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ULTRASOUND FINDING OF THE 'TULIP SIGN' IN A FETUS WITH CHIARI II MALFORMATION AND ASSOCIATED NEUROGENIC INCONTINENCE

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ABSTRACT

RATIONALE FOR THIS CASE REPORT. This chapter reports on the case of a fetus with Chiari II malformation in breech presentation, which was diagnosed prenatally with 33 weeks gestation. Additionally, the 'tulip sign' was diagnosed ultrasonographically, which had previously been described as a "unique sonographic picture of hypospadias".
But in this case, prenatal ultrasound suspicion of hypospadias was not confirmed and, instead of that, the newborn was diagnosed with neurogenic bladder– and anal incontinence.
The author assumed that, in this case, the ventral flexion of the penis was the result of neurologically induced atony of the penis in the fetus with spina bifida in breech presentation, with the help of the gravitational force.
PRESENTING CONCERNS OF THE PATIENT. A 22–year old woman, gravida 1, with no significant medical history, scheduled an ultrasound exam at our clinic at 33 weeks gestation because of previously diagnosed fetal hydrocephalus, in order to get a second opinion.
DIAGNOSIS. Chiari II malformation with ventriculomegaly, corpus callosum agenesis, and suspected hypospadias based on the ultrasound finding of the tulip sign.
OUTCOMES. A term male newborn was delivered by cesarean section at 38 weeks gestation. The newborn was confirmed to have Chiari II malformation with hydrocephalus and agenesis of corpus callosum, but prenatal ultrasound suspicion of hypospadias was not confirmed. The myelomeningocele was surgically repaired during the first postpartum week. After that, the cerebellum spontaneously returned to its normal position and then a shunt was surgically inserted into the lateral cerebral ventricle.
During the postnatal period, the newborn was diagnosed with neurogenic bladder– and anal incontinence.

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MAIN LESSONS LEARNED FROM THIS CASE. The ultrasound finding of the tulip sign is not only specific for hypospadias, and in some special conditions, if a fetus with spina bifida or a complex malformation which includes spina bifida sits in breech presentation, an ultrasound finding of the tulip sign could indicate atony of the penis and neurogenic incontinence in the differential diagnosis.

KEYWORDS: Chiari II, Lemon sign, Beaked tectum, Interhemispheric cyst, Myelomeningocele, Callosal agenesis, Colpocephaly, Tulip sign, Hypospadias, Neurogenic incontinence.

INTRODUCTION

Fetal ultrasound enables the visualization of different anatomical structures, including fetal genitalia. These structures can be seen from the early second trimester of pregnancy, a point in fetal development when the phallus can be identified by its typical sagittal inclination, as opposed to the clitoris, which is situated longitudinally in alignment with the thigh [1–2].The scrotum is also visible from the early second trimester, and as pregnancy progresses, the presence of testicles in the scrotum becomes visible [3].

Hypospadias is a birth defect found in boys in whom the opening of the urethra is not at the tip, but on the ventral side of the penis. Hypospadias is a common developmental disorder of the urogenital tract, with an incidence of between 0.2 – 4.1 per 1,000 live births or occurring in approximately 1 in 125 live male births [4–6].

Urologic classification of hypospadias is based on meatal position: anterior (glandular or coronal), occurring in approximately 50%; middle (penile shaft), constituting 30%; and posterior, accounting for the remaining 20%. A posterior location may occur at the penoscrotal junction, within the scrotum, or along the perineum [7]. In the case of hypospadias, ventral curvature of the penis may be observed and is known as the chordee. The chordee is caused by atresia of the corpus spongiosum distal to the hypospadiac urethral meatus [8].

Embryologically, hypospadias occurs when the fusion of the urethral folds stops proximal to the tip of the glans penis. The recurrence risk of having a second male child with hypospadias is between 4% and 12% [5].

The etiology of nonsyndromic hypospadias is unknown, and is believed to be multifactorial. Recent studies have implicated factors such as familial inheritance, low birth weight, assisted reproductive technology, advanced maternal age, paternal subfertility, and endocrine–disrupting chemicals in the pathogenesis of hypospadias [9].

Hypospadias usually occurs as an isolated defect, but can be part of a recognized syndrome or associated with other genital or extragenital anomalies. The prevalence of associated urogenital abnormalities may be as high as 40%, and extraurogenital anomalies may be present in 7% to 9% [10–11]. The most common extraurogenitally associated anomalies include congenital heart defects, cleft lip and palate, myelomeningocele, anorectal malformations, tethered cord, sacral agenesis, and frontal bossing [12].

