



The Breakdown: Endocrine Issues in FA

What is the endocrine system?

The endocrine system produces hormones that allow our bodies to develop and function. This system consists of glands in the head, neck and abdomen that release many different types of hormones into the bloodstream. These hormones perform a variety of functions in the body, from regulating blood sugar levels to triggering physical changes during puberty. Think about it this way: endocrine cells make a hormone, or message. These “messages” are carried in the bloodstream to other cells, telling the body to grow taller, for example, or to go through puberty.

How is the endocrine system affected in FA?

The DNA damage caused by FA leads to death of some endocrine cells. This loss of endocrine cells results in lower hormone levels. About 80% of children and adults with FA have an endocrine abnormality. These abnormalities can affect the body in a variety of ways:

- Short stature
- Poor weight gain, or overweight status
- Abnormal glucose with low insulin secretion (can contribute to poor

weight gain or diabetes)

- Hypothyroidism (low levels of thyroid hormone, causing poor height growth, delayed puberty, irregular periods, difficulty becoming pregnant)
- Early or late puberty, underactive testes or ovaries, infertility
- Growth hormone (GH) deficiency
- Low bone mineral density

How can these issues be managed?

In FA we talk a lot about comprehensive care, and that is because FA is a disease that affects multiple systems in the body. For this reason, it is important to involve an endocrinologist or pediatric endocrinologist early, a dietician, and for females a gynecologist or reproductive endocrinologist. This team should work in close collaboration with other FA specialists to provide comprehensive care.

According to Dr. Susan Rose, MD, endocrinologist and FA specialist (Cincinnati Children’s Hospital Medical Center/University of Cincinnati), people with FA should have **annual growth screenings** to track height and weight, identify and treat any causes for poor growth, assess the thyroid and treat any deficiencies of thyroid hormone,

vitamin D, GH, or pubertal hormone. **Maintaining a healthy diet and exercise** is important and a **dietician should assess the person’s nutritional intake**. If a child with FA is short, s/ he should be **screened for growth hormone deficiency** (GHD). Growth hormone therapy may be used to treat GHD, though there is no current consensus on safety of this therapy in FA. After age 10 years (or younger if puberty started early), an **annual puberty screening** is recommended to assess hormone levels and to then determine whether treatment is needed for either early or delayed puberty. Finally, after bone marrow transplant or after age 16 years (whichever comes sooner), an annual bone screening to check bone mineral density is recommended. ■

For a full, detailed list of endocrine screening recommendations for patients with FA, see p. 149 in the Guidelines for Diagnosis and Management. Chapter 7 in this book is dedicated entirely to the endocrine system in FA. Also, check out the 2014 Fall Family Newsletter (p. 6) for another detailed summary on endocrine issues (available on our website: www.fanconi.org).