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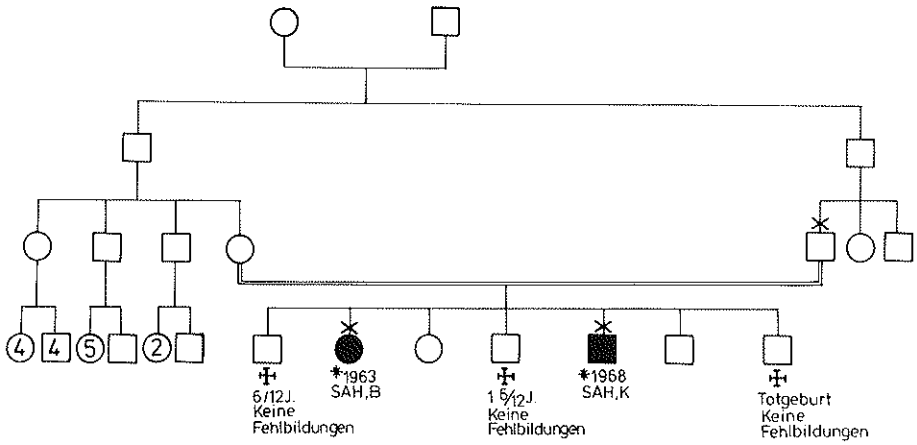

Abb. 23 Sippentafel von Fall 3 und 4.

3. Lange, schmale Grundphalangen der Finger 2 bis 6 (ulnare oder intermediäre Hexadaktylie)
4. kurze, plumpe Metatarsi.

Der Fall 3 und 4 sind Schwester und Bruder aus einer Vettern-Kusinenehe 1. Grades (Abb. 23). Dies ist ein starkes Indiz für autosomal rezessive Vererbung, zu der auch die anderen isolierten Fälle passen. Die Familie aus Taiwan, in der rechtsseitige Syndaktylie der Finger 3 und 4 mit dominanter Vererbung vorkommt, wie sich auch sonst für diesen Syndaktylietyp typisch ist, läßt an die Möglichkeit denken, daß es sich hier um eine Manifestation des Gens bei Heterozygoten handelt. Wenn auch die wenigen Fälle kein endgültiges Urteil über das Wiederholungsrisiko zulassen, so sollte man bei der genetischen Beratung doch darauf hinweisen, daß ein Wiederholungsrisiko von 25% für Geschwister so wahrscheinlich ist, daß man es einer Entscheidung für oder gegen weitere Kinder zugrundelegen sollte.

Nachtrag

Die Eltern von Fall 2 sind nicht miteinander verwandt. Bei der Geburt des Patienten war die Mutter 28, der Vater 30 Jahre alt. Der Patient hatte 2 gesunde Schwestern und einen gesunden Bruder. Gleichartige Fälle waren in der Familie nicht bekannt.

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Spondylo-Costal Dysostosis in two Siblings

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Summary

Two new cases of Spondylo-Costal Dysostosis (SCD) are reported in two siblings with strikingly similar skeletal abnormalities. Parental consanguinity documents in this family an autosomal recessive inheritance of trait. Clinical variability of SCD is discussed on the basis of clinical and radiological features. Its genetic heterogeneity is pointed out even considering the occurrence of cases with autosomal dominant as well as recessive inheritance.

Spondylokostale Dysostosis bei zwei Geschwistern

Es werden zwei neue Fälle spondylokostaler Dysostosis (SKD) bei zwei Geschwistern mit auffallend ähnlichen Anomalitäten des Skeletts beschrieben. Die Blutsverwandtschaft der Eltern in dieser Familie dokumentiert einen autosomal-rezessiven Erbgang des Merkmals. Die klinische Variabilität der spondylokostalen Dysostosis wird diskutiert auf der Grundlage klinischer und radiologischer Befunde. Unter Berücksichtigung des Auftretens von Fällen mit autosomal-dominanten wie auch rezessivem Erbgang wird die genetische Heterogenie von SKD hervorgehoben.

Introduction

Spondylocostal dysostosis (SCD) is a rare and distinct syndrome with mendelian inheritance and characteristic skeletal abnormalities. Dysproportionate short stature, shortness of neck and chest, kyphoscoliosis and bulging abdomen are main clinical features of SCD. Skeletal abnormalities of SCD includes costovertebral agenesis, hemivertebrae, vertebral fusions and ribs malformations. Since its first description by *Jarcho* and *Levin* (1938) (16), SCD have been reported under several different names (Tab. 1). The term „spondylo-costal dysostosis" is recommended in the International Revision of Nomenclature of Constitutional Diseases of Bone (28).

Two clinical variants of SCD exist: a severe one with multiple skeletal abnormalities, cardiac involvement, respiratory tract complications and reduced survival and a mild one with less severe bone abnormalities and prolonged survival. An autosomal recessive inheritance is more frequent in cases with the severe variant; in mild SCD both autosomal recessive and dominant modes of transmission have been described.

Most of SCD cases have been diagnosed after birth, although some clinical reports document the possibility of ultrasonographic prenatal diagnosis (31).

We report on two new familial cases with autosomal recessive SCD, that may provide additional informations on phenotypical variability and genetic heterogeneity of the syndrome.

Clinical Reports

CASE 1 - P. L., a 9 years and 8 months old boy, is the second child of healthy and consanguineous parents (first cousins). The pregnancy was uncomplicated until term; delivery was spontaneous at 40 weeks (the birthweight was 3200 g and the birthlength was 52 cm). A normal child was born from the first pregnancy; a girl with similar clinical and radiological findings was born from the third pregnancy (our case 2). Perinatal events were normal. His psychomotor development during the first year of life was regular; thereafter a slight developmental delay and poor school performances have been noted.

