



# Occult Spinal Dysraphism: Utility of Cutaneous Lesions as a Screening Tool for Early Diagnosis in a Series of 31 Cases

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## Abstract

**Background:** Occult spinal dysraphism (OSD) is a serious and progressive pathology whose prognosis depends on the type of malformation, and how early it is detected and managed. The neurosurgical approach is complex and still debated. However pluridisciplinary monitoring is important for neurological prognosis. The link between lumbosacral midline lesions and OSD is established.

The aim of this study is to analyze the utility of cutaneous lesions as a screening tool of occult spinal dysraphism and show the necessity for an early management of this pathology.

**Methods:** We performed a descriptive study on a series of 31 patients, aged 0-15 years, with congenital midline lumbosacral cutaneous lesions, without antenatal diagnosis referred for urodynamic evaluation.

**Results:** Neonatal Spinal Ultrasound was performed in only 12 patients; Spinal magnetic resonance imaging (MRI) confirmed OSD in 94%. We identified two groups, namely those with a subcutaneous mass, and those without. Average age at spinal MRI in patients with a mass was 3 months, versus 44 months in patients without subcutaneous mass ( $p < 0.004$ ). Complex malformations, which may be potentially severe, were not always associated with a subcutaneous mass.

**Conclusion:** Diagnosis was delayed in those without a subcutaneous mass. Lumbosacral examination should be systematically performed in the neonatal period to screen infants for those requiring spinal ultrasound before the age of 2 months, and if necessary, lesion work-up by full spinal MRI. This will enable early evaluation and multidisciplinary approach to feed the indication for neurosurgery and propose appropriate management aiming to maintain urinary, anal, renal, motor and spinal function.

**Keywords:** Occult spinal dysraphia; Cutaneous anomalies; Ultrasound; Magnetic resonance imaging; Neonatal; Urodynamic

## Abbreviations

MRI: Magnetic Resonance Imaging; OSD: Occult Spinal Dysraphism

## Introduction

Occult spinal dysraphism (OSD) is a heterogeneous group of congenital vertebral-spinal malformations arising from incomplete closure of the spinal bodies with the overlying skin present. The pathophysiology of OSD is not well known and the majority of OSD are thought to be a problem of secondary neurulation i.e. the development of the terminal part of the spinal cord. Normally neural tube closure commences when the two lateral parts of the tube meet. At a given point during the closure process, the cutaneous ectoderm separates from the neuro-ectoderm and fuses in the midline forming an ectoderm that covers the neural structures [1]. Most authors concur that OSD corresponds to premature separation of the neuro-ectoderm from the cutaneous ectoderm or incomplete disjunction of these two structures. Dermal sinus tracts and meningocele result from abnormal development of the neural plate during primary neurulation [1-2]. These defects are loco-regional, and usually without associated cerebral dysplasia.

The incidence of spinal dysraphism (both open and occult) is around 0.5 to 0.8 per 1,000 live births [3]. Since the advent of antenatal diagnostic techniques, the frequency of severe open spinal

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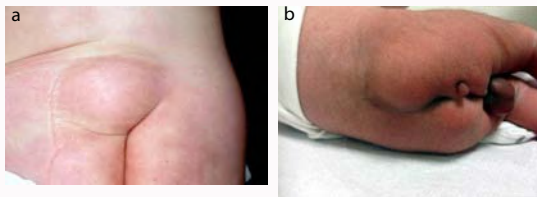


Figure 1: a) Mass. b) Associated Mass.

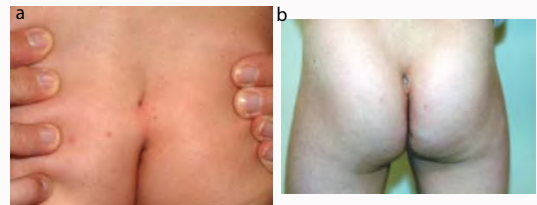


Figure 2a and 2b: Isolated dimple.

