Spinal Sonography in Infants with Cutaneous birth Markers in the Lumbo-Sacral Region – an Important Sign of Occult Spinal Dysrhaphism and Tethered Cord

Spinale Sonografie bei Säuglingen mit kutanen lumbosakralen Auffälligkeiten – ein wichtiger Hinweis auf okkulte spinale Dysrhaphien und Tethered Cord

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Key words
- spine
- occult spinal dysrhaphism
- tethered cord
- diastematomyelia
- lipomyelomeningocele
- myelocystocele
- cutaneous birth markers

Zusammenfassung

Ziel: Kutane Stigmata im lumbosakralen Bereich können Hinweise auf okkulte spinale Dysrhaphien sein. Mit der spinalen Sonografie können anatomische Auffälligkeiten des Rückenmarks in der Neonatalperiode erkannt werden.

Patienten: Wir berichten über 6 Säuglinge mit kutanen lumbosakralen Auffälligkeiten, die mit einem hochauflösenden Linearschallkopf (> 7,5 MHz) zwischen dem 1. Lebenstag und der 9. Woche (m: 26 Tage) untersucht wurden. Bei den Kindern bestanden folgende kutane Auffälligkeiten: asymmetrische rima ani (4); lumbale Weichteilschwellung (2); Dermalsinus (2); Haarnaevus (1); Pigmentnävus (1); häutiges Hautanhängsel (1); Hämangiom (1) sowie einseitige Peroneuslähmung mit trophischer Störung der unteren Exremität.

Ergebnisse: Folgende Diagnosen wurden gestellt: tethered cord (6); Diastematomyelie (2); tight filum terminale (2); spinales Lipom (3); Lipomyelomeningocele (2); Myelocystocele und Hydromyelie (1). Bei allen Kindern konnte die sonografische Diagnose kernspintomografisch und bei der operativen Korrektur im Alter von 2 bis 12 Monaten (m: 7,7 Monate) bestätigt werden.

Schlussfolgerung: Bei allen Kindern mit kutanen lumbosakralen Auffälligkeiten sollte das Rückenmark in der Neonatalperiode untersucht werden, um okkulte spinale Dysrhaphien und ein tethered cord frühzeitig zu diagnostizieren und die operative Korrektur vor Einsetzen neurologischer Störung gegen Ende des 1. Lebensjahres durchführen zu können.

Abstract

Aim: Cutaneous markers in the lumbo-sacral region are indicators of occult spinal dysrhaphism and tethered cord. By means of spinal sonography, anatomical abnormalities of the spinal cord can be shown in the neonatal period.

Patients: We report on 6 infants with lumbo-sacral cutaneous abnormalities who were investigated with a high resolution linear transducer (> 7.5 MHz) and a computer sonographic unit (Sequoia, Acuson). The investigations were performed between the first day of life and the ninth week (m: 26 days). The following cutaneous markers could be found: Asymmetrical gluteal crease (4); dermal sinus (2), hairy tuft (1); pigmented naevus (1); cutaneous appendage (1); haemangioma (1); unilateral peroneal paralysis with hypotrophic correspondic leg (1).

Results: Sonographic evaluation showed the following abnormalities: Tethered cord (6); diastematomyelia (2); tight filum terminale (2); spinal lipoma (3); lipomyelomeningocele (2), myelocystocele and hydromyelia (1). In all infants, sonographic diagnosis could be confirmed by MR imaging and intraoperatively. Surgical correction was performed at the age of 2 to 12 months (m: 7.7 months).

Conclusion: All infants with cutaneous markers in the lumbo-sacral region should be investigated by spinal sonography as long as the vertebral arches are not completely ossified. Sonography of the spinal cord may detect occult spinal dysrhaphism and tethered cord and prevent neurological damage by early surgical correction at the end of the first year of life.

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Introduction

The vertebral arches of newborns and young infants are not completely ossified. They create an acoustic window and allow the sonographic imaging of the spinal cord. High resolution linear transducers with a transmission frequency of > 10 MHz allow excellent detailed imaging

of the spinal cord, the anterior and posterior nerve roots and the cauda equina. Spinal sonography is the imaging modality of choice in the neonatal period for the diagnosis of spinal dysrhaphism.

Spinal dysrhaphism can be divided into three categories based on the presence or absence of a mass on the back [1, 2]. The first category consists of spinal dysrhaphism with a non-skin-covered mass on the back. The second category is spinal dysrhaphism with a skin-covered mass on the back, the third category spinal dysrhaphism without an associated mass on the back (occult spinal dysrhaphism), which encompasses the largest group of anomalies. The first and second category can be diagnosed clinically, whereas the third category of occult spinal dysrhaphism may easily be overlooked. More than 50% of infants with occult spinal dysrhaphism, however, display cutaneous markers in the lumbosacral region which are strong indicators of spinal abnormalities [1, 3 – 9].

All infants with lumbosacral markers should be investigated in the neonatal period by spinal sonography to rule out occult spinal dysrhaphism and associated spinal cord tethering.

The following paper exemplifies the cases of 6 infants with lumbosacral cutaneous abnormalities and associated occult spinal dysrhaphism who were diagnosed by spinal sonography in the neonatal period and first months of life.

Patients and method

The six infants (5 girls and 1 boy) were born between the 38th and 40th week of gestation (m: 39 ± 0.6 week) with a mean birth weight of 3523 ± 77 g and a body length of 53 ± 1.7 cm. The patients showed cutaneous abnormalities in the lumbosacral region, which are listed in ● **Table 1**. The spinal cords of all infants were investigated with a high resolution linear transducer with a frequency of > 7.5 MHz (Sequoia, Acuson). The investigations were performed in prone position.

