



Fetal Genu Recurvatum: A Case Series

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Abstract Genu Recurvatum is a rare congenital anomaly involving the knees. Most of the cases are sporadic, and if isolated, carry a good prognosis. Syndromic association tend to be present, and if so, the diagnosis becomes important. Early diagnosis and counseling remain the key as the couple can make an early decision on the pregnancy. Proper counseling would also help the couple in knowing what to expect postnatally. One, or maybe more than one, surgeries may be required for its correction and the gait may never be completely normal. We present a case series diagnosed by ultrasound at our center between January 2014 and 2020. Through this concise article, we aim to give a key to antenatal diagnosis and postnatal management of this rare condition.

Keyword Genu Recurvatum · Larsens syndrome · Arthrogryposis · Camel girl · Elher Danlos syndrome

Genu is the Latin word for the knee. Recurvatum (*recurvō*: Latin) means bent backwards. Genu recurvatum is also called knee hyperextension and back knee. In this condition there is excessive extension in the tibiofemoral joint. This deformity is more common in females and people with familial ligamentous laxity.

Deformities of the knee are classified on the basis of the tibiofemoral angle (Fig. 1).

Historically, the first case largely identified, dates back to the nineteenth century, Ella Harper, the Camel Girl (Fig. 2). She had a preference to walk on all four limbs,

hence the name Camel. She became famous as part of various Circuses. Also known as ‘freaks’, congenital anomalies were not something well understood or accepted in that era.

Case discussion

1. A 36 year old primigravida, IVF conception, had attended our center for a routine anomaly scan at 29 weeks of gestation. The NT scan done elsewhere was normal. Ultrasound showed abnormal leg position, Genu Recurvatum in both legs, bilateral talipes, right lateral ventriculomegaly of 13 mm and no movement of the legs during the examination of 30 minutes. A diagnosis of distal arthrogryposis was made.
2. A 27 year old, G3P2 at 19 weeks and 6 days gestation attended for a second trimester screening. First trimester screening had not been done. Ultrasound showed increased nuchal fold (measuring 7.9 mm) and features consistent with Arthrogryposis i.e. Bilateral club foot, fixed flexion deformity of both wrists and genu recurvatum of the right leg.
3. A 30 year old, G2P0A1 attended the center for her anomaly scan at 21 weeks and 3 days. She had a normal first trimester screening. On examination, the fetus showed bilateral Genu Recurvatum and rocker bottom foot, overcrowding of digits and clenched fists. A syndromic association was thought of.
4. A 21 year old primigravida, at 19 weeks of gestation for anomaly scan, showed isolated bilateral genu recurvatum. No other anomalies were found. The first trimester combined risk was low.

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Fig. 1 Classification of deformities of genu and types of Genu recurvatum. The three types of Genu Recurvatum are also the grades of it. Grade 1 being hyperextension of the knee (mild form), Grade 2 anterior subluxation of the tibia (mild requires non-surgical corrections), Grade 3 anterior dislocation of the tibia (severe form, surgical correction needed)

