once treatment criteria have been met.

Allogenic stem cell Transplants:
Allogenic stem cell transplants may be offered as a treatment regime. These transplants first involve administrating a dose of chemotherapy via intravenous infusion so that the immune system does not reject the new stem cells. The stem cells are then infused into the body in an effort to produce the absent or deficient enzymes. This specialised treatment option would be discussed by the metabolic specialist with the patient once criteria for this has been met and then a treatment centre could be identified.