In all infants, sagittal sections through the midline and additional transverse sections through the thoracic and lumbar spine were performed. In elder infants with already ossified vertebral arches, parasagittal sections were used. The location of the medullary conus was determined by counting the vertebral bodies from the coccygeal tip upwards as suggested by Beek [10 – 13].

The investigations were performed between the 1st day and the 9th week of life with a mean age of 26 ± 32 days. In all infants, occult spinal dysrhaphism could be shown. The sonographic diagnosis was confirmed by MR-imaging before neurosurgical intervention in all patients.

Corrective surgery was performed between the 2nd and 12th month of life with a mean age of 7.7 months.

Table 1 Cutaneous abnormalities in six patients with occult spinal dysrhaphism[1]

abnormality	n
– asymmetrical gluteal crease	4
– asymmetrical lumbosacral soft tissue mass	2
– dermal sinus	2
– hairy tuft	1
– pigmented naevus	1
– cutaneous appendage	1
– capillary haemangioma	1
– paralysis of the peroneal nerve	1
– hypotrophy of corresponding lower extremity	1

[1] In some infants, more than one abnormality could be found.

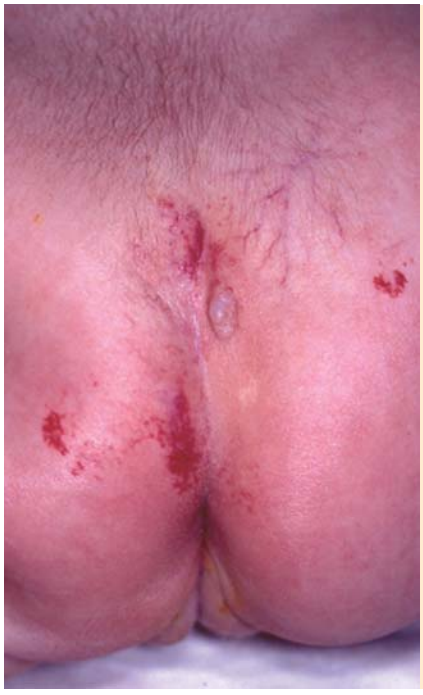

Fig. 1 Patient 1: Multiple capillary haemangiomas in the lumbosacral area. Small area of atrophic skin and cutaneous appendage. Asymmetric gluteal cleft.

Abb. 1 Patientin 1: multiple kapilläre kutane Hämagiome lumbosakral. Zusätzlich atrophisches Hautareal sowie kleines Hautanhängsel und asymmetrische Rima ani.

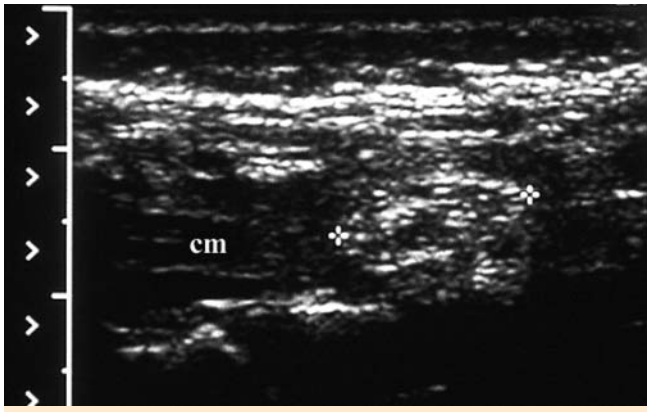

Fig. 2 Patient 1: Longitudinal section through the lumbar spine. Intraspinal echogenic mass (lipoma) in continuity with the cauda equina (+). Low-lying medullary conus (cm), indicating tethered cord.

Abb. 2 Patientin 1: Längsschnitt durch das lumbale Rückenmark. Intraspinale Raumforderung (Lipom) in Verbindung mit der Cauda equina (+). Tiefer Konusstand (cm) im Sinne eines tethered cord.

Results

The sonographic abnormalities and the definitive diagnosis revealed during surgical correction are listed in ● **Table 2**.

- Patient 1 (G.S.) (gestational age 39 weeks, birth weight: 3550 g, length: 51 cm)

  The female patient was investigated at the age of 9 weeks due to several cutaneous markers in the lumbar region to exclude spinal dysrhaphism.

  The girl showed several capillary haemangiomas and a small cutaneous appendage as well as an additional small area of aplasia cutis (● **Fig. 1**). The gluteal crease was asymmetrical due to a subcutaneous mass.

  Spinal sonography showed a subcutaneous echogenic mass which extended into the spinal canal in continuity with the

Table 2 Survey of the cutaneous markers, sonographic findings and definitive (intraoperative) diagnosis

patient	cutaneous markers	sonographic diagnosis	intraoperative diagnosis and surgical correction
1	– cutaneous appendage – haemangioma – asymmetric gluteal crease	– spinal lipoma – tethered cord	– spinal lipoma – resection of lipoma at age of 11 months – myelolysis
2	– peronal paralysis – hypotrophic left leg – asymmetric gluteal crease	– hydromyelia-syringomyelia – myelocystocele	– terminal myelocystocele – hydromyelia – myelolysis at age of 3 months – syringo-subarachnoid shunt
3	– subcutaneous mass – asymmetrical gluteal fold	– spinal lipoma – tethered cord	– resection of lipoma at age of 12 months – myelolysis by resection of filum terminale
4	– subcutaneous left sided mass – asymmetrical gluteal fold	– subcutaneous lipoma and spinal lipoma – lipomyelomeningocele	– sacral lipomyelo-meningocele – tethered cord – resection of lipoma and untethering at age of 5,5 months
5	– pigmented naevus	– diastematomyelia – tight filum terminale – tethered cord	– diastematomyelia type I – pathologic filum terminale – resection of osseus spur – resection of fila terminalia
6	– hairy tuft	– diastematomyelia – spinal lipoma – tight filum terminale – tethered cord	– diastematomyelia type II – pathologic filum terminale – untethering age 7 months

cauda equina (**Fig. 2**). The medullary conus was localised low down in the region of S2/S3, causing tethered cord.

