1. **Cystic Fibrosis** – This results in a malformed ion channel, causing thick secretions in the digestive and respiratory systems. This then causes intestinal obstruction, poor growth, abnormal bowel movements, coughing, wheezing, frequent respiratory tract infections, and fat-soluble vitamin deficiencies. Cystic Fibrosis can be managed with close monitoring, enzyme and vitamin supplements, and preventative therapies. This disorder is more frequent in people of European descent. The diagram below shows the specific organs that are affected by this disease

Airways	Liver	Pancreas	Small Intestine	Reproductive Tract	Skin
Clogging and infection of bronchial passages impede breathing. Repeated infections destroy the lungs. Lung disease accounts for most deaths from cystic fibrosis.	Plugging of bile ducts impedes digestion and disrupts liver function in about 5% of patients	Occlusion of ducts prevents the pancreas from delivering critical digestive enzymes to the bowel in 65% of patients. Diabetes can result as well.	Obstruction of the gut by thick stool necessitates surgery in about 10% of newborns	Absence of the ducts, such as the vas deferans, renders 95% of males infertile. Occasionally, women are made infertile by a dense plug of mucus that blocks sperm from entering the uterus.	Malfunctioning of sweat glands causes perspiration to contain excessive salt. Measurement of Chloride in sweat is a mainstay of diagnosis.

2. **12 types of Fatty Acid Oxidation disorders** – A category of inherited metabolic conditions leading to the accumulation of fatty acids and a decrease in cell energy metabolism. These problems arise because the body is unable to properly break down fat to use as energy. If left untreated hypoglycemia (A lack of sugar in the blood), failure to thrive, and/or death can result. This disorder can be treated with supplemental carnitine, a low-fat diet, and home glucose (blood sugar) monitoring.

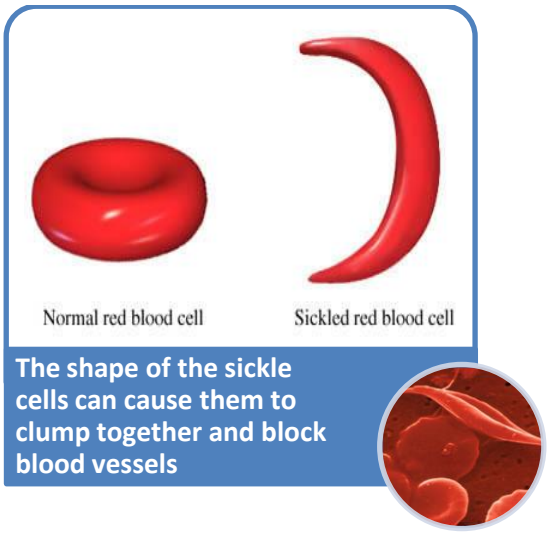
3. **Biotinidase Deficiency** – A disorder where the individual is unable to properly recycle B vitamins. It can be treated with Biotin replacement therapy. If left untreated it can result in seizures, hypotonia (reduced muscle

tone), breathing problems, skin rashes, hair loss, developmental delays, speech problems, ataxia and vision and hearing problems.

4. **Congenital Adrenal Hyperplasia** – This is an error in steroid biosynthesis where the individual is unable to produce enough Cortisol and/or Aldosterone. These hormones are produced in the Adrenal glands located on top of the kidneys:

Congenital Adrenal Hyperplasia results in excess androgen production (including testosterone), salt wasting (over-excretion of salt) in those who can't produce Aldosterone, hypoglycemia (low blood sugar), metabolic acidosis, precocious puberty, dehydration, and shock. It can be treated with hormone replacing therapy.

1. **Sickle Cell Disease** – This disease normally results in sickle shaped red blood cells due to an abnormal hemoglobin protein. It causes anemia, interferes with development, makes children more prone to infection, and causes painful crises with infarctions (clotting) of multiple organ systems. The treatment is supportive and includes supplemental iron, and blood transfusions. This disorder is more frequent in people of African descent.