Clinical data: weight was 20 kg (<3rd centile), stature was 119.8 cm (<3rd centile), weight/height was ratio 0.88 (−2 SD), head circumference was 47.2 cm (−3.18 SD). He had dysproportionate short stature with the lower limbs longer than trunk (trunk/limbs ratio is 0.91, normal value for age 1.02), unusual face, bilateral

Tab. 1 "History" of SCD

	References
1) Hereditary Malformations of the vertebral bodies	16
2) Hereditary multiple hemivertebrae	35
3) Syndrome of bizarre vertebral bodies	18
4) Familial polydysspondylia	22
5) Spondylo-costal dysplasia	29, 21
6) Spondylo-thoracic dysplasia	20, 25, 13, 34, 7, 15
7) Costo-vertebral dysplasia	5, 2, 36, 9
8) Occipito-facial-cervico-thoracic-abdomino-digital dysplasia	23
9) Spondylo-costal dysostosis	12, 3, 10, 17, 32, 28, 6, 24, 37, 14, 30, 11, 19
10) Spondylo-thoracic dysostosis	33
11) Jarcho-Levin syndrome	4, 26, 31, 8

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strabismus, low nucal hairline, short and webbed neck, short trunk, deformed and asymmetrical thorax with Sprengel deformity, severe scoliosis and hypotrophic thorax muscles (Fig. 1a, b, c). Heart and respiratory tract examinations were normal; the abdomen was moderately bulged. Liver, spleen and external genitalia were normal. His tendineous reflexes were hyperelicitable with clonus of feet. QI was 75 (*Terman-Merrill*), confirming a slight developmental impairment.

Chest and spine X-rays showed a generalized severe anomaly of the entire vertebral column. It appeared markedly shortened and malaligned. The vertebral abnormalities were more readily apparent in the thoracic and lumber regions, but also present in the cervical tract. They included a complete C2–C3 vertebral fusion with C4 retrospondylolisthesis (Fig. 2), the absence of two dorsal vertebrae and correspondent ribs, multiple hemivertebrae and partial fusion of the T12-L1 vertebrae (Fig. 3). The

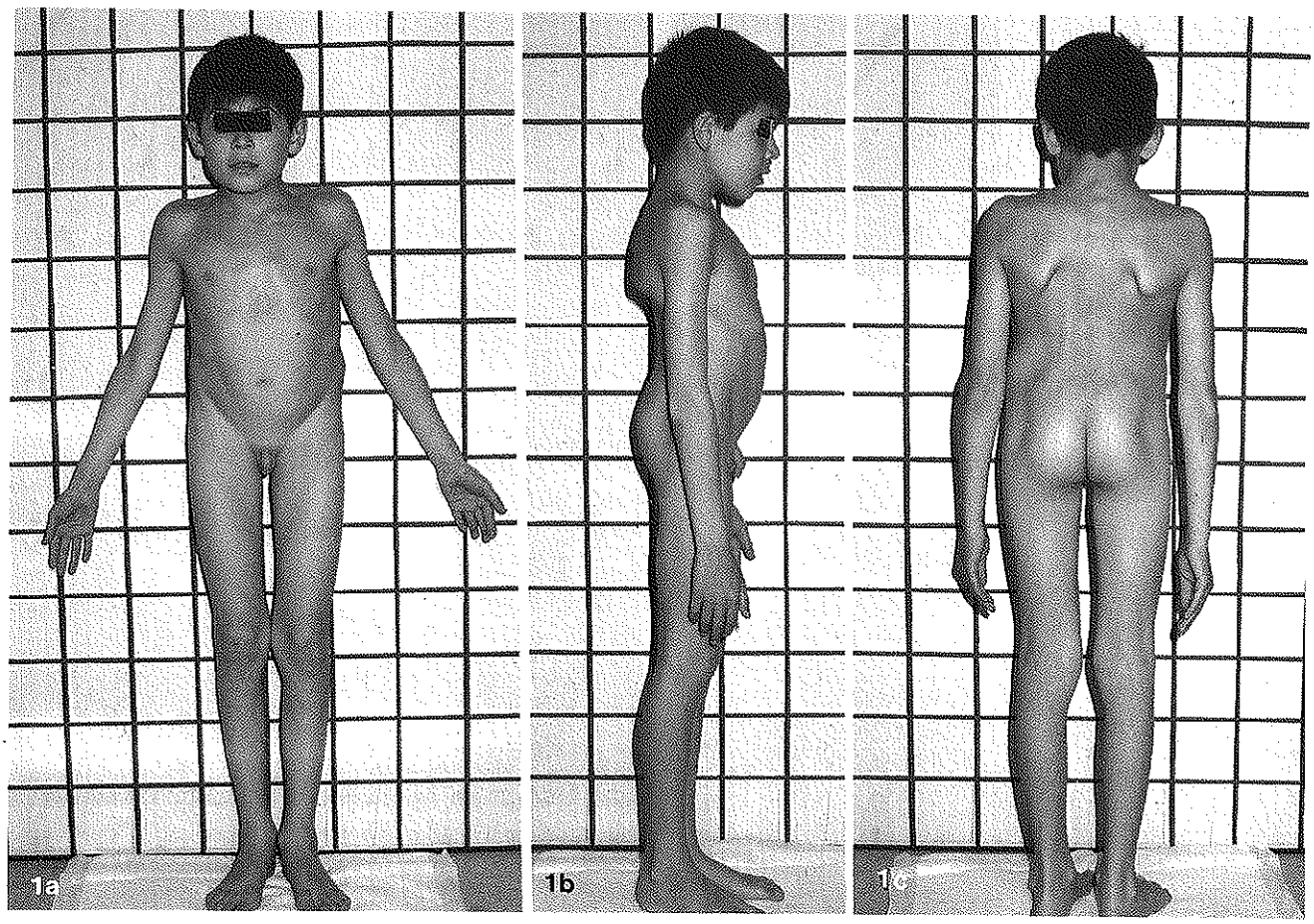


Fig. 1 Case 1 at age 9 years and 8 months. a) Frontal, b) lateral and c) back views.