Spinal MRI demonstrated spina bifida in the sacral region and a sacral lipoma. In addition, low position of the medullary conus could be shown.

Surgery at the age of 11 months confirmed the diagnosis. The lipoma was partially resected and myelolysis performed without complications. The infant has no neurological deficits.

- Patient 2 (S.A.) (gestational age 38 weeks, birth weight: 3540 g, length 53 cm)

The female patient was transferred to our department at the age of 8 weeks due to peroneal paralysis of the left leg since birth. The muscles of the left leg were hypotrophic (**Fig. 3a, b**). The gluteal cleft was asymmetrical.

Spinal sonography showed a polycyclic echo-free mass measuring 29 × 18 mm (**Fig. 4**). The cystic mass extended into a dilation of the central canal due to hydromyelia (**Fig. 4**). The sonographic diagnosis was terminal myelocystocele and tethered cord.

Spinal MRI showed sacral dysplasia and low lying spinal cord. In addition, a presacral cystic mass and syringohydromyelia up to Th5 could be shown.

Surgery was performed at the age of 3 months.

Surgical diagnosis was terminal myelocystocele with a bony defect within the sacral bone and syringomyelia with extension up to the thorax. Myelolysis and a syringo-subarachnoid shunt were performed.

The patient showed persisting peroneal paralysis of the left leg and a neurogenic bladder.

- Patient 3 (J.F.) (gestational age 40 weeks, birth weight 3460 g, length 54 cm)

The female patient was transferred for spinal sonography at the age of 5 days due to an asymmetrical gluteal crease and a subcutaneous mass in the gluteal region (**Fig. 5**)

Spinal sonography showed an intraspinal echogenic mass localised in the region of the cauda equina, due to spinal lipoma. In addition, the medullary conus could be shown in a low position in the region of L4/L5. Sonographic diagnosis was spinal lipoma and tethered cord (**Fig. 6**).

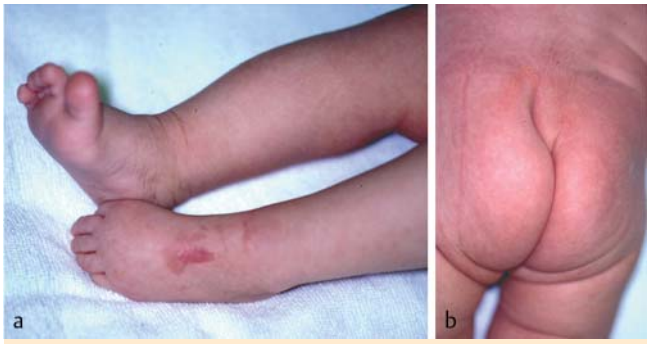

Fig. 3 **a**, **b** Patient 2: Peroneal paralysis of the left leg. Hypotrophic left leg and foot. Asymmetric gluteal cleft and discrete gluteal mass.

Abb. 3 **a**, **b** Patientin 2: Peroneuslähmung und Hypotrophie des linken Beines und Fußes. Asymmetrische Rima ani und diskrete gluteale Raumforderung.

Spinal MRI confirmed the sonographic diagnosis of a subcutaneous lipoma in continuity with a spinal lipoma and tethered cord.

Surgery was performed at the age of 12 months. Surgical diagnosis was spinal lipoma and tethered cord. Partial resection of the lipoma was performed, a tight filum was resected and untethering performed.

- Patient 4 (H.L.) (gestational age 40 weeks, birth weight 3630 g, length 56 cm)

The male patient was transferred for spinal sonography on day 10 because of a low lying left sided mass on the back and an asymmetrical gluteal cleft (**Fig. 7**).

Spinal sonography showed a subcutaneous echogenic mass in the region of the cauda equina with extension to the spinal canal. The medullary conus was localised at the L4/L5 level (**Fig. 8**). Sonographic diagnosis was lipomyelomeningocele and tethered cord.

Spinal MRI confirmed the sonographic diagnosis of a lipomyelomeningocele with sacral lipoma and tethered cord. In addition, sacral dysplasia could be found.

