# Gynecologic issues in CdLS

Adolescents and adult women with CdLS have the same gynecologic issues as all women. In some cases their symptoms can be difficult to diagnose and in other cases routine symptoms can be quite burdensome.

In adolescence, many of the concerns revolve around the menstrual cycle. The first menstrual period (menarche) typically occurs between 11 to 14 years of age. Menses may be irregular, and sometimes heavy for the first year to 18 months, but then generally settle into regular cycles.
Some young adults only start to menstruate in their late teens and about 20% of women with CdLS never menstruate.

Irregular menses are not of concern, unless they occur less often than every 3 months. In this case, treatment with progesterone (eg Provera) every three months should be considered to bring on the menstrual cycle and protect the uterus from overgrowth of the uterine lining. If menstrual cycles remain prolonged or heavy, treatment with the birth control pill (OCP) is usually effective. Progesterone only treatment is another option, but takes longer to be effective.

Pain from menstrual cramps is also an issue. In some girls with CdLS, self injurious behavior can occur as an expression of the pain. Nonsteriodal anti-inflammatory medications such as Motrin or Anaprox, known as NSAIDs, are often effective. If these don't work, then the OCP is usually effective.

Many families worry that the behavioral changes they see in their daughters are an expression of premenstrual syndrome (PMS). PMS is defined as recurrent psychological and/or physical symptoms occurring in the second half of the menstrual cycle. Symptoms can include mood swings, irritability, a state of excitement, sleep disturbance, and headaches. It is important to keep a daily calendar to determined whether symptoms fall in the typical time frame for PMS. There are a variety of treatment options for PMS. Spironolactone, Vitamin B6 and calcium supplementation can help improve mood. NSAIDs are the first choice for pain therapy. The OCP will suppress ovulation, and can be another helpful treatment. In some cases, treatment with drugs directly targeting the behavior problem is necessary. This is best managed by a psychologist or psychiatrist on an individual basis.

Another concern that many caretakers have is dealing with the menstrual cycle and wanting to avoid bleeding completely. Certainly hysterectomy is a definitive solution but this should not be the first choice. The OCP can be given continuously to suppress the menstrual cycle. Depo- Provera, a long acting progesterone, can be administered by injection every three months. This drug is not the best choice in individuals with limited

mobility as it causes bone thinning. Mirena is an IUD which contains progesterone. It will often suppress menses but most likely would need to be placed under anesthesia. Endometrial ablation is an operative procedure which involves cauterizing the lining of the uterus to prevent menstrual bleeding. The advantage of this procedure is that major surgery is avoided but it is not always effective long term. Sometimes hysterectomy is the only feasible option. When this is performed the ovaries are generally preserved, so that they can provide hormones which benefit the bones and other organs.

Contraception is another concern for the families of young women with CdLS. The methods already discussed, namely OCP, Depo-Provera, and Mirena IUD are all good options. Choices should be made based on the individual. The OCP comes in a weekly patch and now in a liquid form to be made available to females who cannot swallow pills. It will decrease menstrual flow and help prevent cramping. Depo-Provera can have a side benefit of reducing seizure activity.

Gynecologic examinations and PAP tests are recommended in asymptomatic women from age 21. If results are normal, the PAP test is only recommended every three years. In some individuals, an exam under anesthesia is needed in order to be able to do an adequate examination. This can be coordinated with other procedures to avoid multiple separate anesthetics. Pelvic ultrasound can be a diagnostic alternative in some cases.