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Case Series

Significance of Short Femur Length in Pregnancy Case Series

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Received: October 12, 2022; **Published:** October 24, 2022

Abstract

Objectives: To compare the etiologies and adverse outcomes of pregnancies with short fetal femur length (FL) categorized based on relative proportion and percentile placement.

Methods: We present 5 examples of prenatally diagnosed and briefly reviewed the differential diagnosis of short femur(s) in utero and associations such as trisomy 21, diabetic embryopathy, IUGR and skeletal dysplasia.

Results: Multiple cases are presented in which a short femur (unilateral or bilateral) is the dominant finding on prenatal ultrasound. A short FL is generally defined as a value that is two standard deviations below the mean FL for a particular gestational age. The etiology of short FL includes normal variant skeletal dysplasias, aneuploidies, and intrauterine growth restriction. Prenatal imaging findings are correlated with postnatal history and imaging.

Conclusion: Prenatal sonography is now widely used as a screening tool, and at times, subtle findings such as an isolated short femur can be seen without other significant anatomic abnormalities. Counseling for the parents can be difficult without some knowledge of the range of associations seen with short femur(s).

Keywords: *Prenatal Ultrasound; Short Femur; IUGR; Skeletal Dysplasia; Diabetic Embryopathy*

Introduction

Prenatal growth estimation and weight assessment are done during the second and third trimesters using a combination of the fetal biparietal diameter, the head circumference, the abdomen diameter and its circumference, as well as the baby›s femur length. During the first trimester, the embryo›s crown-rump length and biparietal diameter are used to gauge and confirm gestational age. This makes measuring the femur's length straightforward [1].

Percentiles were originally used in 1994 by Snijders and Nicolaides to define the normal distributions for biometric parameters. A short fetal femur was defined as falling within the fifth percentile of femur length [2].

The discriminatory power of a prenatally diagnosed "short femur" has been discussed in a variety of ways up to this point because it can be a soft marker for trisomy [6-11], suggest skeletal dysplasia [4,5,12], suggest early growth restriction [3-5], suggest skeletal dysplasia, and also show as a normal variant, especially in the context of different ethnicity [13,14].

This can result from a femur-specific genetic abnormality, a trisomy condition, fetal damage during development, fetal infection during development, or other factors. When a trisomy disorder is mentioned, trisomy-21 (Down syndrome), trisomy-18 (Edward's syndrome), or trisome-13 are usually included (Patau syndrome).

Femur length is one of the basic biometry metrics used in prenatal sonography to gauge gestational age and fetal growth (FL). A short FL is a fetal FL that deviates by no more than two standard deviations from the mean FL for a certain gestational age [4].

Objective of the Study

In this study, we investigated the etiology and outcome of fetuses from various categories case series and controlled the significance of short femur length in pregnancy using percentile and proportion as classifying criteria.

Methods

In a private clinic, ultrasound imaging for cases 1, 3, 4 and 5 was carried out utilizing a Medicon R7 and a 6-MHz transducer. Using a 6-MHz transducer and a Fukuda machine, case 2 was photographed.

Case 1

A 24-year-old pregnant lady got standard care at our outpatient prenatal clinic. She and her husband both had ordinary and diabetes-filled family histories. The discovery of serious malformations occurred in the 13th week of pregnancy. The nasal bone was absent, the NT was greater than 1 cm, the FL was short, and the biparietal diameter and the abdominal circumference were all within the normal range for gestational age at that time (Figure 1-3). Trisomy 21 was discovered during amniocentesis at 16 weeks and the patient chose to bring the pregnancy to term. But at 30 weeks (Figure 4-6), she stopped feeling the baby move and delivered a dead fetus. Down syndrome has been confirmed by the fetus' Karyotype.