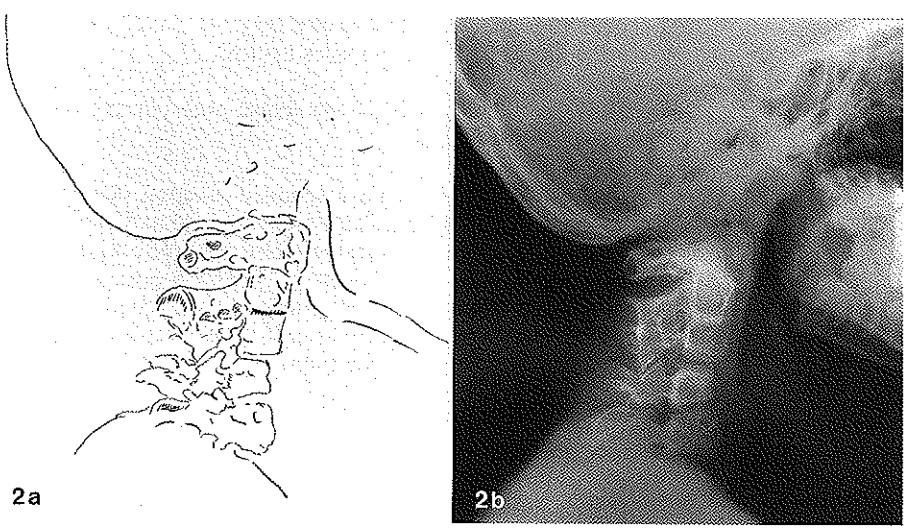


Fig. 2 Case 1: lateral view of cervical spine. a) Schematic and b) radiographic demonstrations. Note complete C2–C3 vertebral fusion with C4 retrospondylolisthesis and deformities of C1 and C5 vertebral bodies.

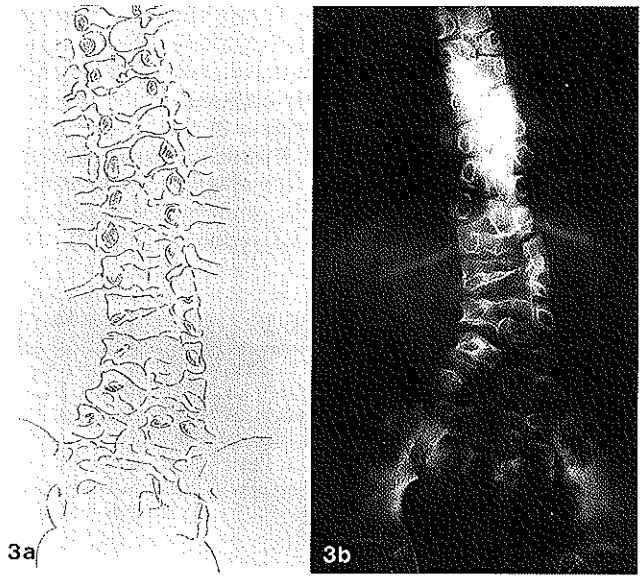
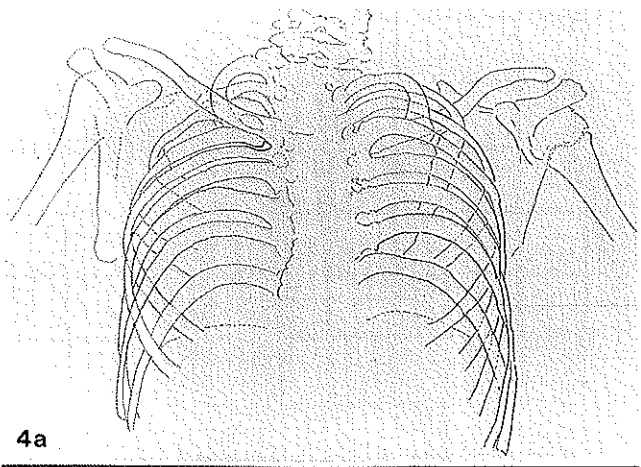


Fig. 3 Case 1: thoraco-lumbar regions of spine. a) Schematic and b) radiographic demonstrations. Note multiple hemivertebrae, scoliosis and T12-L1 vertebral fusion.