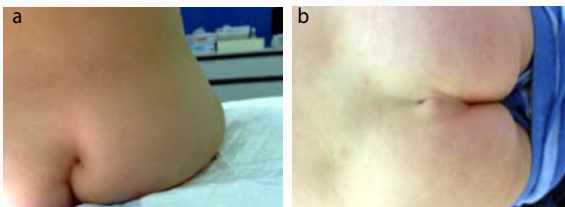


Figure 3a and 3b: Deviation of the gluteal fold and dimple.

dysraphisms has progressively decreased [4].

The diagnosis of OSD is guided by the clinical examination, particularly examination of the midline lumbosacral region. This examination is of paramount importance because it substantiates the need for early spinal ultrasound and enables detection of children requiring a full MRI scan. Management should be initiated as early as possible in order to limit the sequelae, stabilize and even improve the neurological status.

The aim of this study was to investigate whether these cutaneous anomalies had permitted an early diagnosis of OSD and if not for what reasons.

## Methods

### Population

We performed a retrospective, descriptive study on a non-consecutive series of pediatric cases followed up in the Department of Pediatric Surgery of the University Hospital of Reims, France, from 1995 to 2016. All patients were referred for urodynamics consultation, and presented an existing lumbosacral cutaneous anomaly from the neonatal period.

**Exclusion criteria were:** cutaneous anomalies corresponding to open spinal dysraphism; patients with cutaneous anomalies who had had antenatal diagnosis of OSD; simple sacrococcygeal dimples in the gluteal fold defined according to established criteria (soft tissue depression < 5 mm wide, appearing up to 2.5 cm above the anus) [5,6]; Mongolian spots and pilonidal cyst; isolated deviated gluteal fold and OSD occurring in the context of polymalformative urogenital and anorectal syndrome (bladder exstrophy, VACTERL syndrome, imperforate anus).

### Data recorded

From medical files we recorded age and sex, the type of cutaneous anomaly, whether or not spinal ultrasound was performed during the neonatal period, whether or not spinal MRI was performed, age at diagnosis of OSD, how patients were recruited, the type of malformation discovered, the presence or absence of clinical symptoms (neurological, neuro-orthopedic, bladder sphincter) before diagnosis and after surgery.

### Statistical analysis

Data are reported as mean±standard deviation for quantitative variables, and as number and percentage for qualitative variables. Comparisons between groups were performed using the Student t test. All analyses were performed using SAS version 9.4 (SAS Institute Inc., Cary, NC, USA).

### Ethical considerations

This study was approved by the Institutional Review Board of the University Hospital of Reims. The parents of all the included patients provided informed consent for the use of photographic.

## Results

Thirty-three patients were followed during the study period (14 boys, 19 girls), all born between 1994 and 2016. The duration of follow-up was variable, spanning a period of 21 years. Ten patients were identified from the periods between 01/1995 and 12/2004, and 23 patients were identified in the period between 2005 and 01/2016. Two patients were excluded because of missing data; thus 31 patients were included in the final analysis.

In terms of midline lumbosacral cutaneous markers, 13 patients had a palpable subcutaneous mass associated or not with other cutaneous signs characteristic of OSD (Figures 1A and B). Eighteen patients had no subcutaneous mass but presented isolated or multiple cutaneous lumbosacral markers. Among these 18 patients, we identified 7 groups: isolated dimple above the gluteal fold (2 patients, Figures 2A and 2B); dimple associated with deviation of the gluteal fold (6 patients, Figures 3A and 3B); dimple associated with another cutaneous anomaly (2 patients, Figure 4); vascular lesion (isolated or associated with other anomalies) (4 patients, Figure 5); area of cutaneous atrophy (2 patients, Figure 6); human tail (1 patient, Figure 7), and 1 patient with a deep, Y-shaped gluteal crease.

Recruitment occurred through various pathways; 17 patients were referred by their pediatrician and/or general practitioner; 7 by a pediatric urological surgeon, and 7 by a pediatric orthopedic surgeon. Among the patients with palpable subcutaneous mass, 11 were referred by the pediatrician of the maternity ward and 1 by an urologist. Among the patients with no mass, 6 patients were referred by their urologist, 6 by the orthopedic surgeon, and 6 by pediatricians



Figure 4: Dimple and small mass.