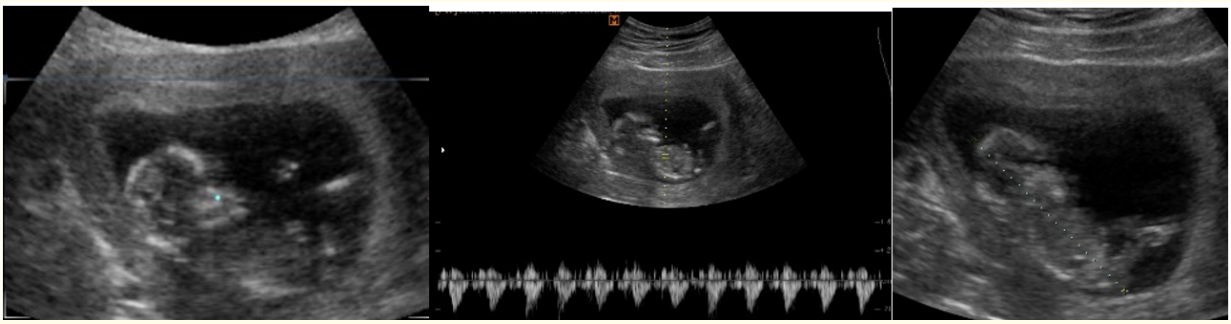

Figure 1-3: *At the 13th week of pregnancy, we seen the atrioventricular communication on the heart, the nasal bone absent, the NT > 1 cm, the FL short.*

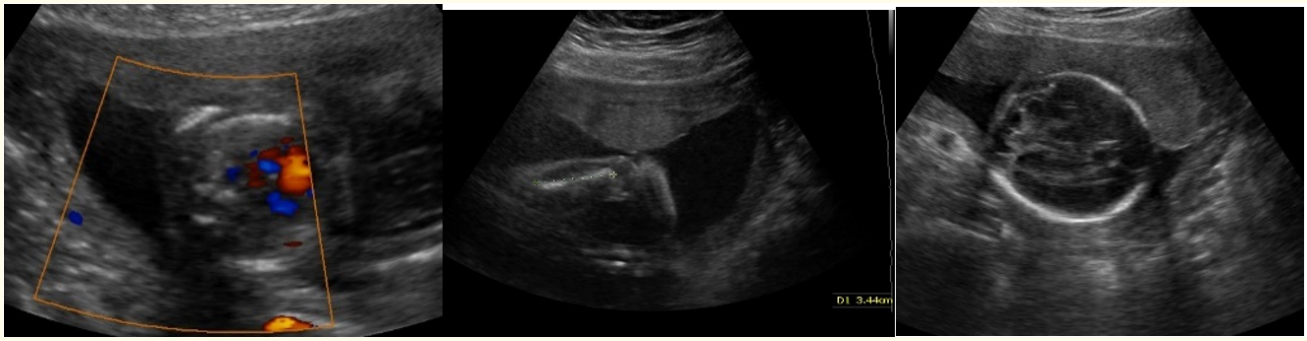

Figure 4-6: *The atrioventricular communication and FL short on the heart at 30 weeks. The abdominal size and biparietal diameter were both within the usual range for gestational age.*

Case 2

A 23-year-old lady who was gravida 1, parity 0, and outpatient pregnant received routine treatment. Her personality was typical. The discovery of nanism skeletal dysplasia family histories between the two people suggests first-degree consanguinity. The discovery of serious malformations occurred in the 13th week of pregnancy. NT superior at 1 cm, FL bilateral was short at 13 weeks.

While all measurements were in line with gestational age, the results of ultrasound at 23 weeks showed that the fetal femur was 5 weeks shorter than expected (Figure 7). The extremities' bones were all short and symmetrically angular in shape, with bilateral talipes equinovarus, a small scapula, and a thin thorax, according to a thorough ultrasound.

Figure 7: *An ultrasound at 23 weeks revealed that the fetal femur was 5 weeks shorter than anticipated.*

The patient elected to carry the pregnancy to term despite these problems, however at 40 weeks the patient complained of an unusually enormous tummy. He was born, but due to severe hydrocephalus, he went away very quickly (Figure 8 and 9).

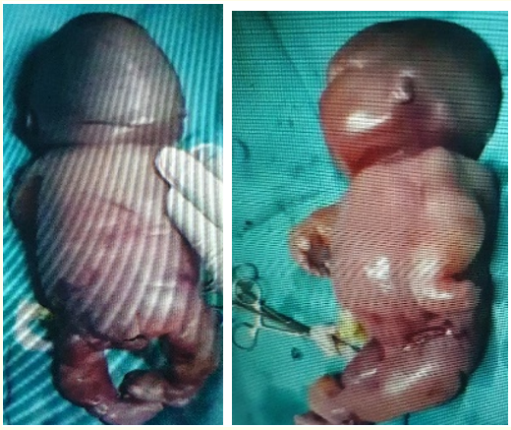

Figure 8 and 9: *The bones in the extremities were all short, and the belly was large.*

Case 3

25 years old a G2 P1, diabetic, insulin-dependent, 1cesarean section.

