

Highlights from Camp Sunshine Presentations

Camp Sunshine attendees were treated to comprehensive presentations from Jill Huppert, MD, MPH, Cincinnati Children's Hospital, on gynecologic issues; Sarah Jane Schwarzenberg, MD, University of Minnesota, on gastrointestinal, liver and nutritional problems in FA patients and Susan Rose, MD, Cincinnati Children's Hospital, on endocrine problems in FA. All three presenters contributed to or were the primary authors of chapters dealing with these subjects in our 2008 *Guidelines for Diagnosis and Management Handbook*. The reader is referred to the *Guidelines* for detailed information on the above subjects. Some brief observations from the Camp Sunshine presentations include the following:

Gynecological Issues

- The median age for cervical cancer in FA women is 25, and the median age for vulvar cancer is 27. FA females are at extraordinarily high risk for gynecological cancers.
- The human papillomavirus (HPV) vaccine protects against HPV subtypes 16 and 18. These two subtypes are implicated in 70% of cervical cancers and 50% of vulvar cancers. FA girls between the ages of 9 and 26 are strongly advised to undergo vaccination.
- FA patients should abstain from sexual activity or greatly limit the number of sexual partners, insist on condoms, and abstain from oral sex, which is implicated in cancers of the throat in the non-FA population.
- A colposcopy allows a physician to magnify the vulvar area by ten to one hundred times. To prevent unnecessary biopsies, colposcopy should only be used if there are visible lesions.

Gastrointestinal, liver and nutritional problems

- FA children frequently have poor oral intake, nausea, pain or diarrhea. Dr. Schwarzenberg detailed the components of a thorough exam to diagnose the problem, while emphasizing that *radiographic (x-ray) imaging should be avoided, if possible*. Appropriate therapies for various symptoms were reviewed in detail.
- Twenty-two percent of FA children are underweight for their height. Appetite stimulants can have worrisome side effects and lose effectiveness when discontinued.
- Some FA children can benefit from supplemental feeds, which are most effective when given through the GI tract. The goal is to enable children to reach their genetic potential and give them sufficient energy to meet daily demands, plus a reserve in case of illness.
- Androgens can cause liver adenomas, carcinomas and liver lakes. Liver abnormalities must be thoroughly evaluated prior to transplant. Liver lakes persist after discontinuing androgens and can complicate transplantation.
- FA patients should have diets high in vegetables and fruits with anti-oxidant properties, believed to reduce the risk of some cancers. Individual vitamins cannot reproduce the effectiveness of foods and can actually be toxic in high dosages.

Endocrine abnormalities

- Four out of five FA patients have some level of endocrine abnormality.
- Two-thirds of FA patients have mild abnormalities in thyroid function. Seven of eight patients treated with thyroid hormone achieved an improved growth rate.
- DEXA scans (measurement of bone density) are not recommended for FA children prior to transplant, but are important post-transplant. DEXA scans adjust for age but not height, so FA patients sometimes inaccurately appear to have abnormally low bone density. FA patients short for their age need a “z score,” which takes height into account. Eighteen FA pediatric patients evaluated by “z score” were, as a group, in the normal range.
- Half of FA children have impaired glucose tolerance, and 8% are diabetic. Most have normal glucose while fasting, but demonstrate an abnormally slow release of insulin after eating. *Glucose tolerance should be measured one to two hours after eating.* Concentrated sugars should be avoided, as they greatly increase the demand for insulin.