Metabolism Project Ideas

1. Maple Syrup Urine Disease (MSUD) → Member #1
    a. Caused by a deficiency in the branched-chain alpha-keto acid dehydrogenase complex, which mainly affects the metabolism of branched-chain amino acids (leucine, isoleucine, and valine)
    b. Primary symptoms include poor feeding, vomiting, and lethargy, which can quickly devolve to severe neurological complications if left untreated.
    c. Management: Treatment involves dietary restrictions of the branched-chain amino acids. Similar to the management of the disease we learned about in lecture, galactosemia.

2. Fructose Intolerance (Hereditary Fructose Intolerance - HFI) → Member #2
    a. Fructose intolerance is caused by a deficiency in aldolase B enzyme. This enzyme is necessary for fructose metabolism.
    b. Symptoms of HFI include hypoglycemia, jaundice, liver failure, and vomiting upon intake of fructose due to the inability to metabolize fructose.
    c. People with HFI cannot digest fructose, so they must avoid foods containing fructose, sorbitol and sucrose. Examples are fruit, honey, and some vegetables.

3. Gout → Member #3
    a. Gout is a form of inflammatory arthritis that is caused by high levels of uric acid in the blood. This often leads to the formation of urate crystals in joints, which in result, causes acute pain and swelling.
    b. The acid crystallizes and the crystals remain in the joints, tendons, and surrounding tissues. The presence of the crystals in the joint fluids are a way to diagnose this disease.
    c. Connection to gout is seen in individuals with high blood pressure, high blood sugar levels, excess fats, abnormal cholesterol levels, and closely related to individuals with insulin resistance - leading to an accumulation of uric acid in the body.

4. Gaucher disease → Member #4
    a. Gaucher disease is inherited and caused by a deficiency in the glucocerebrosidase enzyme. The enzyme's role is to break down glucocerebroside fats, so people with Gaucher's disease cannot properly break down glucocerebrosides. This causes accumulation of fats in the liver, spleen, and bone marrow.
    b. Symptoms of gaucher disease include pain, fatigue, jaundice, bone damage, anemia, and death.

c. Symptom severity vaires greatly among people with Gaucher disease, but those with more severe symptoms can be treated with enzyme replacmenet therapy or medicines to rebuild lost bone density.