The patient's initial ultrasound at 14 weeks revealed severe oligohydramnios alteration, and short femurs with leg fusions (Figure 10 and 11). The patient had spontaneous abortion a16 weeks (Figure 12-15).

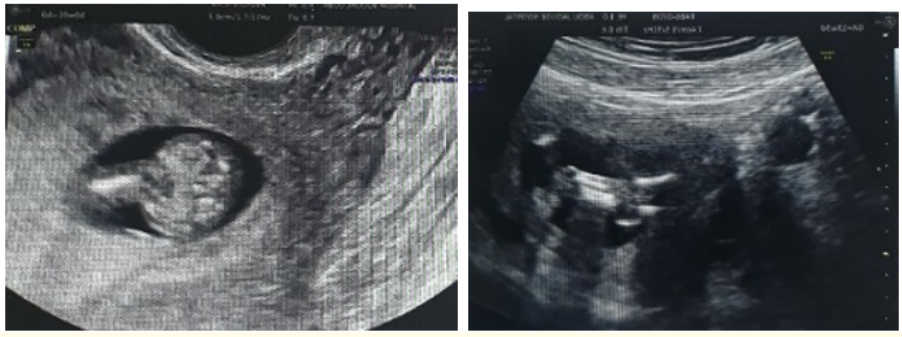

Figure 10 and 11: *Pelvic ultrasonography showed significant oligohydramnios, a shortened femur and leg fusion.*

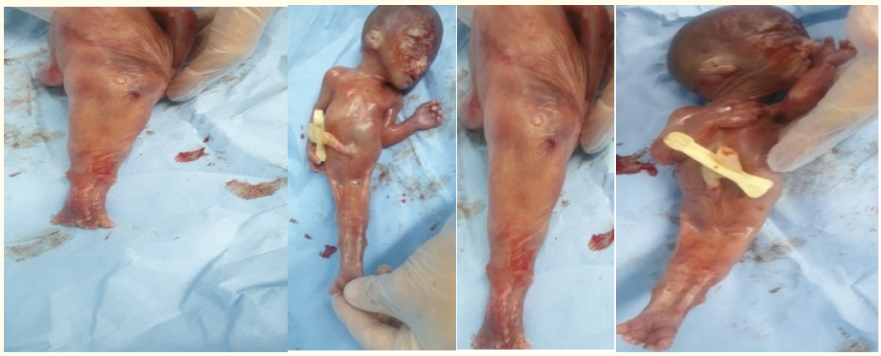

Figure 12-15: *Sirene fetus.*

Case 4

A 23-year-old G1P0 healthy, overweight, non-smoker who had a first-degree relative in marriage came up at the clinic as well for her first prenatal check-up. The first-trimester feto-maternal ultrasound results showed NT = 2.2 CM with a positive nasal bone. Iron, multivitamins, and progesterone were among the first-trimester medications examined. The results of the scan at 24 weeks of gestation showed a short, seemingly normal-appearing femur measuring 3.77 cm in length, which correlates with 22 weeks of gestation with 1.42%, bilateral normal tibia and fibula, no other indications of skeletal dysplasia, an increase in the amount of amniotic fluid, and no other abnormalities (Figure 16-18). Cost factors were taken into account even though the patient desired an amniocentesis. A follow-up ultrasound showed that the EFW at 28+6 weeks of gestation was 1.6 kg and that the femur was less than 1% short. Ultrasound at 36 weeks of pregnancy showed a short femur bone of less than 1% and a humerus bone of 10%. Due to the imbalance between the fetus and pelvis, the patient underwent a c-section and delivered a boy weighing 3,6 kg and measuring 52 cm in length. The newborn's characteristics were entirely normal and showed no indications of aneuploidy. The infant's growth chart over a year was typical, measuring 74 cm in length.