Figure 5: Vascular dysplasia.



Figure 6: Area of atrophy.



Figure 7: Human tail.

(from both neurology and maternity ward).

Ultrasound was performed in 12 patients' altogether (39%); i.e. in 7 of the 13 with a mass (53.8%), and in 5 of the 18 with no mass (27.7%). MRI was performed in all patients and confirmed a diagnosis of OSD in 29/31 patients (93.5%). Two patients had no OSD. The time when MRI was performed was considered as the date of diagnosis for the purposes of this study. The average age at diagnosis was 27.6 months  $\pm$ 44.3 for all of them; the diagnosis was done earlier in the group with subcutaneous mass (3.6 months  $\pm$ 2.1) than in the group with no mass (44 months  $\pm$ 52.1) the difference was significant ( $p < 0.004$ ). The observed malformations were summarized in the (Table 1).

Among the two patients without spine lesions, the both had bladder dysfunction, one related to a posterior urethral valve and the other due to idiopathic overactive bladder. One dermal sinus had no spinal prolongation, with the distal extremity of the dermal sinus terminating at the coccyx. Among the 29 confirmed patients with OSD 16 were symptomatic prior to management. Fourteen patients had neuro-orthopedic and/or bladder-sphincter sequelae.

## Discussion

Our study points out the existence of a strong relation between the presence of congenital midline lumbosacral markers and the existence of OSD, which is in line with previous reports in the literature [3,6-7]. Sarikaya Solak et al. [8] reported that the prevalence

of OSD was 4.8% and that cutaneous lesions were various and often interlinked but could also be isolated [7,8]. Our series also shows a twofold increase in the number of cases reported to our centre in the last 10 years.

The different types of lumbosacral cutaneous lesions observed in our series are concordant with those described in the literature [6,7]. The lesions were on or near the midline, in the lumbosacral region, and rarely dorsal or cervical. We identified two main groups of patients, namely those with a subcutaneous mass corresponding to lipomatous tissue or nerve tissue, particularly characteristic of OSD; and a second group including patients with lesions but no mass (hypertrophy or scarring, human tail or caudal appendage, cutaneous aplasia or dermal sinus or dimple, dyschromia, vascular, hairy or deviation of the gluteal fold associated with a dimple).

The lesions appeared in various combinations and the presence of several lesions indicates a higher likelihood of underlying OSD. These markers are present in more than 90% of OSD cases with rates ranging from 50 to 100% according to different studies [1,7].

We excluded from our series isolated deviation of the gluteal fold as this is more difficult to categorize among the markers of OSD when it occurs strictly in isolation [5,9]. However, when associated with a dimple above the gluteal crease, this defect was more informative in our series [10]. Regarding anomalies of the filum and dermal sinus, none of the 9 affected patients had a subcutaneous mass. For the myelolipomas which was the most frequent lesion in our series and in the literature [7,11], subcutaneous lipomas did not always exist, which is also in line with previous reports [7].

In terms of diagnosis of OSD, there was a clear difference in the confirmation time of diagnosis by MRI between the group with a subcutaneous mass and the group without, i.e. 3 months on average for patients presenting a mass, versus an average of 44 months (ranging widely from 4 months to 7.5 years) among those without a mass. There is paucity of data regarding the differences in time intervals to diagnosis between those with a mass and those without a mass.

OSD is a congenital loco-regional progressive pathology due to myelodysplasia, secondary traction of the conus and spinal compression. It is widely acknowledged that it exists a significant risk of bladder-sphincter dysfunction as well as renal, anorectal and neuro-orthopedic repercussions whose functional and social consequences may be major [1,12,13]. The search for cutaneous signs is thus particularly important when there is no clinical evidence at birth. Indeed neuro-orthopedic signs are commonly absent, either in terms of spinal column balance or the lower limbs (e.g. scoliosis, high arched foot etc.). Bladder-sphincter dysfunction and anorectal disorders can be difficult to detect in the first months of life and are generally absent with the exception of acute affections such as urine retention or pyelonephritis.