Hypospadias may also occur as a part of a number of syndromes, including Opitz, Opitz–Frias and Smith–Lemli–Opitz syndrome, Nager syndrome and acrofacial dysostosis. Hypospadias is one of the most prominent and characteristic midline defects in male infants with Wolf–Hirschhorn (4p–) syndrome [13].

The sonographic findings for the suspected diagnosis of hypospadias are

- anomalousdistal morphologic characteristics of the penis,
- small lateral folds (dermal remains of the prepuce),
- a small penis,
- ventral incurving and
- an anomalous urinary stream [14,9].

Lin et al. reported a rare variant of hypospadias (penoscrotal type) in which the penis and scrotum had a normal appearance but a urethrocutaneous fistula and a cystlike protruding lesion on the ventral side of the penis, evident during urination, were observed [15].

THE TULIP SIGN, a sonographic marker for severe hypospadias, was firstly reported by Meizner in 6 fetuses with single umbilical artery and absent kidney, as "helpful in diagnosing severe hypospadias"[9]. This sign, described as "a unique and specific sonographic sign for prenatal detection of severe penoscrotal hypospadias", represents the severe curvature of the penis in association with penoscrotal transposition. The tulip flower is formed by the ventrally bent penis, located between the two scrotal folds [15–16].

This chapter reports on the case of a fetus with Chiari II malformation in breech presentation, with the ultrasound finding of the tulip sign. But in this case, prenatal ultrasound suspicion of hypospadias was not confirmed and, instead of that, the newborn was diagnosed with neurogenic bladder– and anal incontinence. The author assumed that, in this case, the ventral flexion of the penis was the result of neurologically induced atony of the penis with the co–work of the gravitational force in the fetus with spina bifida in breech presentation.

PRESENTING CONCERNS

A 22–year old woman, gravida 1, with no significant medical history, scheduled an ultrasound exam at our clinic at 33 weeks gestation because of previously diagnosed fetal hydrocephalus, in order to get a second opinion.

PATIENT INFORMATION

The patient was born on April 23, 1986. As a child she had mumps and chickenpox.

Menarche occurred at the age of 12. Menstrual cycles were regular, from 27 to 30 days, with periods lasting 4 days.

CLINICAL FINDING

A detailed fetal transabdominal sonography with high–frequency ultrasound transducer and three–dimensional (3D) technology was performed and male fetus in breech presentation with adequate amniotic fluid and posterior wall placenta was diagnosed. Fetal biometric measurements correlated well with the expected gestational age according to the dates of the patient's last menstrual period.

Fetal Head and Brain

Head shape, brain texture and anatomical integrity were examined using the three standard views – the transventricular plane, transcerebellar plane and transthalamic plane [17].

The transventricular plane, which usually demonstrates the anterior and posterior portion of the lateral ventricles, revealed asymmetrically extended lateral ventricles at the level of the atrium, wherein the width of the left ventricle was 14.9 mm – "moderate ventriculomegaly", and the width of the right ventricle was 17.5 mm –"ventriculomegaly of severe degree" or hydrocephalus (Figure 1/A). Between the ventricles there was an 8.4 mm interhemispheric cyst, which is usually due to expansion of the posterior recesses (pineal and suprapineal recesses) of the third ventricle. Third ventricle enlarges often and early in aqueductal stenosis, even if ventriculomegaly is minor. Interhemispheric cysts can be seen in a variety of both normal and abnormal circumstances in fetuses, which include interhemispheric arachnoid cysts associated with callosal dysgenesis, pineal cyst and cava vela interpositi [18].

Occipital horns of both ventricles showed an angular deformity or a "pointed" appearance, known as ventricular or occipital 'point' [19], which is a common supratentorial feature of the Chiari II malformation (Figure 1/B).

Three–dimensional reconstruction of the fetal head demonstrated the 'lemon sign', medial inward displacement and scalloping of the frontal bones, that gives the fetal calvarium the configuration of a lemon [20]. It is commonly seen as a sign of a Chiari II malformation and also seen in the majority of fetuses with spina bifida (Figure 2/A).