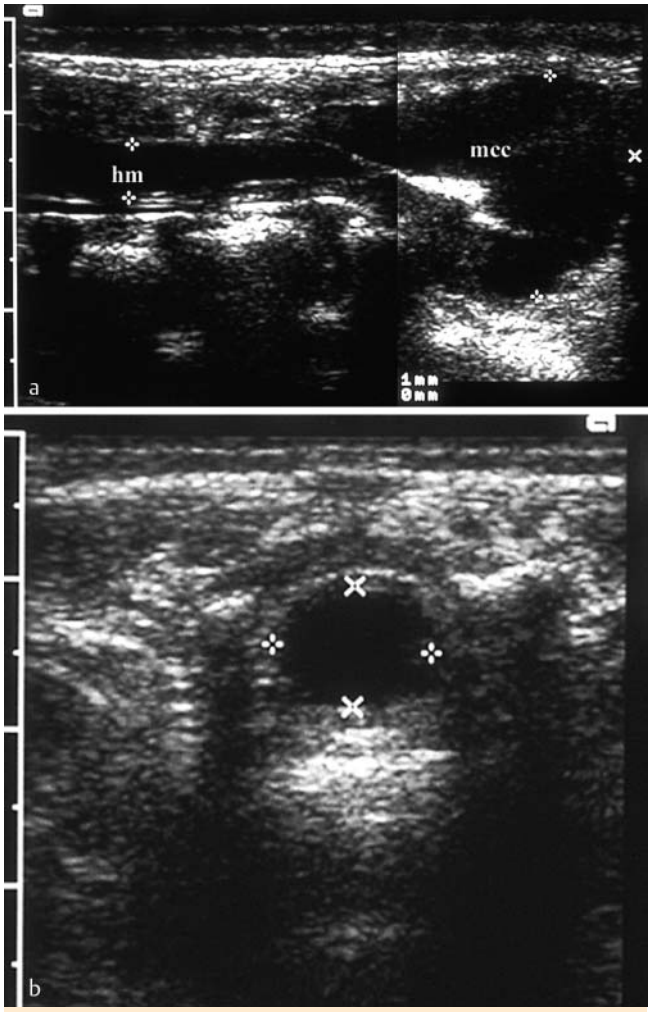

Fig. 4 **a** Patient 2: Longitudinal section through the lumbo-sacral area shows a cystic mass (mcc = myelocystocele) which is connected with a dilated central canal (hm = hydromyelia). Diagnosis: terminal myelocystocele (mcc) and associated hydromyelia (hm). **b** Patient 1: Transverse section through the lower thoracic spine: dilation of the central canal (hydromyelia) (markers).

Abb. 4 **a** Patientin 2: Längsschnitt lumbosakral: Darstellung einer zystischen Raumforderung (mcc = Myelozystocele), die sich nach ventral erstreckt und mit einem erweiterten Zentralkanal (Hydromyelie) (hm) kommuniziert. Diagnose: terminale Myelozystozele und Hydromyelie.
b Patientin 2: Querschnitt durch das untere Thorakalmark: deutliche Erweiterung des Zentralkanals (Hydromyelie) (Markierung).

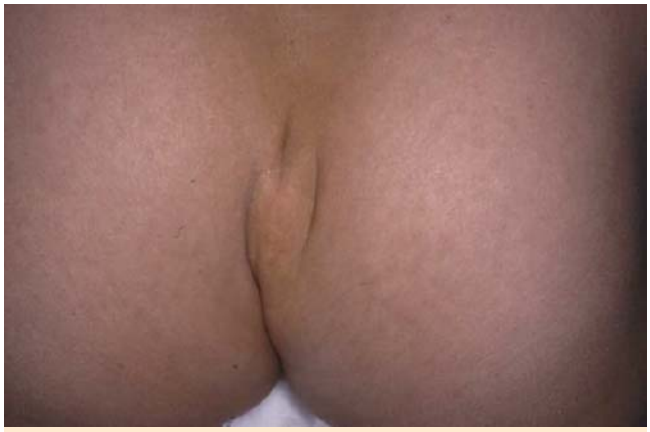

Fig. 5 Patient 3: Asymmetric gluteal crease and subcutaneous gluteal mass.

Abb. 5 Patientin 3: Asymmetrische Rima ani sowie subkutane gluteale Raumforderung.

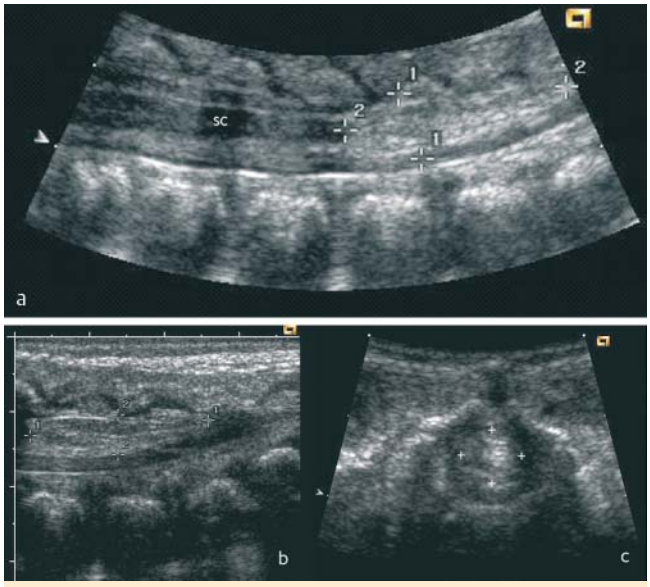

Fig. 6 **a–c** Patient 3: Longitudinal **a**, **b** and transverse section **c** through the lumbar spine shows an echogenic mass (lipoma) in the region of the cauda equina (markers), causing tethered cord. sc = spinal cord.

Abb. 6 **a–c** Patientin 3: Längsschnitt **a**, **b** und Querschnitt **c** durch die lumbale Wirbelsäule: echogene Raumforderung (Lipom) im Bereich der Cauda equina (Kreuze), die zum tethered cord geführt hat. sc = Rückenmark.

Surgery performed at the age of 5.5 months confirmed the diagnosis of sacral lipomyelomeningocele and tethered cord.
Partial resection of the lipoma and untethering was performed. The child is doing well without neurological deficits.