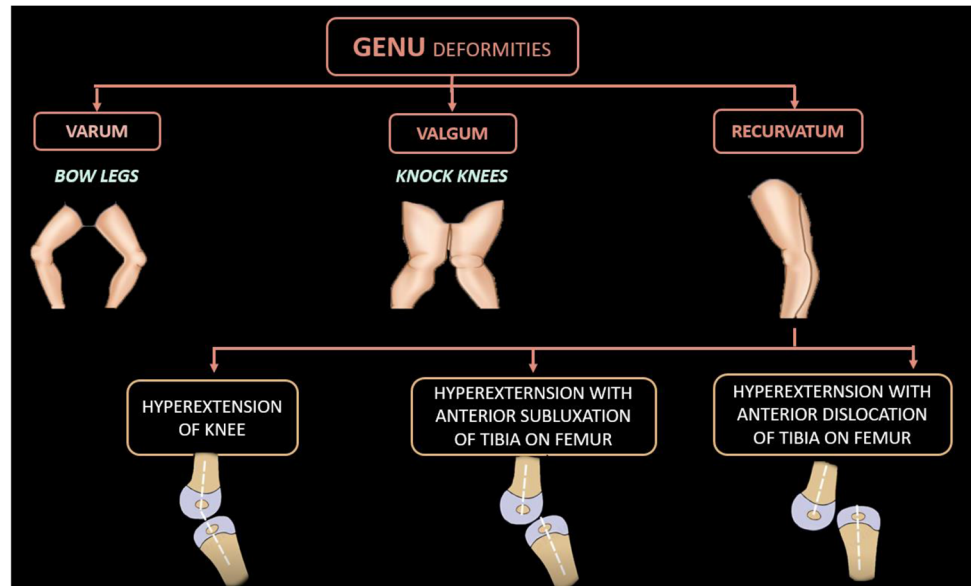


Fig. 2 This image is of Ella Harper. She was widely known as “camel girl”. She became famous as one of the few people in the world having Genu Recurvatum

All the patients opted for termination of pregnancy. The first three cases had associated findings and the couples did not want any invasive procedures and opted directly for termination. The last case also, however, wasn’t willing to continue the pregnancy. There have been cases reported in the past stating corrective surgeries for genu recurvatum, but this couple wanted to terminate (Fig. 3).

Discussion

Genu recurvatum is a rare congenital anomaly also known as congenital dislocation of the knee (CDK) or congenital dislocation of the patella (CDP). It is a rare condition occurring in 1 in every 100,000 newborns [1]. It is defined as a pathological hyperextension of the knee greater than 30°, associated with limited flexion. The degree of

recurvatum is not in itself correlated with prognosis [2]. This depends on the presence of a dislocation of the knee [3] (Fig. 4).

Figure 1 describes the three pathological variants of congenital genu recurvatum [4, 5].

CDK has an unknown physiopathology. Congenital Genu Recurvatum can be divided into two groups [6]:

- Malformative Congenital Genu Recurvatum, involving anomalies of the elastic tissues and
- Postural Congenital Genu Recurvatum, as a consequence of abnormal presentation or oligohydramnios.

The most common associated anomalies are congenital dislocation of the hips and foot deformities. [7, 8]

CDK can occur in isolation or as part of different conditions like arthrogryposis multiplex congenita, myelomeningocele, Marfan, Ehlers-Danlos, and Larsen syndromes [9]. Genetic syndromes characterized by joint hypermobility include Ehlers-Danlos syndrome, Marfan syndrome, Larsen syndrome, and osteogenesis imperfecta [10]. Congenital tibiofemoral subluxation, arthrogryposis multiplex, spastic cerebral palsy, and cervical myopathy are among other conditions to be considered in the differential diagnosis. Table 1 describes the various common syndromic associations with genu recurvatum.

Oligohydramnios and footling presentation may be associated with postural genu recurvatum. Ischemia of quadriceps leading to fibrosis can also lead to Congenital Genu Recurvatum. [16] Contracture of quadriceps due to any reason may also lead to Congenital Genu Recurvatum.

To the best of our knowledge and extensive literature search, there are no defined criteria for diagnosis of genu recurvatum on ultrasound. A routine screening suggestive

Fig. 3 An Image like ‘A’ should increase the suspicion for Congenital Genu Recurvatum (yellow arrow demonstrating extended knee). A more thorough image of the same patient showed hyperextension at the knee joint (image ‘B’). Volume Analysis can aid in diagnosis. A 3D/4D reconstruction can be diagnostic in cases of doubt. Features like 3D magicut can enhance the perception by clearing the point of focus

