



Alkaptonuria: A Fact Sheet for Patients

What is Alkaptonuria?

Alkaptonuria (pronounced al-KAP-tone-ur-ee-a) is a rare disease that can cause serious damage to many different parts of the body. Alkaptonuria usually causes painful joint disease involving the back, hips, and knees. It also can cause bones to break easily, dangerous hardening of heart valves, formation of prostate and kidney stones, and the appearance of dark spots in the white part of the eyes and on the ear.

Alkaptonuria is a genetic disease. It is present from birth, but may not produce symptoms until early to mid-adulthood. Although most people with Alkaptonuria are able to live normal life spans, the disease is often painful and disabling later in life.

Who is at risk for Alkaptonuria?

People all over the world can have Alkaptonuria. However, doctors know the disease occurs more commonly in some groups of people, including people from Czechoslovakia or the Dominican Republic and their descendants.

How might you know if you have Alkaptonuria?

Sometimes, dark urine stains in diapers can indicate that a baby has Alkaptonuria. Usually, however, there are no signs until adulthood. In adults, the clearest signs of Alkaptonuria are:

- Painful joints, especially in the back, hips, and knees.
- Unusual brown or dark blue coloring on the ear.
- Dark spots in the whites of the eye.
- Urine that turns dark if it is exposed to air for several hours.
- Dark bones, which may be noticed only by doctors during operations.

How do doctors diagnose Alkaptonuria?

Doctors usually can diagnose the disease by looking at a combination of factors: damage to joints (especially of the back and hips), unusual dark color in the ear or eye, family history, and some medical tests. There is no approved treatment or cure for the disease.

What research is being done on Alkaptonuria?

The National Institutes of Health (NIH) is one of the few places in the world currently studying Alkaptonuria. This research should help improve the understanding of the disease and help scientists find ways to treat it. NIH doctors invite people with Alkaptonuria to consider taking part in a new research study. Please call for more information.

Please call for more information
1-800-411-1222 (USA)

Se habla español

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