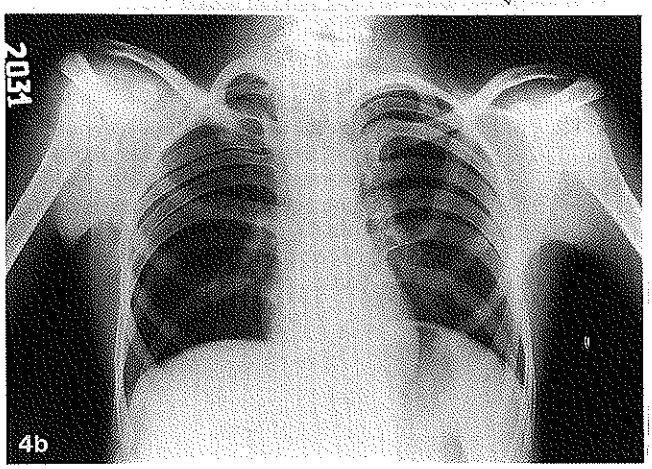


Fig. 4 Case 1: Chest X-rays. a) Schematic and b) radiographic demonstrations. Note multiple ribs abnormalities with synosthosis near the costovertebral joints.

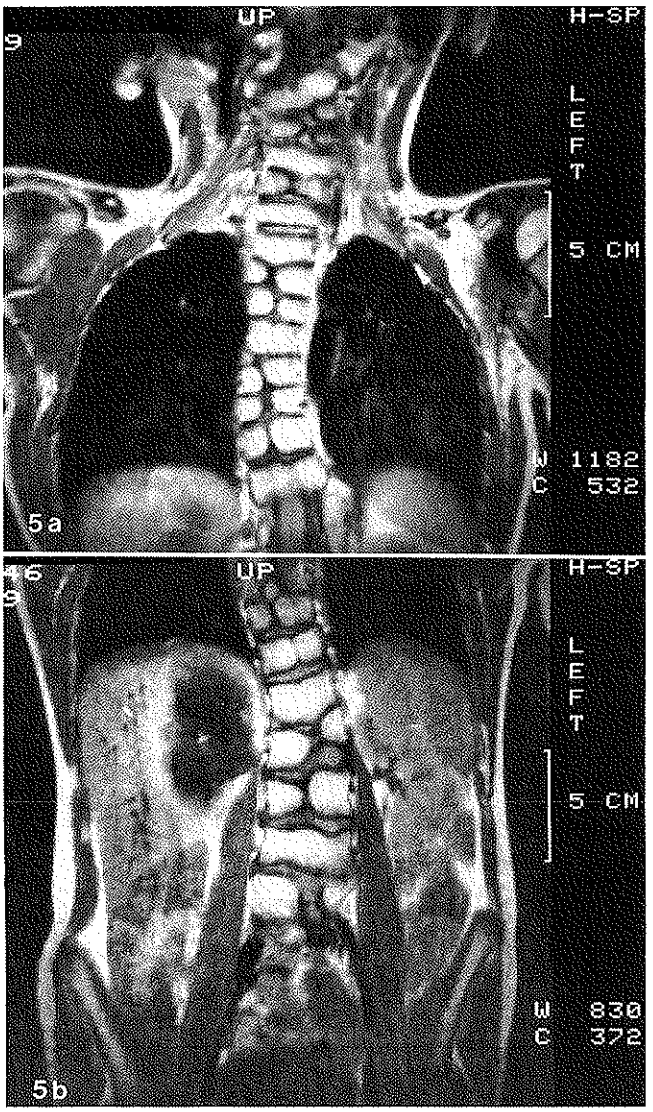


Fig. 5 Case 1: RMI of the spine. a) Upper and b) lower views, showing multiple hemivertebrae and several irregularities of the intervertebral discs.

ribs were irregular in shape and course, with partial fusion near the costovertebral joints (Fig. 4). Skull, limbs and bone age (TW2 Tanner) were normal.

X-rays of the spines both parents were interpreted as normal.

Magnetic resonance imaging (MRI) scans of the spine confirmed multiple vertebral and ribs abnormalities, showing the hemivertebrae and the abnormalities of the intervertebral discs (Fig. 5) with a greater number of details. The dural sac was normal until L3–L4 vertebral level, from where it was poorly developed and partially replaced by some adipose tissue.

Blood routine analysis were normal. Karyotype from peripheral blood was 46,XY. An abdominal ultrasound scan was normal. His electroencephalogram (EEG) showed an alpha-type rythm, with interposition of theta waves, particularly on the left emisphere. Left trape-

zium and right deltoid electromyography (EMG), auditory brain responses (ABR) and fundus oculi were normal. Visual evoked potentials (VEP) showed bilaterally low and slight delayed waves, more consistent on the right side (Fig. 6).

CASE 2 – P. G., an 8 years and 4 months old girl, is a sister of our case 1. The pregnancy was uncomplicated until term; no ultrasound scan was performed during gestation. Delivery was spontaneous (the birthweight was 2750 g, the birthlength was 48 cm). Her psychomotor development during the first year of life was normal; thereafter a developmental delay and poor school performances have been noted.

Clinical data: weight was 20 kg (<3rd centile), length was 115.2 cm (<3rd centile), weight/height was ratio 0.98 (50th centile), head circumference was 49