The transcerebellar plane, that includes visualization of the frontal horns of lateral ventricles, cavum septi pellucidi (CSP), thalami, cerebellum and cisterna magna demonstrated an empty posterior fossa. The cerebellum could not be displayed, and the author suspected it was the downward displacement of the cerebellar tonsils through the foramen magnum (Figure 2/B). Instead of normal, rounded appearance of the tectum, fusion of the colliculi and upward deflection of the tectum resulted in prominent beaking and elongation of the tectum, known as a 'beaked tectum' (Figure 2/B). The severity of tectal beaking correlates with the size and extent of the myelomeningocele and a severe tectal deformity is the likely cause of upward gaze paresis and other oculomotor problems in children with the Chiari II malformation [21].

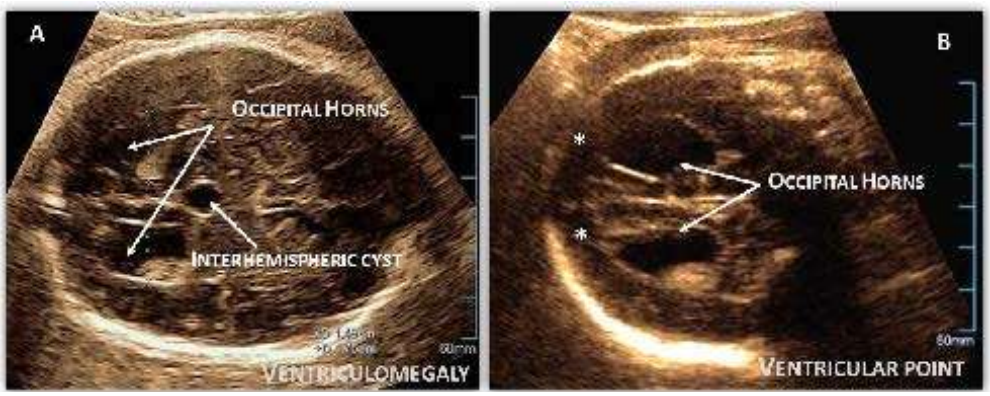

Figure 1. Supratentorial abnormalities in the Chiari II malformation.
A – The transventricular view: Ventriculomegaly with enlarged occipital horns and interhemispheric cyst. B – Ventricular 'point', angular deformity of the occipital horns of the lateral ventricles (*).

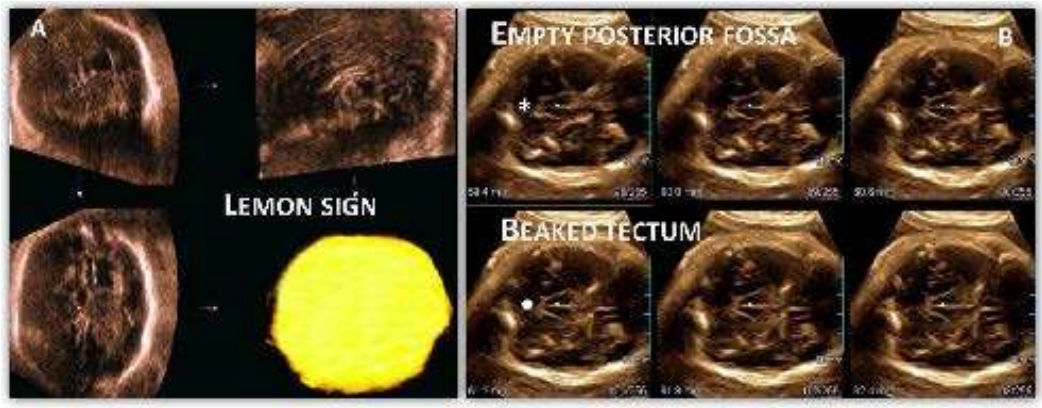

Figure 2. Cranial signs of myelomeningocele.
A – 'Lemon sign': 3D multiplanar view of the medial inward displacement and scalloping of the frontal bones, which gives the fetal calvarium the configuration of a lemon.B – The transcerebellar view: Instead of a 'banana sign' and normal, rounded appearance of the tectum, it demonstrated an empty posterior fossa (*) due to complete herniation of the cerebellar tonsills and 'beaked' tectum (•), prominent beaking and elongation of the tectal plate.

The basis of the neurosonographic examination of the fetal brain is the multiplanar approach, which is obtained by aligning the transducer with the sutures and fontanelles of the fetal head [17]. The coronal view at the level of the frontal horns of lateral ventricles (the transcaudate plane) depicts the midline interhemispheric fissure the continuity of which is interrupted by the genu or anterior portion of the corpus callosum. The cavum septi pellucidi is depicted as an anechogenic triangular structure under the corpus callosum, between the lateral ventricles.

