Seckle Syndrome

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Seckel Syndrome is a rare (<1:10,000) constellation of malformations, presumably inherited as an autosomal-recessive trait. The primary characteristic features of Seckel Syndrome (SS) include; severe intrauterine growth restriction, microcephaly, orofacial dysmorphology with characteristic "bird-headed" appearance, and mental retardation.

Case Report
The patient is a 43 year old gravida 5, para 3, with a history of a child diagnosed with SS. The effected child died at the age of three years secondary to respiratory complications sustained under general anesthesia during a minor surgical procedure.

The patient presented for ultrasound evaluation and genetic amniocentesis at 15.0 weeks gestation by menstrual history. Ultrasound demonstrated a normal appearing female fetus. Fetal biometry was consistent with 15.0 weeks gestation, with all parameters equal to the mean for gestational age. Menstrual dates were therefore confirmed as accurate. Genetic amniocentesis was performed without complications. In light of the patient's history of an SS effected child, repeat ultrasound evaluation were recommended to monitor fetal growth.

Chromosomal analysis showed a normal 46,XX karyotype. Subsequent ultrasound examinations are summarized below.

19.0 weeks

Biometry
Head circumference = 13.9 cm (16.8 weeks) >2SD below the mean for gestational age. Abdominal circumference and femur length within normal ranges.

Abnormal Anatomy
No gross anatomical abnormalities noted.

22.0 weeks

Biometry
Head circumference = 15.9 cm (18.7 weeks), >2.8SD below the mean for gestational age. Abdominal circumference = 15.1 cm (20.0 weeks), at the lower limit of the normal range, Femur length = 3.3 cm (20.8 weeks), at the lower limit of normal range.

Estimated Fetal Weight = 12th percentile.

Abnormal Anatomy
Cisterna magna appeared prominent at 8mm in width, splaying of cerebellar hemispheres, cerebellar vermis not identified, abnormal profile with receding forehead and micrognathia

At this point, probable recurrent Seckel Syndrome was diagnosed, and the patient counseled. Patient elected to continue pregnancy.
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Figure 1: Facial profile at 22 weeks fetal age: receding forehead and micrognathia.

Figure 2: At 22 weeks fetal age: Transverse view of the posterior fossa showing absence of cerebellar vermis.

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27.0 weeks
Biometry
Head circumference = 19.8 cm (21.9 weeks), below 5th percentile
Abdominal Circumference = 19.9 cm (24.4 weeks), below 5th percentile
Femur Length = 3.9 cm (23.1 weeks), below 5th percentile
Humerus Length = 3.1 cm, below 5th percentile
Estimated Fetal Weight = 9th percentile

Abnormal Anatomy
Enlarged Cisterna Magna (14mm), splayed cerebellar hemispheres, unilateral pyelectasis = 6mm in AP diameter, pronounced abnormal profile

34.3 weeks
Biometry
Head Circumference = 23.5 cm (25.4 weeks), below 5th percentile
Abdominal Circumference = 25.1 cm (29.3 weeks), below 5th percentile
Femur Length = 4.9 cm (27.2 weeks), below 5th percentile
Humerus, Radius, Tibia, Fibula measurements all below 5th percentile bilaterally
Estimated Fetal Weight = 7th percentile

Abnormal Anatomy
Abnormal facial profile with sloped forehead, micrognathia, and large beaked nose, Cerebellar vermis hypoplasia, unilateral hydronephrosis (AP renal pelvis measurement = 1.4 cm), suspected agenesis of corpus callosum
A female infant was delivered by vaginal delivery at 38 weeks gestation. The infant weighed 1163 grams. APGAR scores were 8 (1 min.) and 9 (5 min.). Seckel syndrome confirmed at birth. Child currently living at home.

**Discussion**

Seckel syndrome is a form of primordial dwarfism with a set of primary diagnostic criteria, however, there are also several secondary characteristics that are less well defined. There appear to be several sub-sets of this type of dwarfism, and the literature reports some controversy in the application of this diagnosis. One author reports three types of Seckel-like syndrome. Another report guards against the over-diagnosis of this syndrome, and states that there is a group of microcephalic dwarfism variants which have yet to be clearly defined. The syndrome is assumed to have an autosomal-recessive pattern of inheritance. Advances in molecular genetics may soon characterize a specific chromosomal aberration that may help delineate true Seckel syndrome from other similar microcephalic dwarfs.

The majority of the literature concerns the postnatal diagnosis of the syndrome, but there are several articles describing the prenatal sonographic diagnosis of SS. The primary diagnostic features are; severe intrauterine growth restriction, microcephaly, characteristic "bird-like" facies, and mental retardation. With the exception of mental retardation, the other primary characteristics are well suited for sonographic diagnosis.

One author describes a technique for evaluating the palpebral fissure slant angle to help identify...
fetuses with Seckel syndrome. Others describe techniques of fetal face evaluation and the detection of several fetal abnormalities including SS.

Serial ultrasound examinations to evaluate fetal growth have been shown to be very useful in detecting the severe, symmetric growth restriction involved in this syndrome. Children affected with SS can live for an extended period of time, and often have profound mental and physical deficits. In patients with a history of SS pregnancies, serial ultrasound examinations for fetal growth, and a careful evaluation of the fetal face and cranial anatomy can help identify recurrent cases of this often heartbreaking disease.

References:

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