- Patient 5 (Z.L.) (gestational age 39 weeks, birth weight 3550 g, length 53 cm)

The female patient was transferred to our unit at the age of 6 days because of a pigmented naevus in the lumbosacral region (**Fig. 9**).
Spinal sonography showed diastematomyelia from L2 to L4 with low lying medullary conus ending at L4 (**Fig. 10**). The cord was fixed in the low position by two echogenic fila terminalia.
Sonographic diagnosis was diastematomyelia and tethered cord.
Spinal MRI confirmed the diagnosis of diastematomyelia from L2 to L4, associated with an osseous spur in the region of L4 and tight fila terminalia causing tethered cord.
Surgery was performed at the age of 10 months. During surgery, the sonographic and MRI diagnoses were confirmed. The osseous spur and the pathologic fila were resected and untethering performed.
The child is doing well without neurological deficits.

- Patient 6 (S.J.) (gestational age 40 weeks, birth weight 3410 g, length 52 cm)

The girl was born in our department with a hairy tuft in the lumbo-sacral region (**Fig. 11**). No other abnormalities could be found. The infant showed no neurological abnormalities.

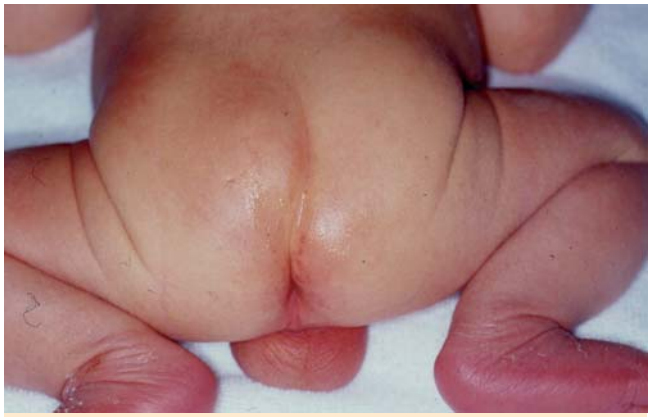

Fig. 7 Patient 4: Dorsal mass low on the left buttock and asymmetrical gluteal crease.

Abb. 7 Patient 4: Subkutane gluteale Raumforderung im Bereich der linken Gesäßbacke und asymmetrische Rima ani.

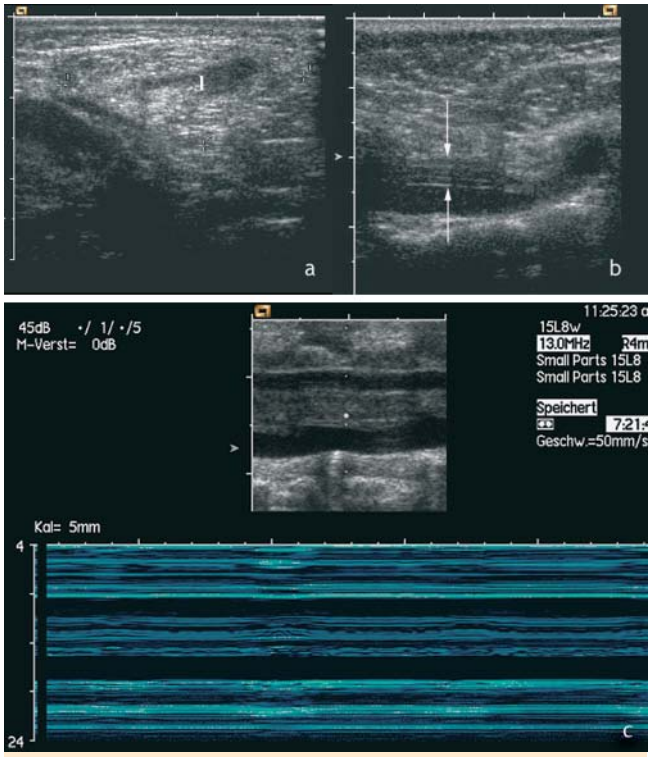

Fig. 8 **a**, **b** Patient 4: Paraspinal echogenic mass (l = lipoma) which communicates with the spinal cord (arrows), representing lipomyelomengingocele. Low lying and deformed medullary conus due to tethered cord. **c** Patient 4: M-mode of the low-lying spinal cord shows no oscillations due to tethering of the cord.

Abb. 8 **a**, **b** Patient 4: Paraspinale echogene Raumforderung (l = Lipom), die mit dem Rückenmark (Pfeile) im Sinne einer Lipomyelomengocele kommuniziert. Tief stehender und deformierter Konus medullaris im Sinne eines tethered cord. **c** Patient 4: M-mode des tiefstehenden Konus medullaris zeigt keine Oszillationen aufgrund eines tethered cord.

Spinal sonography performed on day 1 showed diastematomyelia from L1 to L4 and associated tight filum terminale, filar lipoma and tethered cord.

Spinal MRI confirmed the sonographic diagnosis. In addition, lumbo-sacral dysplasia and incomplete closure of the lumbar vertebral arches L4 and L5 could be shown.