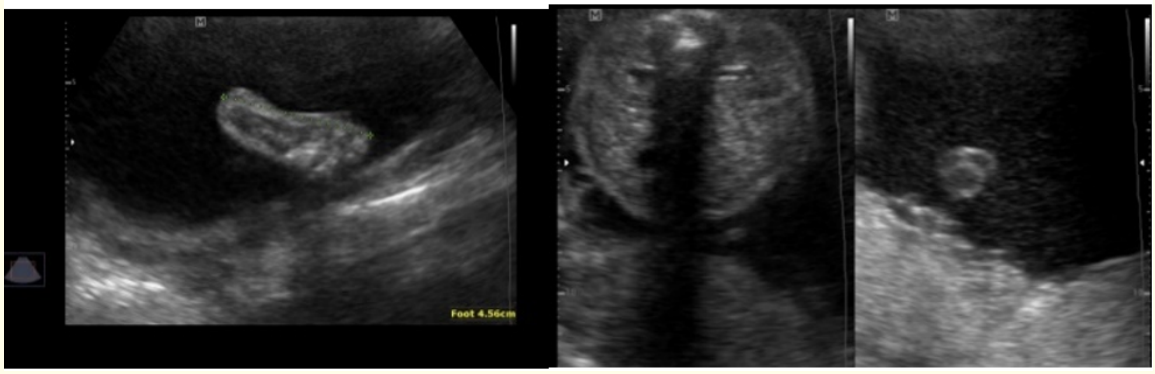

Figure 16 and 17: *The abdominal circumference was all within the normal range for gestational age other structure normal.*

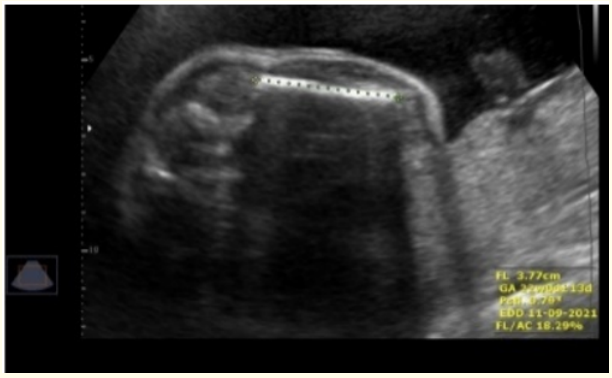

Figure 18: *A short, visible femur measuring 3.77 cm in length, which, with 1.42% accuracy, corresponds to a gestational age of 22 weeks rather than 24 weeks.*

Case 5

24 years, G3P2, no pertinent medical background. She showed up for a screening ultrasound and dated 20.6 weeks pregnant. The left femur was measured at that time to be 23 mm, 3 SDs below the mean gestational age (Figure 19). The anatomy of the left femur was otherwise usual. The size of the right femur was 35 mm. The remaining long bones were measured and confirmed to be healthy. The pelvis appeared typical. At the 34th week of pregnancy, FL measured 5.4 cm, which is equivalent to a gestational age of 29 weeks. Measurements of the biparietal diameter (8.2 cm) and belly circumference were used to determine the typical range for gestational age, which was associated with a gestational age of 29 weeks (26.4 cm) with abnormal Doppler results (Figure 20-22).

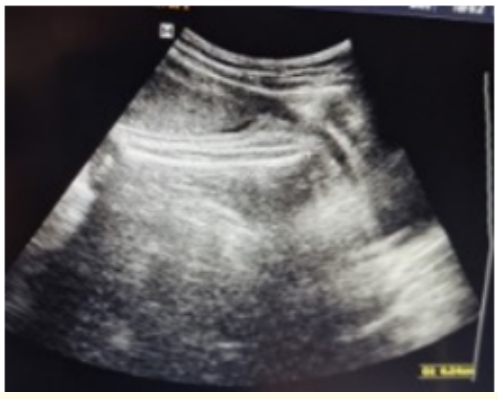

Figure 19: *FL measured 5.4 cm at the 34th week of pregnancy, which corresponded to a gestational age of 29 weeks.*