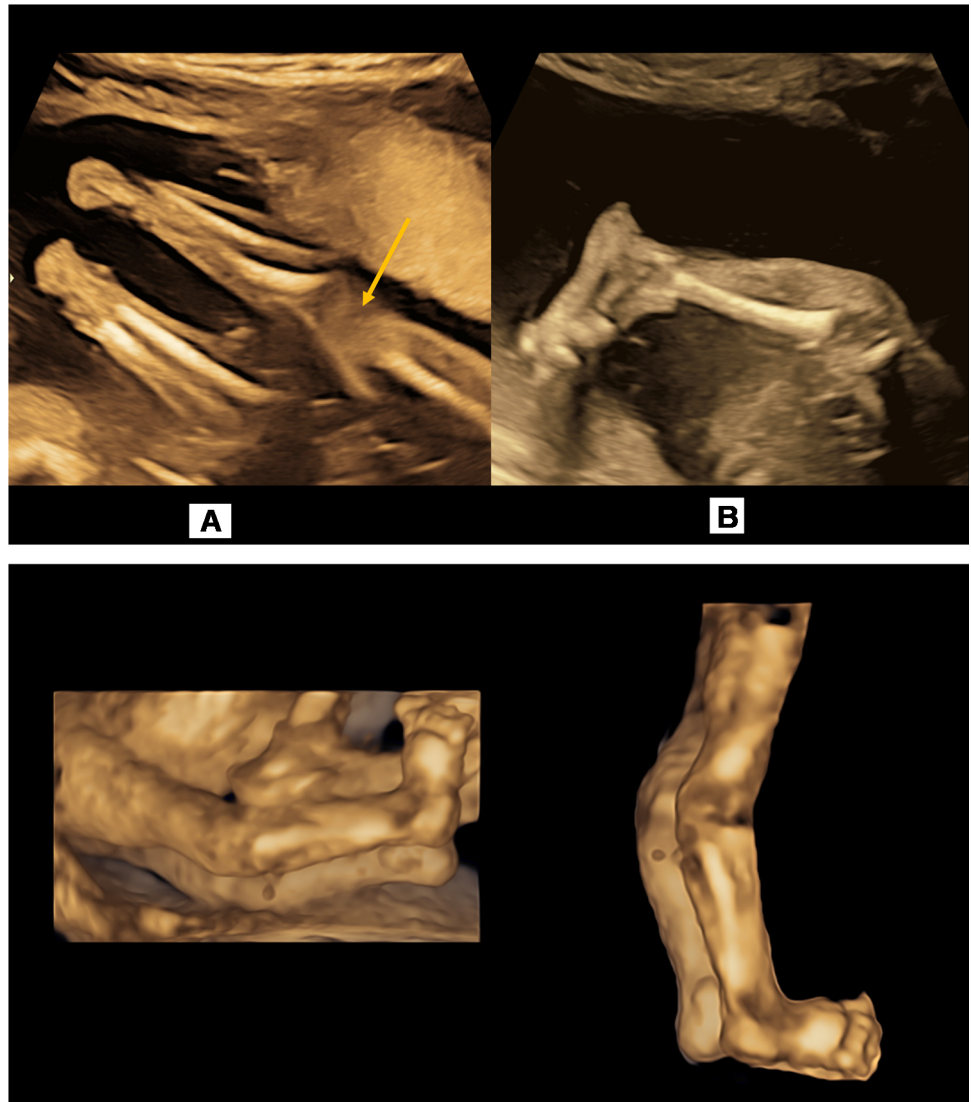


Fig. 4 G2P0A1 attended the center for her anomaly scan at 21 weeks and 3 days. The fetus showed bilateral Genu Recurvatum and rocker bottom foot. (patient number 3 in the text.)