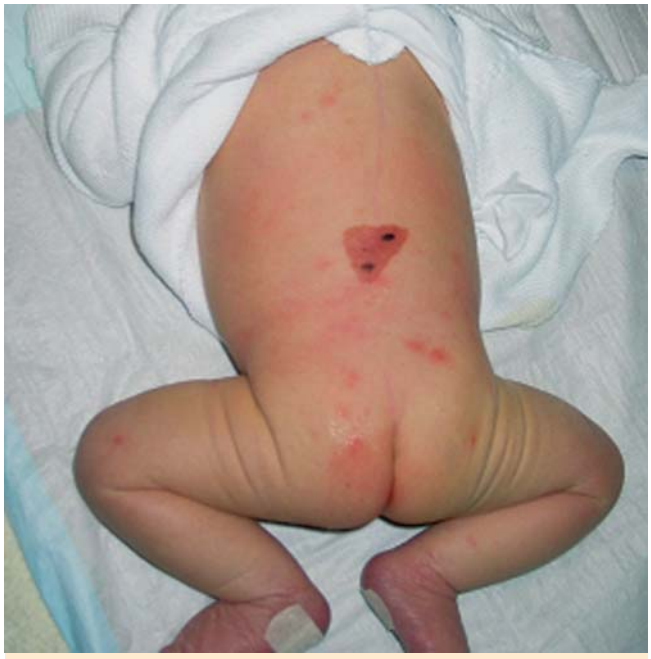

Fig. 9 Patient 5: Pigmented naevus in the lumbar region.

Abb. 9 Patient 5: Lumbaler Pigmentnaevus.

Surgical correction was performed at the age of 7 months. The intraoperative diagnosis was diastematomyelia type II, tight filum terminale and filar lipoma. The tight filum was transsected and untethering performed.

Discussion

Spinal cord development occurs through three consecutive periods: Gastrulation (weeks 2 – 3), primary neurulation (weeks 3 – 4), secondary neurulation and regressive differentiation (weeks 5 – 6) [1, 2]. Defects in these early embryological stages may produce spinal dysrhaphism, characterised by anomalous differentiation and fusion of dorsal midline structures. According to Tortori-Donati, spinal dysrhaphism can be categorised clinically into open and closed spinal dysrhaphism [1, 2]. In open spinal dysrhaphism, the neural tissue is exposed to the environment. Closed spinal dysrhaphism is covered by skin, although cutaneous stigmata usually indicate its presence. The overlying skin is often dysplastic. In closed spinal dysrhaphism, two subsets may be distinguished based on whether a subcutaneons mass is present in the low back or not [1, 2].

Open spinal dysrhaphism includes the following diseases: myelomeningoceles, myeloschisis, hemimyelomeningoceles and hemimyeloceles [1, 2].

Closed spinal dysrhaphism with mass comprises the common lipomas with a dural defect, lipomyeloschisis and lipomyelomengioceles and the incommon meningoceles and terminal myelocystoceles [1, 2]. Closed spinal dysrhaphism (CSD) with a subcutaneous mass represents 18.8% of all CSD. Lipomas with dural defect account for 87.4% of all CSD with a subcutaneous mass [2]. Almost all of these abnormalities occur in the lumbosacral spine [2].

Closed spinal dysrhaphism without a mass comprises a number of complex dysraphic states ranging from complete dorsal enteric fistula to neurenteric cyst, split cord malformation, der-

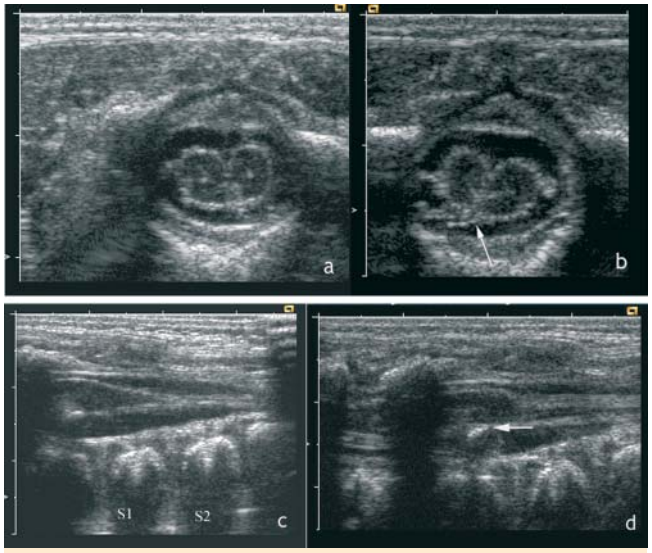

Fig. 10 **a**, **b** Patient 5: Transverse section of the spinal cord shows duplication of the spinal cord due to diastematomyelia. The anterior and posterior root of the right hemicord originate correctly on the right side, whereas the anterior root of the left hemicord originates falsely on the right side of the spine (arrow). **c**, **d** Longitudinal section through the lumbo-sacral spine. Low position of the medullary conus, terminating in the sacral region at the level of S1/S2. Osseus spur associated with diastematomyelia (arrow).

Abb. 10 **a**, **b** Patientin 5: Querschnitt durch das Lumbalmark. Komplette Duplikatur des Rückenmarks im Sinne einer Diastematomyelie. Die Radix anterior und posterior des rechten Rückenmarksanteils entspringen regelrecht auf der rechten Seite, während die Radix anterior des linken Rückenmarksanteils ebenfalls rechts entspringt (Pfeil). **c**, **d** Längsschnitt lumbosakral. Tiefer Konusstand, der in Höhe S1/S2 endet. Knöcherner Sporn (Pfeil), der mit der Diastematomyelie assoziiert ist.

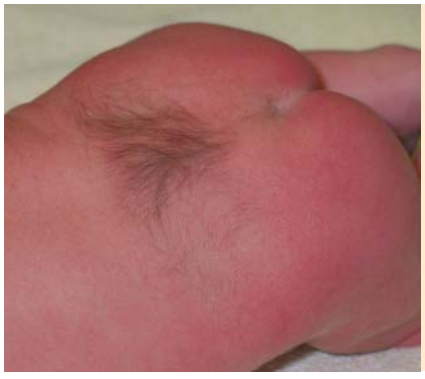

Fig. 11 Patient 6: Hairy tuft in the lumbar region.