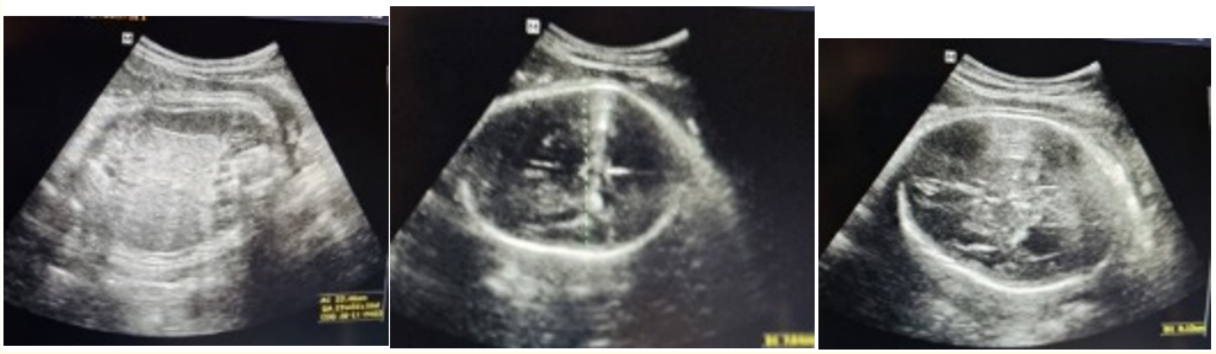

Figure 20-22: *Biparietal diameter (8.2 cm) and abdominal circumference (26.4 cm).*

A baby was delivered at term with fetal cardiac hypertrophy (Figure 23) and the left femur was identified as being linked to IUGR while the infant was receiving evaluation and therapy in the NICU. identified as having a connection to IUGR.

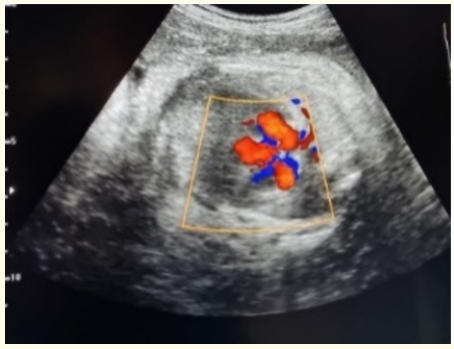

Figure 23: *Fetal heart hypertrophy.*

Discussion

Prenatal medication is being developed at a breakneck pace. Possibilities have greatly expanded in recent years, particularly in the field of genetic testing. Fetal ultrasound is the cornerstone of every prenatal diagnostic. The more thorough the examination, the clearer the diagnosis, which serves as the basis for the best possible advice for expectant parents. One of the standard procedures for biometric measurement is the sonographic assessment of femur length, which is simple to carry out in the era of high-resolution ultrasound technology. A short femur is defined as having a femoral length below the fifth percentile of defined biometric measurement criteria, with normal, healthy children of short stature as a standard variant.

Additionally, it is thought that a short femur is a "soft marker" that, when combined with other soft markers, raises the chance of trisomy, particularly trisomy 21. It has been hypothesized that there is a connection between these conditions and other fetal abnormalities, chromosomal problems, placental insufficiency, or other birth-related hazards such as preterm birth and low birth weight infants [16].

When a short FL is discovered, a variety of factors must be investigated in order to create an efficient management strategy. Compared to high-level markers like the nuchal skin fold, short FL is a low-level marker for aneuploidies [7]. People with Down syndrome commonly exhibit several aneuploidy indicators, such as choroid plexus cysts, duodenal atresia, nuchal translucency or pyelectasis.

According to earlier research like our first case, early pregnancy with Down syndrome is characterized by a short femur along with other soft markers including NT and an atrioventricular septal defect, which was confirmed by an amniocentesis at 16 weeks. Typically, the diagnosis is made via the Down syndrome screening tests and the identification of many soft markers while receiving prenatal care.

Mathiesen., *et al*. presented the largest data set in 2014 [17]. The definition of a short femur varies in current literature, and only a small number of studies make a distinction between a biometric short femur alone and one combined with other deformities or within the context of a syndrome, making it difficult to compare these studies with one another. The first ten years of data collecting did not include any documentation of ethnicity in clinical practice.

Beginning in the early 2000s, it became clear that numerous characteristics, including mother and paternal height, BMI or birth weight, and ethnicity, had an impact on certain medical issues.