The indications for neurosurgery remain debatable since OSD represents an heterogeneous group of malformations and progression varies widely. For symptomatic patients, the indication for surgery is more consensual whereas it is more controversial in patients who are asymptomatic. Some authors purport that it is not appropriate to wait for symptoms to appear before operating since the neurological impairment is rarely reversible. Furthermore, the surgery itself may be complicated by post-operative neurological sequelae. Mankahla et al. [2] recommend that the operation be performed under intra-

**Table 1:** Types of malformations description.

Type of malformations	Symptoms before diagnosis	n =	Subcutaneous mass		Age in months at MRI	Neurosurgery		Post-management
			Yes	No	++	Yes	No	
Isolated tethered cord (n=3)	OAB, UR	1		1	83	1		OAB, dysuria
	no symptom	2		2	4, 4	2		no symptom
Lipoma terminal filum (n=2)	UR, Scoliosis	1		1	60	1		scoliosis
	no symptom	1		1	21	1		no symptom
Myelolipoma (n=11)	OAB, dysuria	4	2	2	4, 3, 3, 3	4		OAB, dysuria, NOD
	no symptom	7	4	3	3, 8, 12, 8, 7, 3, 3	7		no symptom
Dermal sinus (n=2)	Meningitis	1		1	22	1		Neurologic sequela
	no symptom	1		1	94		1	no symptom
Diastematomyelia (n=2)	Scoliosis	1		1	3	1		Scoliosis
	no symptom	1	1		3		1	no symptom
Syringomyelia (n=2)	OAB, dysuria, Scoliosis	1		1	77		1	OAB, Scoliosis
	OAB	1		1	50		1	no symptom
Meningocele (n=1)	no symptom	1	1		3	1		no symptom
Complex malformations (n=6)								
Myelocystocele	OAB, dysuria	1	1		3	1		OAB, dysuria, NOD
Angiomyelocystocele	UR	1	1		0	1		OAB, dysuria
Lipomyelocystocele	UR	2	2		3, 3	2		OAB, dysuria
Lipomyelomeningocele	OAB	1	1		3	1		OAB, UR
Tethered cord + syringomyelia	HAB	1		1	131	1		no symptom
No OSD (n=2)	OAB, Scoliosis	1		1	185		1	scoliosis
	OAB	1		1	45		1	no symptom

Used abbreviations: OSD = occult spinal dysraphism - OAB = overactive bladder – UR = urinary retention - HAB = hypoactive bladder - NOD = neuro-orthopedic deficit  
 ++ : Ages at MRI are referenced on the case number

operative neurophysiological control and to operate between the ages of 6 to 9 months unless there is progressive deterioration before that time.

However, for dermal sinus early diagnosis makes it possible to prevent meningitis. One child in our series presented bacterial meningitis that revealed a dermal sinus. Lallemand et al. [14] reported a case of dermal sinus complicated by medullar abscess with major neurological sequelae in a 12-month old patient underlining the importance of early diagnosis and regular surveillance. It exists a general consensus that there is an indication for surgery in all patients with dermal sinus to resect the tract fully and correct the malformation as soon as the diagnosis is confirmed [15,16].

Short filum syndrome and filum lipoma, is a widely acknowledged indication for surgery if the tethering is low-lying, although there is no systematic approach [17]. In our series, prophylactic surgery was performed in 3 patients and curative surgery in 2 other patients who had persistent post-operative sequelae.

For lipomas of the conus and other complex malformations, there is currently no consensus regarding the indications for neurosurgery which range from preventive purposes [18] to surgery when deterioration occurs [19]. Depending on where the lipoma is located the nerves roots can emerge outside the lipoma or may transgress the lipoma rendering surgery more difficult and at higher risk [19].

White et al. [20] reported that diurnal incontinence and

overactive bladder are the most frequently encountered symptoms and usually improved after surgery. Furthermore, post-operative recovery is inversely proportional to the duration of compression of the nerve fibers. Surgery before the appearance of symptoms is therefore indicated. Isolated lesions and pre-operative continence are not predictors of post-operative continence as regards filum lipomas and tethered cord. Urodynamic studies pre- and post-surgery do not predict continence, but are necessary to monitor for re-tethering [21]. Numerous authors have reported late neurological deterioration in pediatric and adult patients with undetected OSD [22,23].