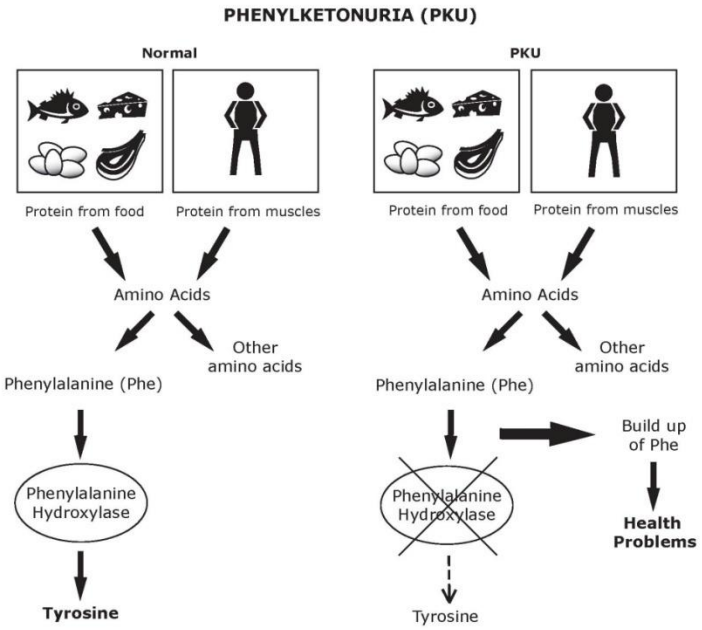


2. **Hypothyroidism** – A lack of thyroid hormone resulting in a lowered basal metabolism. Since this hormone is necessary for normal growth and metabolism, the child must be treated with hormone replacement therapy.

3. **Galactosemia** – This results in an inability to convert galactose to glucose. It can be treated by the exclusion of galactose and lactose from the diet. If left untreated it can result in vomiting, diarrhea, cataracts, failure to grow, mental retardation, and death.

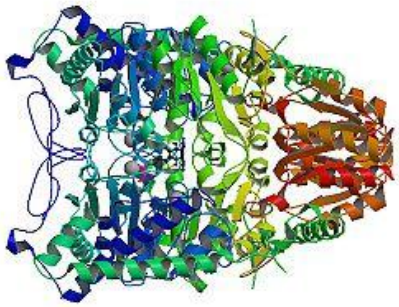
4. **11 different Organic Acid disorders** – A category of inherited metabolic conditions. If left untreated brain damage and/or death results. They can usually be treated with low protein diets and, in some cases, supplemental vitamins or enzymes.

5. **Phenylketonuria** – A defective enzyme makes the child unable to oxidize phenylalanine to tyrosine. If left untreated the buildup of tyrosine in the blood causes brain damage and other symptoms. This disorder can be treated with a diet low in phenylalanine (a protein). The diagram below maps out this relationship:



6. **Homocystinuria** – This is a defect in an enzyme used to break down or process methionine (a protein). The build up of methionine in the body causes developmental delay, ectopia lentis(displacement of the lens in the eye), and Osteoporosis. Homocystinuria can be treated with a diet lacking methionine, medication, folic acid, and vitamins B_{12} and B_6.

7. **Maple Syrup Urine Disease** – People with this disorder lack the enzyme necessary to catabolize (break down) leucine, isoleucine, and valine (all of these are proteins). The affected enzyme looks like this:

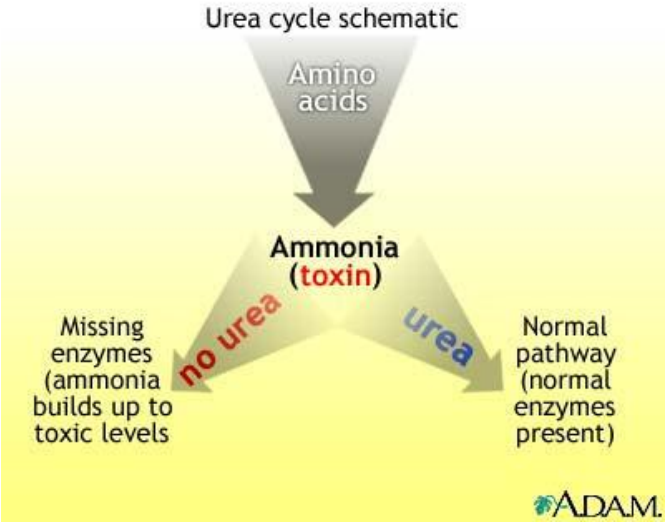


Treatment of Maple Syrup Urine disease involves a diet lacking in the proteins broken down by the above enzyme, and supplemental vitamin B_1. If left untreated it can result in poor feeding, hypoglycemia (low blood sugar), mental and neurological deficits, seizures, coma, ketoacidosis, and death.

8. **2 Types of Tyrosinemia** – This is a group of inherited disorders affecting tyrosine (a protein) catabolism (breakdown). It normally affects the liver, kidneys and nervous system, and can be treated with a restricted diet.

9. **Glutathione Synthetase deficiency** – This disorder results in a lack of Glutathione (a protein antioxidant). The lack of this protein leaves the cells open to damage by oxidation (The same process that turns apples brown). The effects can range from mild to severe depending on what types of tissues are affected. In the most severe form the condition results in acidosis (acidic blood). This can be treated with sodium bicarbonate or citrate (both are buffers), blood transfusions, and vitamins E and C.