Abb. 11 Patienin 6: Lumbaler Haarnaevus.

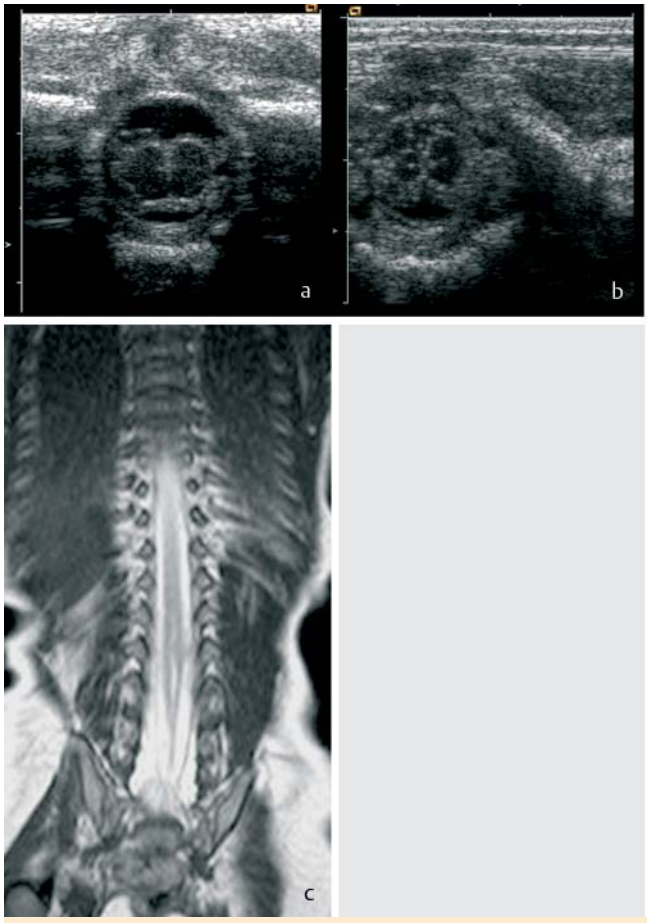

Fig. 12 **a**, **b** Patient 6: Transverse section through the lumbar spine shows duplication of the medullary conus of the spinal cord due to diastematomyelia. The right hemicord is smaller and ends higher than the left one. **c** MRI image of diastematomyelia. It shows complete duplication of the conus medullaris.

Abb. 12 **a**, **b** Patientin 6: Querschnitt durch das Lumbalmark: Duplikatur des Konus medullaris des Rückenmarkes bei Diastematomyelie. Der rechte Rückenmarksanteil ist deutlich schwächer ausgebildet und endet höher als der linke Anteil. **c** MRI der Diastematomyelie. Das Bild zeigt eine komplette Duplikatur des Konus medullaris.

mal sinus, caudal regression and spinal dysgenesis to tight filum terminale, filar and intradural lipoma and persisting terminal ventricle [1, 2].

Whereas the diagnosis of open spinal dysrhaphism is apparent, closed spinal dysrhaphism without a mass may be overlooked especially easily. This may be fatal as all cases of spinal dysrhaphism may be associated with a tethered cord [1, 2, 8, 14, 15]. Tethered cord is related to traction of the spinal cord and distortion or kinking of the small arteries, veins, capillaries and nerve fibers of the spinal cord. Metabolic impairment may result in functional deterioration of spinal cord nerve cells. Progressive neurological deterioration including motor and sensory dysfunction of the lower extremities, muscular atrophy, decreased or hyperactive reflexes, urinary incontinence, spastic gait or orthopaedic deformities such as scoliosis or foot and hip deformities may occur [1, 2, 14 – 16]. As most of these neurological symptoms are not reversible, early diagnosis before the commencement of deterioration is essential [14].

Diagnosis of open spinal dysrhaphism is easy due to the typical clinical malformations which cannot be overlooked. Clinically, myelomenigoceles are characterised by elevation of the neural placode by underlying expanded subarachnoid space, whereas in myeloschisis the placode is flush with the back surface. Such differentiation is a clinical matter, and usually, no imaging modality needs to be employed.

Diagnosis of closed spinal dysrhaphism may be difficult in the individual case especially in the absence of a mass on the back. Often, these malformations are not clinically evident at birth. Patients attract clinical attention later in infancy when complications related to tethered cord occur.

In general, however, careful clinical examination may identify more than half of these patients due to numerous cutaneous markers on the back associated with spinal dysrhaphism.

A critical factor is the presence of a subcutaneous mass located at the lumbosacral level of the patient's back. Four malformations will present with a mass on the back: lipomyeloschisis, lipomyelomeningocele, meningocele and terminal myelocysteocele.

Cases of closed spinal dysrhaphism without a subcutaneous mass on the back may benefit from a thorough clinical assessment of the back. 50% of cases of occult spinal dysrhaphism are associated with cutaneous markers [8]. On the other hand, 3–8% of all infants with skin abnormalities in the region of the lower spine suffer from occult spinal dysrhaphism [8]. Most infants show cutaneous markers associated with spinal malformations: focal hirsutism, capillary haemangioma, dermal sinus, rudimentary "tail" and atretic meningocele [1, 3–9]. These cutaneous birth markers may be isolated but often occur in combination.

It is not surprising that malformations of the skin and central nervous system occur simultaneously, as during closure of the neural tube, the ectoderm differentiates into neural and epithelial tissues in early pregnancy [2, 16].