At 7 to 10 weeks of gestation, the limb skeleton organogenesis begins. However, between 12 and 26 weeks of gestation, it is possible to test for severe limb deformities. The morphological evaluation of the limbs is still a challenging procedure that must take fetal motion into consideration. A spondylo-epi-metaphyseal dysplasia known as metatropic dwarfism, or dysplasia, is characterized by kyphoscoliosis, enlargement of the joints, and short limbs with a normal-height trunk. It is a highly uncommon type of dwarfism, with an estimated prevalence of fewer than 1/,000,000 [9]. TRPV4 gene mutations are to blame for the pathogenesis. Transmission can be dominant or recessive [2]. These mutations cause the chondrocytes' calcium levels to rise, which prevents endochondral ossification from occurring. Skeletal dysplasia must be considered in cases of shortening of the long bones. There are more than 450 different kinds of skeletal dysplasia, including achondroplasia and hypochondroplasia [8].

If a short FL is found on sonography, it is generally advisable to compare all other long bones to the expected usual values. Skeletal dysplasia is associated with short FL before 24 weeks of gestation and is frequently accompanied by anomalies of other tubular bones, metaphyseal changes, or bowing. The diagnosis is made using the results of DNA testing and family history. In our situation, the patient completed her pregnancy despite declining the test due to a known history of dwarfism.

Sirenomelia, also known as Mermaid Syndrome, is an extremely unusual congenital deformity that is characterized by genitourinary and gastrointestinal system malformations, thoracolumbar spinal anomalies, sacrococcygeal agenesis, and varying fusion of lower limbs. It was given the common term Mermaid Syndrome because these misshapen fetuses resemble the mermaid (Siren) from Greek mythology [30-33]. The male-to-female to female ratio for sirenomelia is 3:1, with a frequency of 0.8 - 1 case per 100,000 births. Although several theories have been put up regarding the origin of sirenomelia, none are thought to be definitive. Given that 22% of cases have a diabetic mother, there is a clear link between maternal diabetes and the condition [34-36]. Fusion of the lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia [30-41]. Various anomalies present in sirenomelia include varying degree of fusion (symelia) and/or hypoplasia of lower limbs, vertebral anomalies (sacral agenesis), anorectal agenesis, urinary tract anomalies and genital anomalies [34-37]. Sirenomelia is fatal and about 50% of cases are stillborn because of bilateral renal agenesis and

associated visceral anomalies [38-40]. Many researchers have considered sirenomelia to be a severe form of caudal regression syndrome because of the observed abnormal development of fetal caudal mesodermal structures before the 4th week of gestation, that later extends to different craniocaudal levels. It may also lead to the absence of genitalia and renal agenesis if paramesonephric and mesonephric ducts are involved. Survival depends upon visceral anomalies instead of sirenomelia. The syndrome is classified into three types according to the number of feet: simpus apus, no feet present; simpus unipus, one foot; simpus dipus, two feet [39-41]. Our case falls under this group.

Numerous studies have shown a link between isolated short FL and IUGR. However, IUGR frequently displays other abnormal biometric measurements including Doppler results. The abnormal femur growth pattern in IUGR can start at any time, however, this usually manifests on sonography as a small belly circumference. If more than one soft indication is seen during prenatal care, serial ultrasonography to monitor growth restriction and karyotype analysis can be performed to confirm the diagnosis [19-23].

Children with short femurs have LBW fetuses, which are 7.1%–8.6% higher than LBW fetuses with normal femur biometry [24,26-28].

Our case the short femur associated with IUGR start at 6 months.

Summary

In the event that an isolated short FL is detected on ultrasonography, a conservative management approach is suggested [15,18-20]. Typically, isolated short FL is discovered in the second trimester. In this case, short FL was not identified until the third trimester. Serial ultrasonography is required to rule out skeletal dysplasia, IUGR, and aneuploidies [21-25]. If there are sonographic signs of aneuploidies, such as the nuchal skin fold, karyotype investigation is indicated. Short FL due to IUGR should be looked into if sonography results reveal a tiny belly circumference or abnormal Doppler parameters, particularly of the uterine and umbilical arteries [26-28]. It is vital to discuss potential diagnoses, plans for the newborn and red flags for future pregnancies with the parents.

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Volume 11 Issue 11 November 2022