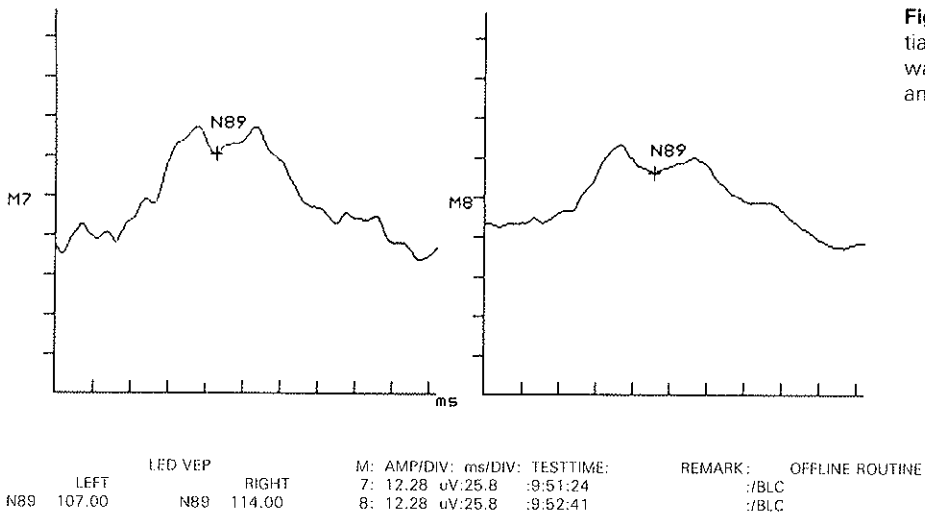


Fig. 6 Case 1: Visual Evoked Potentials. Note the low amplitude of the waves, particularly on the right side and their slight delay.

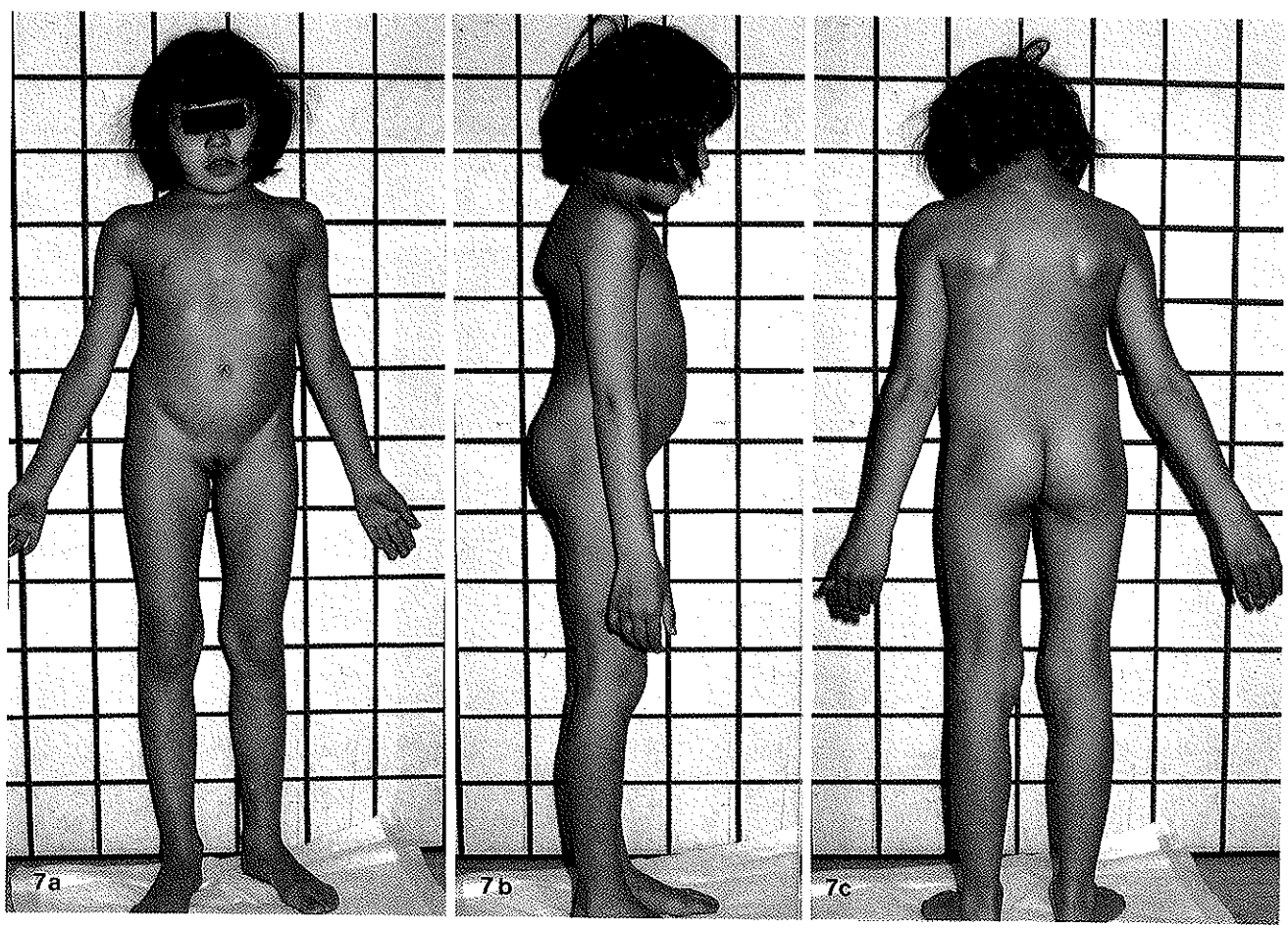


Fig. 7 Case 2 at age 8 years and 4 months. a) Frontal, b) lateral and c) back views.

cm (−2.18 SD). She had dysproportionate short stature with the lower limbs longer than trunk (ratio trunk/limbs was 0.96, with normal value for age 1.23), unusual face, bilateral strabismus, highly arched palate, low nucal hairline, short and webbed neck, hypotrophic sternocleidomastoid and trapezium muscles, short trunk, deformed and asymmetrical thorax with severe scoliosis and Sprengel deformity (Fig. 7a, b, c). Her tendineous reflexes were hyperelicitable with bilateral clonus of feet. QI was 65 (*Terman-Merrill*), confirming a developmental impairment.