The transcaudate plane depictured uninterrupted continuity of the midline interhemispheric fissure with absent cavum septi pellucidi (Figure 3/A), and inferiorly beaked frontal horns.

Additionally, the sagittal view showed the disproportionate enlargement of the occipital horns of the lateral ventricles. The normal reduction in diameter of the occipital horns of the lateral ventricles is critically dependent upon correct morphogenesis of the corpus callosum. Therefore, the finding of enlargement of occipital horns is commonly associated with partial or complete agenesis of the corpus callosum and it is called 'colpocephaly' (Figure 3/B).

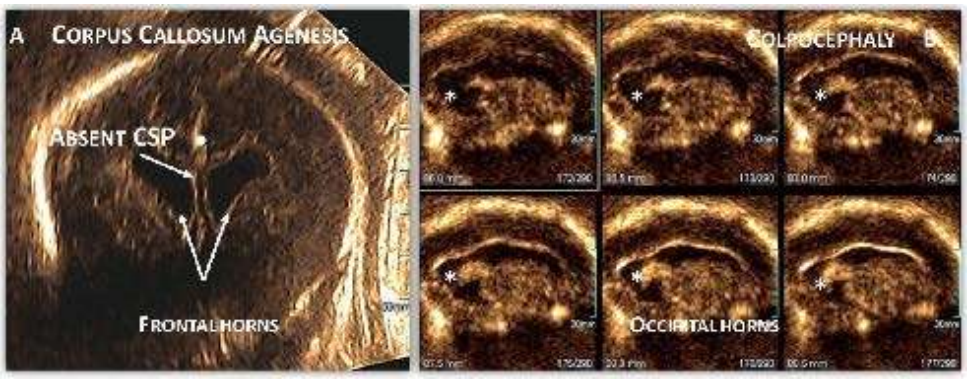

Figure 3. The agenesis of the corpus callosum.
A – The coronal view: Absent cavum septi pellucidi (CSP); angular deformity of the frontal horns, uninterrupted interhemispheric fissure (•). B – The saggittal section: Colpocephaly(*), enlarged occipital horns of the lateral ventricles.

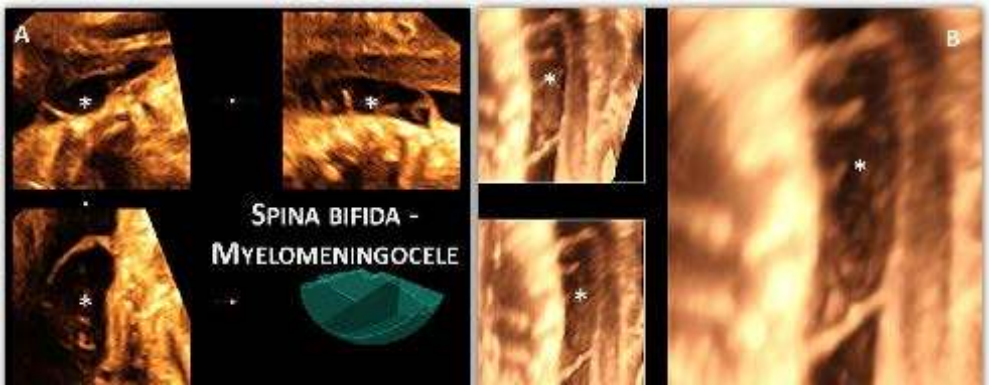

Figure 4. The three–dimensional ultrasound of myelomeningocele (*).
A –3D multiplanar view. B – 3D surface view.

All three findings – absent cavum septi pellucidi, uninterrupted continuity of interhemispheric fissure and colpocephaly, were significant for the agenesis of the corpus callosum.

Fetal Spine

Evaluation of the spine as part of neurosonographic examination was performed using a combination of axial, coronal and sagittal planes, using two–/ and three–dimensional techniques.

In this case, the examination of the fetal spine revealed a 27.9x11.9 mm cystic expansion in the lumbosacral area, which was diagnosed as spina bifida (Figure 4). The diagnosis of myelomeningocele in a fetus with the hindbrain findings and supratentorial abnormalities were significant for the diagnosis of the Chiari II malformations.