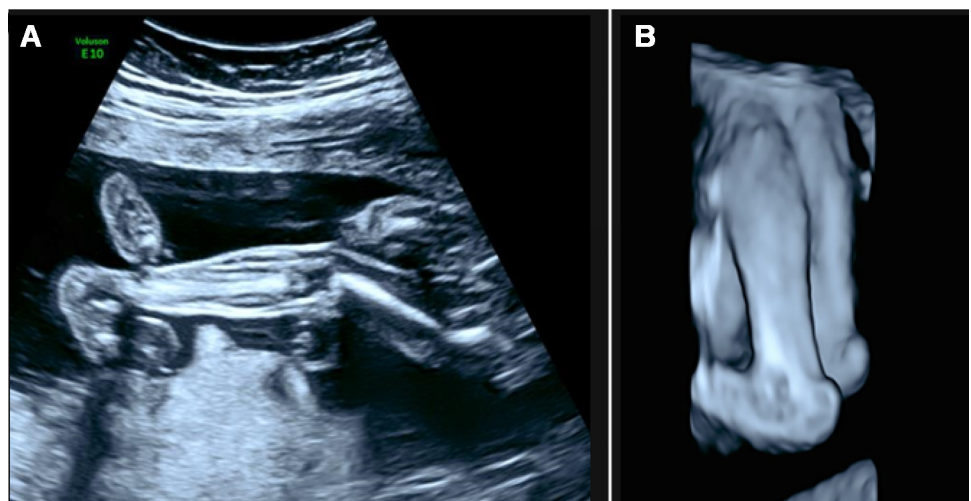


Table 1 Common syndrome associated with genu Recurvatum

Elher Danlos syndrome [11]	Autosomal dominant Defect in collagen production Skin extensibility, Hypermobility of the joints, and Tissue fragility	Arachnodactyly, Clubfoot, Micrognathia, Radial dislocation, kyphoscoliosis, Spondylolysis and spondylolisthesis
Marfan syndrome [12, 13]	Autosomal dominant, 25% sporadic Connective tissue disorder	Cardiovascular, ocular and skeletal systems (wide variety)
Larsen syndrome [14, 15]	Autosomal dominant or recessive Generalized mesenchymal connective tissue disorder	Bilateral hip and elbow dislocations, Genu recurvatum, Club foot, Short metacarpal bone (cylindrical finger) Prominent forehead, Depressed nasal bridge, Round faces, Micrognathia, Hypertelorism Cleft palate, Hydrocephalus, Cervical kyphosis, etc

of a hyperextended knee should prompt further inspection. Imaging advances have led to an improvement in diagnostic abilities. Bone imaging has also peaked with the addition of VCI and 3D/4D. A general routine anomaly scan will be able to diagnose severe Congenital Genu Recurvatum and also associated anomalies. So when - should we look for Congenital Genu Recurvatum? In cases where the associations are present, the knee should also be taken a view of. General cases with mild Congenital Genu Recurvatum, even if missed, carry a good prognosis. If found, the couple wish to continue the pregnancy, it is better to offer invasive investigation to rule out associated anomalies. Early diagnosis with proper counseling should be done to ensure that the couple makes an informed decision.

There is no consensus on the mode of delivery of the fetus with Congenital Genu Recurvatum. As the association with footling breech is more, these cases might as well have a C section for obstetric indications.

Babies born with Congenital Genu Recurvatum should be immediately identified and referred to a multidisciplinary approach of orthopedic surgeries and pediatricians. For isolated congenital genu recurvatum, conservative orthopedic treatment using casts, corrective splints, physical therapy, braces, and gait training is preferred. [14, 17]. Several classifications and new grading systems [18] are being studied for the treatment in postnatal life. Severe

cases are surgically corrected. According to some authors [19, 20] surgical treatment must be performed before the infant begins to stand or put weight on the affected knee, preferably around 6 months. Long term follow up is needed to evaluate treatment and detect non-identifiable syndromes that might be more evident later in life [19].

Conclusion

Given the imaging technology currently available, direct visualization of the fetal skeletal system is now possible with techniques like VCI and 3D. Prenatal recognition of Congenital Genu Recurvatum is significant not only because of the important distinction from other skeletal dysplasia abnormalities but also because identification and characterization of the disease may have implications for the optimal mode of delivery. Prenatal diagnosis may also help in providing parents with the option of pregnancy termination if the parents desire appropriate obstetrical management and planning; surgical and medical treatment; or adequate counseling about the long-term prognosis of the disease.

Declarations

Conflict of interest The authors declare that they have no conflicts of interest.

Informed Consent Consent from the patients in the series have been taken to publish the respective case details and USG. (The couples refused to share the photographs of the abortus and their wish was duly respected).

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