Chest and spine X-rays showed a generalized severe anomaly of the entire vertebral column, which appeared markedly shortened and severely malaligned. The vertebral abnormalities were readily apparent in the thoracic, lumbar and cervical regions. They included a complete C3–C4 vertebral fusion with C2 and C3–C4 complex retrospondylolisthesis (Fig. 8), the absence of the two dorsal vertebrae and correspondent ribs, and a large number of hemivertebrae (Fig. 9). The ribs were irregular in shape and course, with partial fusion near their costovertebral joints (Fig. 10).

RMI scans of the spine confirmed the severe vertebral abnormalities, showing the hemivertebrae and the abnormalities of the intervertebral discs with a greater number of details (Fig. 11).

Blood routine analysis were normal. Karyotype from peripheral blood was 46,XX. Her EEG was abnormal with slow rhythm and interposition of fast

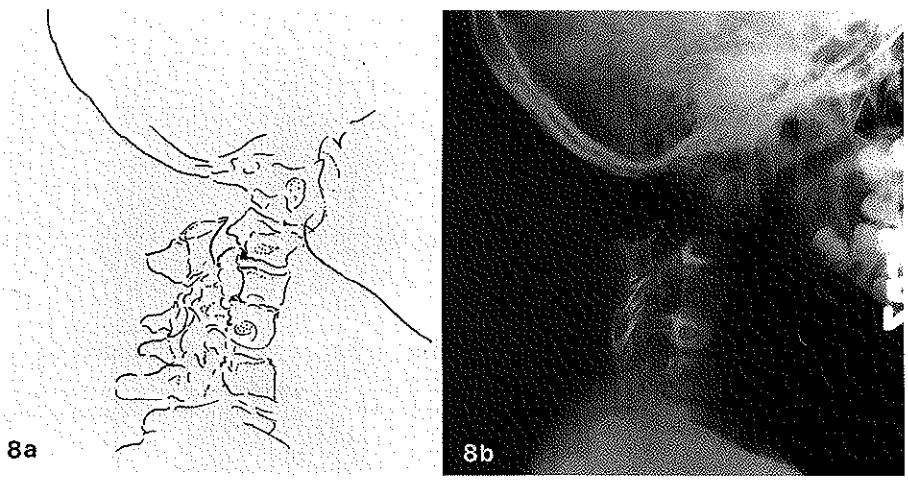


Fig. 8 Case 2: lateral view of the cervical spine. a) Schematic and b) radiographic demonstrations. Note massive fusion and retrospondylolisthesis of the C3–C4 vertebrae and the severe irregularity of the other vertebral bodies.

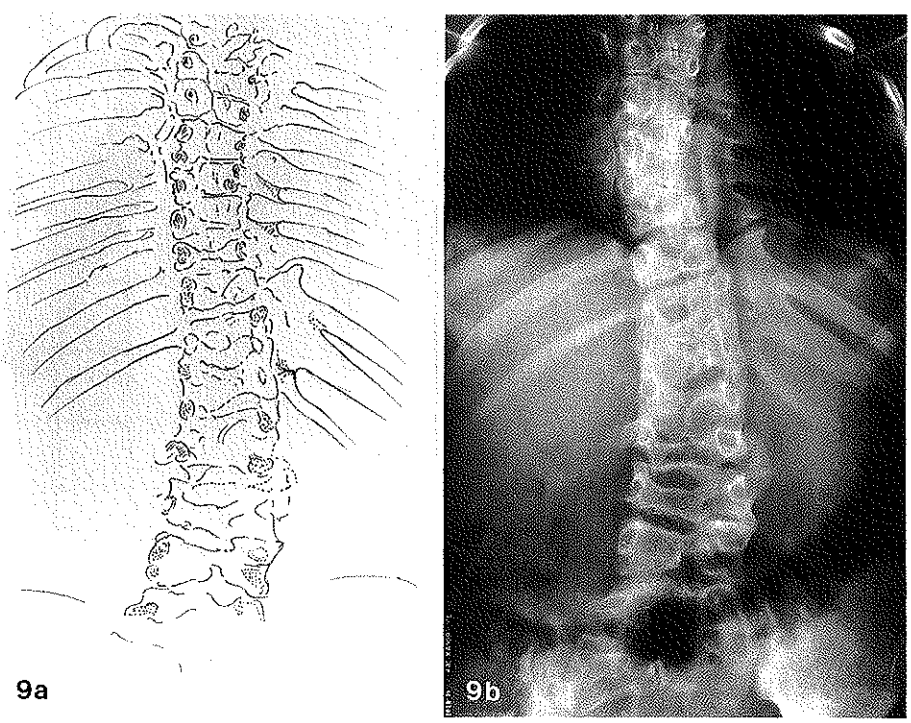
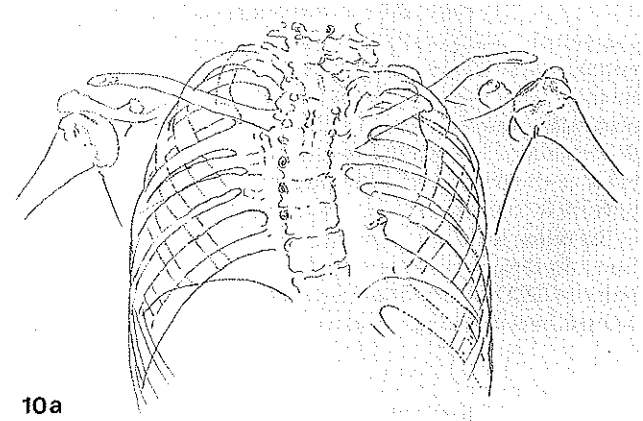


Fig. 9 Case 2: thoraco-lumbar tract of the spine. a) Schematic and b) radiographic demonstrations. Note multiple hemivertebrae, several irregularity of the vertebral bodies, spinal malalignment and ribs fusion near the costovertebral joints.