Nonetheless, pluridisciplinary monitoring is important for prognosis and also contributes to therapeutic decision-making [12,20], particularly regarding urodynamic studies [12,13,24]. Averbeck et al. [25] reported that the main goal of neuro-urological management is the best possible conservation of upper and lower urinary tract function. Regular urodynamic studies allow detection of risk factors before irreversible damage occurs. Early treatment of high pressure in the bladder makes it possible to avoid repercussions on the upper urinary tract (e.g. uretero-hydronephrosis, reflux, alteration of renal function), preserve bladder compliance and improve continence [25]. More than 50% of children with early management maintain satisfactory continence [12,18].

The interest of early detection of cutaneous markers suggestive of OSD lies in the possibility of establishing an early diagnosis, if possible even before progressive neurological symptoms, and allowing



appropriate monitoring and management. Prenatal diagnosis of OSD remains rare [12]. The presence of cutaneous markers calls for spinal ultrasound, ideally performed during the first 2 months of life [17]. It is useful for the analysis of the spinal cord, the position of the conus, dural cul de sac and the filum terminale.

The conus medullaris is situated above L3 at birth and is contiguous with the filum terminale which should be less than 2 mm thick. The conus is surrounded by cerebrospinal fluid and nerve roots [26,27]. The position of the conus medullaris is informative since in the majority of OSD the conus are low-lying, except in syringomyelia or diastematomyelia [28]. This can allow the diagnosis of isolated tethered cord or more complex dysraphisms [28]. In this last case, it is recommended to perform transfontanelar ultrasound as well as ultrasound of the urinary tract [29]. Ultrasound is a non-irradiating, painless, sensitive and specific exam for the detection of OSD before the age of 2 months [5,17,28]. Beyond this age, the ossification of the posterior arch of the spine renders interpretation of the spinal ultrasound more difficult. According to the literature, normal spinal ultrasound performed by an experienced operator can rule out a serious dysraphism and obviate the need for MRI [5,29], except in the case of dermal sinus, although there is no confirmed correlation between MRI and operative findings [16].

Ultrasound allows detection of patients requiring complementary assessment of lesions by spinal MRI [17,24,30,31] and possibly also cerebral MRI. At such a young age, the immobility required for successful MRI imaging is usually achieved by simple contention. When the diagnosis is being considered at a later stage, MRI is the only option. Depending on the centre and the patient's age, the immobility required for the MRI is achieved either by sedation or by general anaesthetic [32].

According to Henriques et al. [33] even when skin lesions are minor in the neonate, spinal ultrasound is justified by the fact that it is a simple, feasible, inexpensive and reliable examination in terms of the benefit to be yielded from early treatment.

## Conclusion

OSD is rarely diagnosed prenatally. Examination of the lumbosacral region for skin markers should be systematically included in the battery of neonatal tests in the maternity ward or during the first month of life, particularly since cutaneous markers may be discreet when there is no associated subcutaneous mass. Any such findings should be systematically noted in the baby's medical records. Targeted patients can undergo spinal ultrasound to detect OSD and although the time window during which spinal ultrasound can be used is limited, it remains a simpler exam than MRI performed at a later stage under general anesthetic.

Given that OSD is sometimes overlooked, this investigation should be included in the work-up of any orthopedic consultation for disorders of spinal equilibrium or lower limb disorders. It should also be performed systematically at the first consultation for bladder-sphincter and anorectal disorders. MRI is always indicated to evaluate the lesions. There is no consensus regarding the optimal neurosurgical therapeutic approach. However, pluridisciplinary management should be initiated as early as possible with neuro-urological and neuro-orthopedic monitoring, in order to guide decision-making and propose appropriate early management with a view to preserving urinary, anal, renal, motor and spinal function, and consequently, quality of life.

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## Author Contributions

NM contributed in preparation of manuscript, MLPM and SB proposed patients to inclusion, DJ contributed to statistical analysis and in preparation of manuscript, CJ initiated the project, made review of all medical records, and prepared the manuscript.

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