The most common skin marker is a simple midline dimple found in 74% of patients with cutaneous abnormalities [7]. If these dimples are localised low down in the sacrococcygeal region and are hidden by the gluteal cleft, they are usually not associated with occult spinal dysrhaphism and tethered cord. If these cutaneous pits are localised at the level of the coccyx and the skin at their base is intact, they need no further investigation [8]. The higher they are localised, the more likely they are to be associated with spinal dysrhaphism.

If the bottom of the dimple cannot be seen, if associated skin abnormalities are present or if they are localised above the gluteal crease, they should be investigated by spinal sonography [7, 8]. Simple midline dimples or pits have to be distinguished from atypical dimples which are large (> 5 mm), occur higher up on the back (> 2.5 cm from the anus) or appear together with other lesions [7] often associated with spinal dysrhaphism. The lesion which is most clearly associated with underlying intradural pathology is focal hirsutism or hairy tufts localised over the thoracic or lumbar spine [1, 2]. In more than 50% of these infants, diastematomyelia is the associated spinal malformation [1].

Of the cutaneous birthmarks, the least sensitive in predicting underlying spinal dysrhaphism is capillary haemangioma [1]. Only 10% of capillary haemangiomas in the lumbar region are associated with underlying spinal pathology [1]. Further indicators of spinal dysrhaphism include deformities of the lower extremities such as clubfoot, asymmetry of length and muscle mass of the legs or feet, anorectal and genitourinary malformations [1, 7, 17, 21].

As lumbosacral cutaneous markers and mass on the back are indicators of underlying spinal dysrhaphism and tethered cord, these infants should be thoroughly investigated by MRI imaging or spinal sonography.

In the neonatal period, the vertebral arches are not completely ossified. This offers an acustic window for the sonographic evaluation of the spine to rule out spinal dysrhaphism and tethered cord. The study of Gusnard and coworkers showed good correlation between cryomictrotome, CT and spinal sonography [18]. Several comparative studies between MRI and sonography have shown excellent correlation [19–21]. Sonography needs no sedation and is much cheaper than MRI. It should be performed as early as possible before the vertebral arches and spinous processes ossify. Several sonographic reports have described the normal anatomy of the spinal cord [10–13, 19, 22–28]. At birth, the medullary conus is situated above the level of L2/L3 [5, 10, 12, 13, 24]. If the medullary conus is located below L3, tethered cord should be suspected. To determine the end of the medullary conus, the echogenic vertebral bodies should be visualised. In newborns with a straight lumbar spine, the promontorium forms an angle with the convex sacral spine. By identifying the lordotic transition at the lumbosacral junction and counting the lumbar vertebra upwards, the level of the medullary conus can be determined exactly by ultrasound [10].

Beside the normal anatomy, the different forms of pathology such as spinal lipoma, lipomyelomeningoceles, myelocystoceles, hydromyelia and syringomyelia as well as tight filum terminale and diastematomyelia can be shown [7, 9, 19, 23, 25, 28–33].

Spinal lipomas may appear as echogenic or hypoechoic masses on sonography. They may be localised completely intradurally within the spinal canal and the filum terminale (filar lipoma). They may extend from the subcutaneous plane into the spinal canal through a defect in the vertebral arch and dura (lipomyelocele). The connections of the subcutaneous fibrofatty masses with the spinal cord may cause secondary spinal cord tethering.

In hydromyelia, the dilated central canal can be shown in longitudinal and transverse sections of the spine as an echofree area.

In cases of a terminal myelocystocele, the hydromyelic cord herniates through a posterior spinal defect into an echofree mengingocele. The spinal cord transverses the meningocele and inserts in its posterior wall. At the caudal end of the cord, the central canal opens into the cyst.

Tight filium terminale is an echogenic filum with a diameter of more than 2 mm which tethers the cord below the level of L3 [2]. Thickening is often caused by a fibro-fatty mass.

Diastematomyelia refers to a partial or complete sagittal clefting of the spinal cord, usually in the lumbar region. It may be associated with a fibrous, cartilaginous or bony spur tethering the cord at the level of the spur [16, 32]. The division of the cord into two hemicords can clearly be shown by spinal sonography in transverse sections. The divison may be complete or partial, and the two hemicords are not necessarily symmetrical. Often, thickening of the filum terminale and associated subcutaneous and/or spinal fibrolipoma and fibrous bands may be found.

Consequences

In the case of sonographically demonstrated pathology, the patient should be presented to a neurosurgeon experienced with spinal cord malformations. Surgical correction should be done before neurological deterioration occurs [14, 31]. Usually, surgical correction will be performed towards the end of the first year of life. In cases of neurological problems such as paresis of the legs and neurogenic bladder, surgical procedures have to be performed earlier.

As long as the vertebral arches are not completely ossified, spinal sonography should be the imaging modality of choice to rule out spinal dysrhaphism and tethered cord.

As the vertebral arches progressively ossify during the first year of life, spinal sonography is no longer possible.

As surgery is usually performed at the end of the first year of life, MRI of the spinal cord should be delayed until operative correction is planned. In the neonatal period, high resolution sonography constitutes a much better imaging modality than MRI.

Conclusion

Spinal dysrhaphism should be suspected in all infants with a mass on the back or cutaneous markers in the lumbo-sacral region such as haemangiomas, dermal sinuses, pigmented naevi, hairy tufts, skin appendages or atrophic cutaneous areas. These patients should be investigated by spinal sonography as early as possible during the first weeks of life, as long as the spinal arches are not completely ossified. All forms of spinal dysrhaphism including secondary spinal cord tethering can be diagnosed sonographically.

Early diagnosis is essential for adequate surgical correction before neurological deterioration has occurred.

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