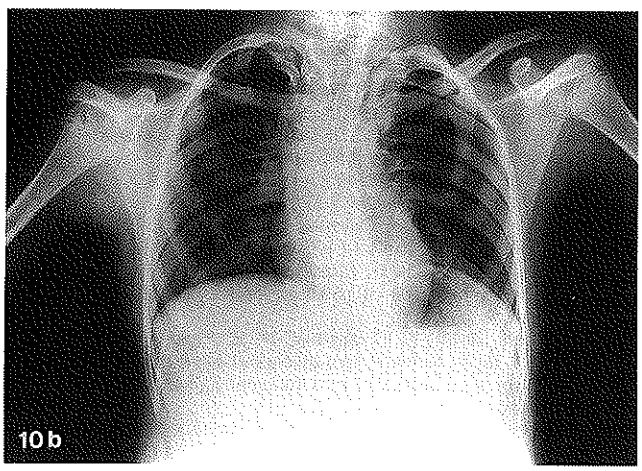
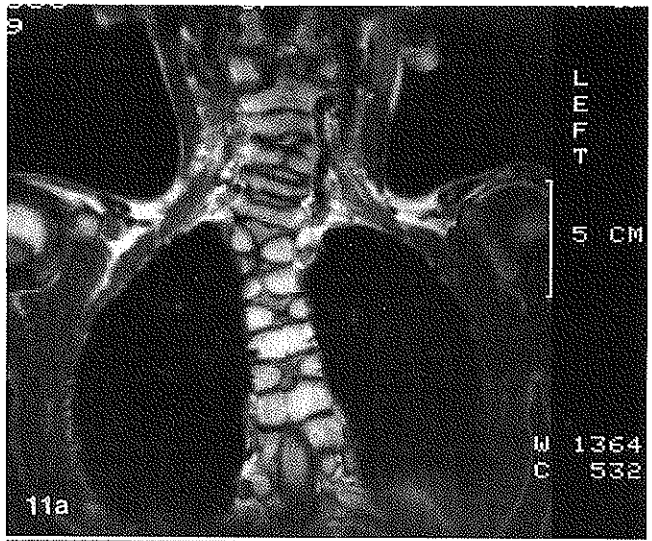


Fig. 10 Case 2: chest X-rays. a) Schematic and b) radiographic demonstrations. Note the severe ribs irregularities with partial synostosis near the costovertebral joints.



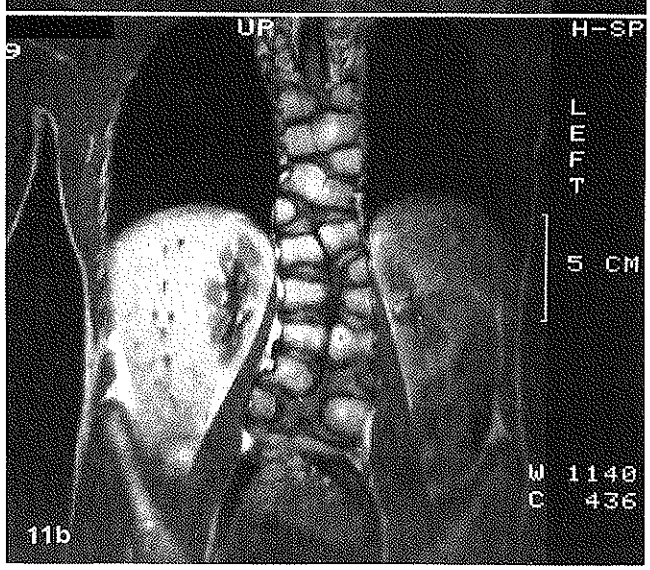


Fig. 11 Case 2: RMI of the spine. a) Upper and b) lower views. Note multiple hemivertebrae, spinal malalignment and abnormalities of the intervertebral discs.

waves, particularly on the right emisphere. Right trapezium EMG, fundus oculi, PEV were normal. ABR demonstrated a mildly elevated threshold.

An abdominal ultrasound scan showed a pyelo-uretero-hydronephrosis on the right side. An urographic evaluation identified a retrocaval ureter at L2 level. From this point in fact the appeared thinner and medially deviated (Fig. 12). She underwent surgical correction, with decussation of the ureter from cava vein and subsequent ureteral-ureteral anastomosis. Clinical and radiological follow-up of the little patient demonstrated a progressive reduction of the hydronephrosis.