Fetal Genitalia

The sonographic determination of the fetal gender showed an abnormally curved and shortened fetal penis between the two scrotal folds in the coronal view (Figure 5).The aspect of the penis contrasted markedly with the expected image of the penis.Ambiguous genitalia were excluded with 3D ultrasound (Figure 6/A).The ultrasound image resembled the tulip sign described by Meizner and the diagnosis of hypospadias was suspected.

Fetal Face

The 3D surface display of the fetal facedid not diagnose any pathological changes (Figure 6/B).

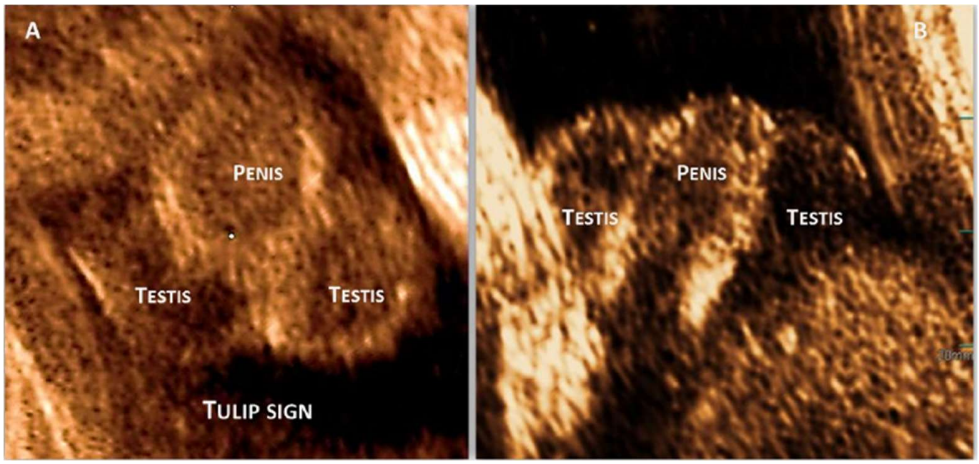

Figure 5. The tulip sign.
A – The coronal view and B – The transversal view: The tulip flower is represented by the ventrally bent penis, located between the two testicles.

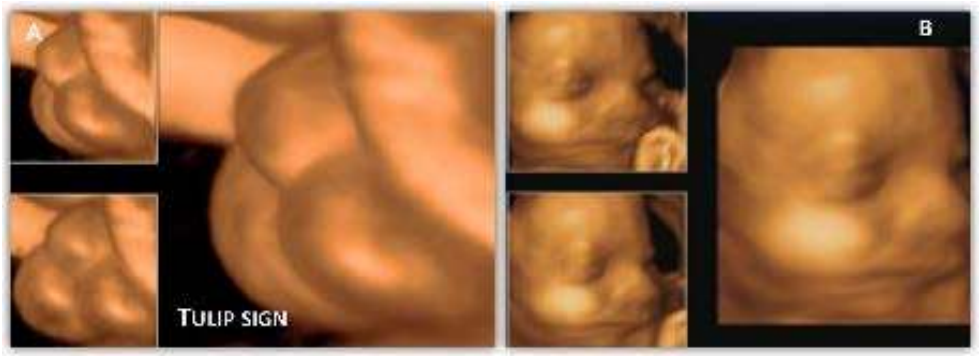

Figure 6. Three–dimensional ultrasound images.
A – The tulip sign: abnormally curved and shortened fetal penis between the two testicles. B – Fetal face without any pathological changes.

On the basis of these sonographic findings, prenatal diagnosis of the Chiari II malformation with ventriculomegaly, agenesis of the corpus callosum and hypospadias was made.

THERAPEUTIC INTERVENTION/OUTCOME

The patient had been informed in detail about her ultrasound findings, the possibilities and limitations of ultrasound diagnosis and the possible consequences, after which she decided to continue further monitoring of the pregnancy at the University Department of Obstetrics and Gynecology, Petrova 13, Zagreb.

During the clinic monitoring, the fetal MRI was performed at the University Department of Diagnostic and Interventional Radiology in Zagreb, which confirmed the suspicion of Chiari II malformation with hydrocephalus and agenesis of the corpus callosum. At 38 weeks' gestation, a term male newborn (3,130 g; 49 cm) was delivered by Cesarean section. The newborn was confirmed to have Chiari II malformation with hydrocephalus and corpus callosum agenesis. Ultrasound documentation was compared with postnatal clinical findings.