10. **3 different Urea Cycle Disorders** – This category of disorders is caused by a deficiency of enzymes in the urea cycle.

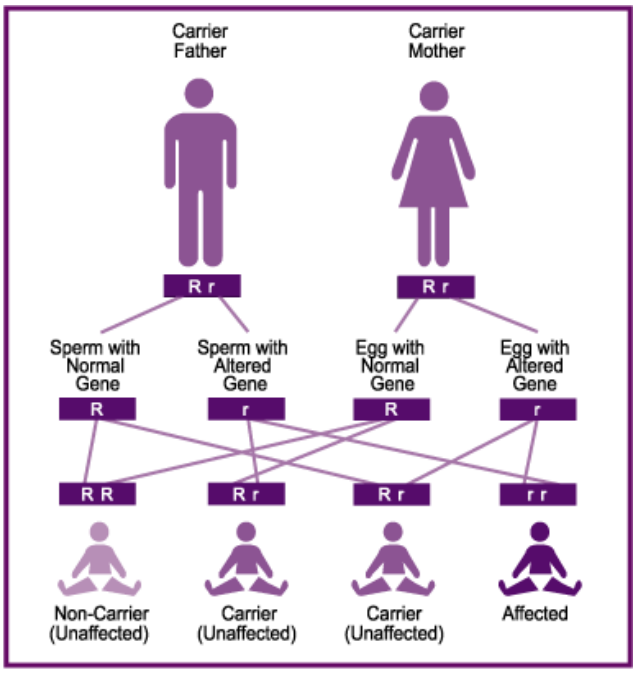


The disorder then leads to a build up of ammonia in the body. This buildup then results in hyperammonemia, poor feeding , coma, muscle tone abnormalities, seizures and death. It can be treated with a special medical formula, arginine supplements, high caloric intake, and medication.

For more complete information on these disorders and the state testing policies, take a look at the state website: Illinois Department of Public Health

Most of these disorders are relatively rare. This is in part because a child can only be born with one of these disorders if:

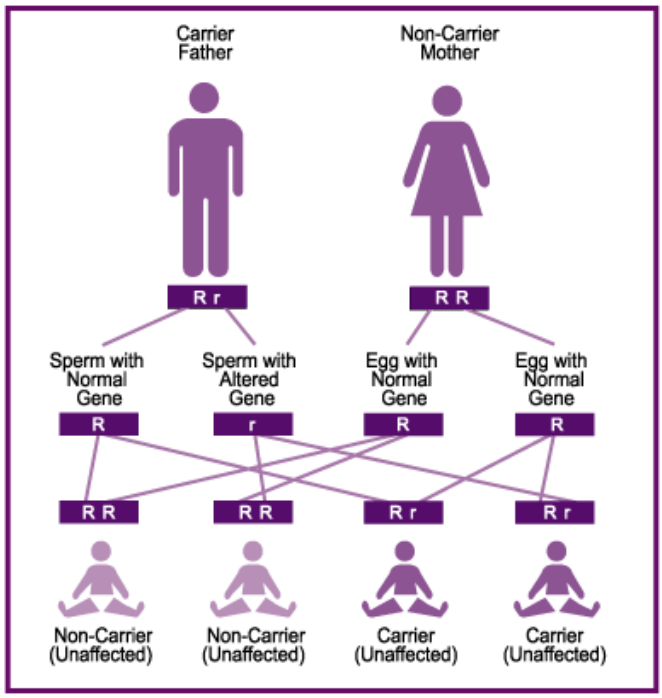
1. Both the mother and father have one normal copy of the gene in question and one defective copy of the same gene – In this case each child they have has a 25% chance of having the disorder. This scenario can be diagrammed like so:



2. Both the mother and father have the disorder (each parent has two defective copies of the gene in question) – In this case each child they have has a 100% chance of having the disorder.

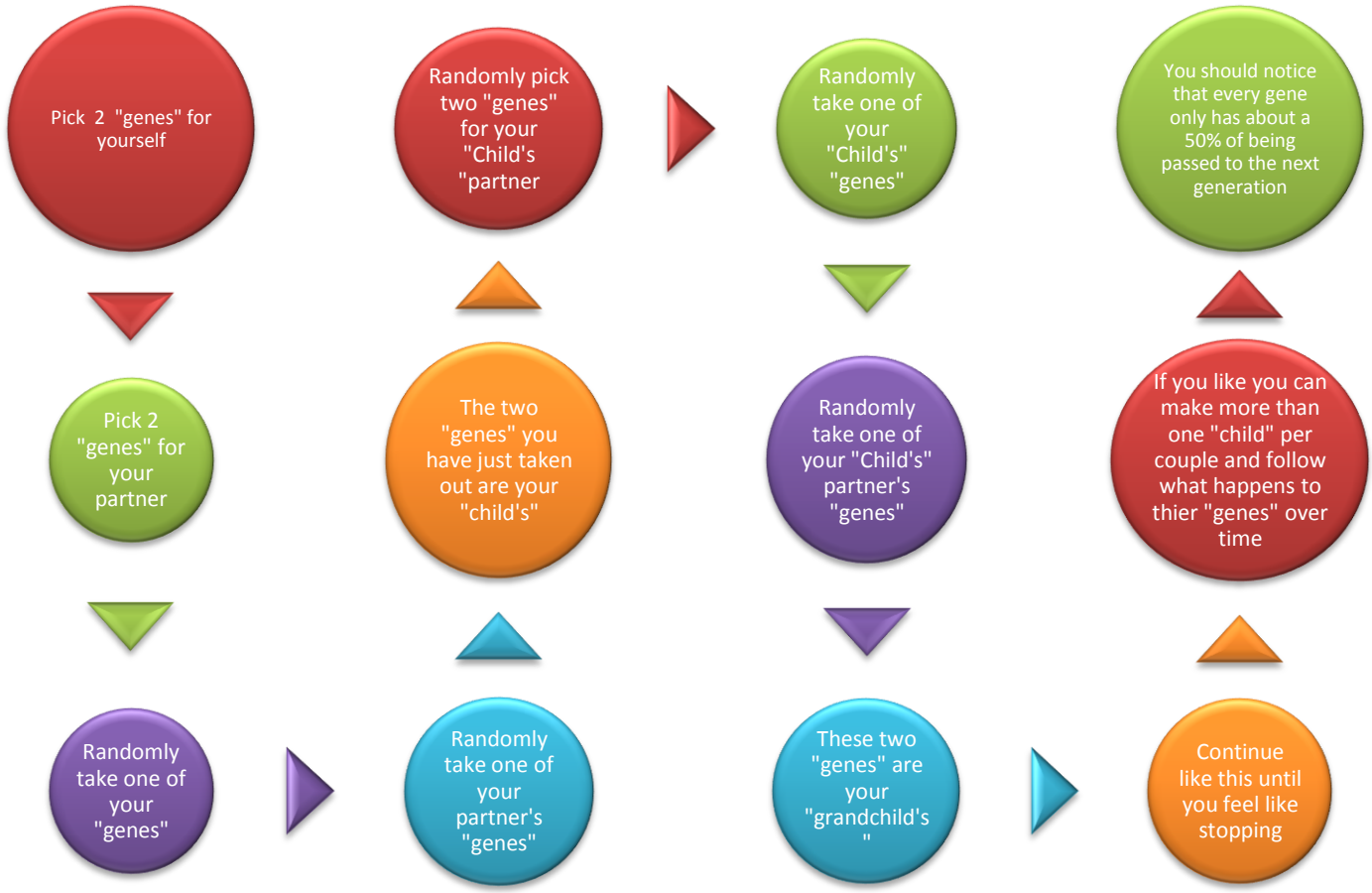
3. Either the mother or father have the disorder (two defective genes), and the other partner has one normal and one defective version of the same gene – In this case each child they have has a 50% chance of having the disorder.

4. However, if one parent carries the defective gene, or is even affected by the disorder, and the other parent has two normal copies of the same gene, it is impossible for any of the children to actually be affected by it because they are guaranteed to get at least one normal copy of the gene. This can be shown with a diagram:



Since a child can only have one of these disorders if both parents have a defective version of the same gene, it is important to know the health history of both your family and that of your partner's. However, because these defective genes are fairly rare, they can lurk undetected for many, many generations before someone is actually born with the disorder.

To better understand how these genes are passed from one generation to the next. Have one color represent the defective gene, and have another color represent the normal gene. Choose a "genotype" for yourself by selecting two strings of either color. Then choose a "genotype" for your partner by selecting two stings of either color. To make your "child's genotype" randomly take one string from your set of 2, and one string from you partners set of 2. Try doing this a couple of times to see the different types of "children" you can have. Also try it with different "genotypes" for yourself and your partner. You can also use this same principle to map out a couple generations worth of gene passing.



Newborn Hearing Screening

Every year in Illinois, up to 500 babies are born with hearing loss. Also, catching hearing difficulties early is important to that the affected child can receive the help they need from an early age. For these reasons, children born in Illinois are screened at birth for certain types of deafness or hearing difficulties. As with all the other tests of this page, the hearing screening test is only meant for screening, and a positive result simply means that further testing is needed. Some babies will not pass the first test but be fine by a later one, while others may pass the first test and lose their hearing later. This is why it is important to listen to the recommendations of your doctor regarding further testing, as well as to know what the age-appropriate behavior is for your child with regards to noise.