Clinical examination of the external genitalia confirmed male gender. The testicles were descended into the scrotum but the short penis showed atony. The external urethral meatus was of normal appearance and located in the normal position in contrast to hypospadias with morphological abnormalities of the urethra. Although the appearance of the external genitalia at birth corresponded to prenatally made 3D ultrasound images, postpartal examination did not confirm prenatal suspicion of hypospadias.

In the telephone conversation 18 months after birth, the mother stated that the boy showed a delay in psychomotor development with nystagmus. He had also developed a neurogenic bladder– and anal incontinence.

The author assumed that, in this case, prenatal ultrasound findings of the tulip sign and ventral flexion of the penis were the result of neurologically induced atony of the penis due to spina bifida, and breech presentation of the fetus with the co–work of the gravitational force.

DISCUSSION

The Chiari type II malformation is characterized by myelomeningocele followed by cerebellar hypoplasia and varying degrees of caudal displacement of the lower brainstem into the upper cervical canal through the foramen magnum. Myelomeningocele is a complex congenital spinal anomaly that results from failed closure of the caudal end of the neural tube, resulting in an open lesion or sac that contains dysplastic spinal cord, nerve roots, meninges, vertebral bodies and skin.

This deformity impedes the flow and absorption of cerebrospinal fluid and causes hydrocephalus, which occurs in more than 90% of infants with myelomeningocele. Cerebral cortex dysplasia, including heterotopias, polymicrogyria, abnormal lamination, fused thalami, and corpus callosum abnormalities, also occurs frequently. Mesodermal structures surrounding the neural tube, such as the vertebra and ribs, may also be malformed.

Commonly associated anomalies are facial clefts, heart malformations, and genitourinary tract anomalies. Urinary tract anomalies, such as solitary kidney or malformed ureters, may contribute to increased morbidity in the presence of neurogenic bladder dysfunction.

The ability of sonography to show fetal myelomeningocele has improved steadily and the greatest improvements occurred when the focus of diagnosis shifted from observation of the spina bifida to the cranial findings associated with the Chiari II malformation [22-28]. The Chiari II malformation results in a number of cranial abnormalities that can be sonographically imaged prenatally. The most useful features of the Chiari II malformation are the inftratentorial findings, which include effacement of the cisterna magna and the so-called 'banana sign'. The banana sign consists of displacement of the inferior vermis, lower pons and medulla into the upper cervical canal with an associated elongated fourth ventricle with ultrasound finding of the flattening of the cerebellum that often results in an appearance simulating a banana. Tonsillar herniation, when the cerebellar tonsils move downward through the foramen magnum, also called downward cerebellar herniation, transforaminal herniation or 'coning', may result in an 'empty' posterior fossa. This is often attributable to tethering of the spinal cord at the site of the myelomeningocele with downward displacement of the brain as the fetus grows.

Although the dominant abnormalities of Chiari II malformation are related to the cerebellum, many supratentorial features have also been described. The important ones among them are the lemon sign, polymicrogyria, callosal dys-/agenesis, ventriculomegaly, colpocephaly, the ventricular or occipital point, the interhemispheric cyst, beaked tectum and the 'too-far-back ventricle'.

Given the wide range of anatomical severity as well as the large number of associated abnormalities which are sometimes encountered, it should be no surprise that the clinical presentation of patients with Chiari II malformations is also varied both in character and severity.

Neurogenic incontinence is one of the conditions, that could not be seen by ultrasound examination, but this chapter demonstrates that the 'tulip sign' is not only specific for hypospadias, but in some special conditions could indicate atony of the penis and neurogenic incontinence in the differential diagnosis.

The tulip sign, previously described as "a unique and specific sonographic sign for prenatal detection of severe penoscrotal hypospadias", formed by the ventrally bent penis, located between the two scrotal folds, was accidentally found in the fetus with Chiari II malformation in breech presentation, with postnatally confirmed neurogenic bladder– and anal incontinence. In this case, the ventral flexion of the penis was probably the result of a neurologically induced atony of the penis with the co–work of the gravitational force in this specific position, where the fetus with spina bifida was in breech presentation.

PATIENT PERSPECTIVE

During the first postpartal week, the myelomeningocele was surgically repaired. Thereafter, the cerebellum spontanously returned to its normal position and then a shunt was surgically inserted into the lateral cerebral ventricle.

At the age of 18 months, the boy had a delay in psychomotor development and nystagmus. The neurogenic bladder– and anal incontinence were developing during the postnatal period.

Informed Consent: The patient provided informed consent for this case report.

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