Discussion

All the skeletal abnormalities in SCE are the result of multiple segmentation defects of the axial skeleton, whose morphogenesis complete before the 20th day of gestation (24). Clinical variability of DSC is broad; chest deformities and costovertebral abnormalities vary from asymptomatic to severe life-threatening forms. Agenesis of vertebrae and ribs, hemivertebrae, vertebral and ribs fusion can be variably associated. The most severe cases are characterized by a typical chest with „fan-like" or „crab-like" appearance on X-rays. In these cases

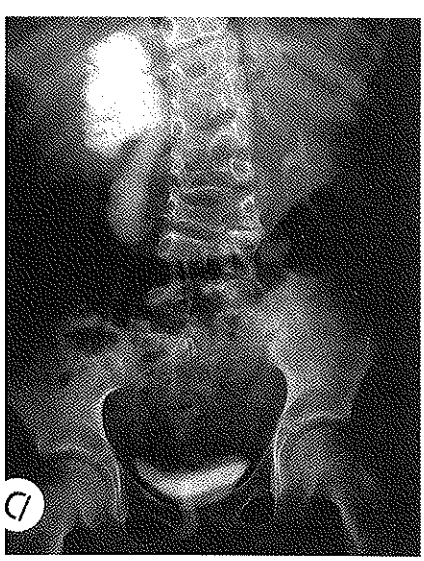


Fig. 12 Case 2: excretory urography. Note the severe dilatation of the renal calyces and ureter on the right side. From L2 vertebral level the ureter appears thinner and medially deviated, due to a retrocaval malposition. The left kidney and ureter appear normal.

Tab. 2 Costovertebral malformations in present SCD cases

Costovertebral abnormalities	Case 1	Case 2
Vertebral and rib agenesis	+	+
Hemivertebrae	+	+
Vertebral fusions	+	+
Vertebral body deformations	+	+
Spondylolisthesis	+	+
Spinal malalignment	+	+
Ribs synosthosis	+	+
Other ribs anomalies	+	+
Sprengel deformity	+	+

Tab. 3 Associated malformations in SCD

Prominent occiput
Lower limbs abnormalities
Spina bifida occulta
Camptodattily
Abdominal wall defects
Mycrocephaly
Triangular mouth
Sindattily
Cleft of palate
Ano-rectal abnormalities
Genito-Urinary tract abnormalities
Heart defects
Single umbilical artery

the features of the syndrome are quite remarkably and permit its recognition at birth.

Differential diagnosis of SCD includes all the syndromes in which the segmentation defects of vertebrae are likely to be found. Into this etiologically heterogeneous group of diseases, COVESDEM syndrome, dyssegmental dysplasia, Klippel-Feil anomaly, MURCS and VACTERL associations, Goldenhar syndrome can be easily excluded on clinical grounds.

All the skeletal abnormalities found in our cases are listed in table 2. Radiologically, they are very similar to those present in the severe SCD variant. Nevertheless, they can be ascribed to the mild variant of SCD, even considering their normal survival and the lack of heart failure and respiratory tract complications.

Several malformations can be found in association with SCD (table 3); we found microcephaly in both our cases and urinary tract abnormalities in case 2. Although some reports exist in genito-urinary abnormalities in SCD cases (6, 10, 25), this is the first report of a retrocaval ureter. Because of the close embryologic relationship between the nephronic and mesodermal somitic structures, urinary tract anomalies can be expected in at least some of the subjects with SCD.

The mild developmental delay observed in our two cases, can be related to the mycrocephaly, which is unfrequently reported in SCD.

Parental consanguinity and the absence of clinical and radiological features in both parents prove the autosomal recessive inheritance of SCD in our family. Although mild cases of SCD usually show an autosomal dominant transmission, we think that it is impossible to differentiate between the two genetic types of SCD on the basis of the clinical and radiological findings. A recent report on a case with dominant inheritance and severe chest deformities supports previous considerations (19). Therefore, doubtlessy genetic heterogeneity and clinical variability of SCD do not allow satisfactory subdivisions of the disease into different variants.

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Buchbesprechungen

Helmut Lechner (Herausgeber): **Epilepsien und ihre Grenzgebiete.** 1990, 120 Seiten, 23 Abbildungen, 20 Tabellen. Polyleinenumschlag, DM 32,—. Werk-Verlag Dr. Edmund Banaschewski, München-Gräfelfing. ISBN 3-8040-0384-2

Wie der Herausgeber im Vorwort anführt, haben verschiedene Disziplinen zu Fortschritten auf dem Gebiet der Epileptologie beigetragen. Dies dem praktisch tätigen Arzt zu vermitteln, ist Anliegen der elf Beiträge, die von Spezialisten österreichischer Kliniken verfaßt wurden. Knapp und instruktiv werden die neurophysiologischen Grundlagen epileptischer Anfälle (Petsche) sowie pathologisch anatomische Befunde bei Epilepsien dargestellt (Kleinert), wobei auch auf die wichtige Bedeutung von Mikrodyskinesien verwiesen ist. Welchen Stellenwert moderne neurophysiologische und bildgebende Verfahren haben (Lechner u. Pump, Fazekas et al.) ist an Beispielen deutlich gezeigt. Genetische Aspekte der Epilepsien finden Berücksichtigung (Rosenkranz), die Differentialdiagnose nerschiedener Anfälle bei Erwachsenen (Mamoli) und bei Kindern (Groh) wird praxisnah diskutiert. Im Kapitel über Epilepsien des Kindesalters (Graf) und über das klinische Bild bei Erwachsenen (Deecke) findet man viele wertvolle Informationen zu Systematik, Diagnose, Prognose und Therapie. Ein eigener Abschnitt ist dem Wert der Serumspiegelbestimmung von Antiepileptika gewidmet (Bohr u. Bauer), ebenso den Problemen der Begutachtung bei epileptischen Krankheiten. – Es ist gelungen, in einer straffen, gut gegliederten Darstellung alle praktisch wichtigen Aspekte zu berühren und fundiert über den derzeitigen Kenntnisstand zu berichten. Für den in der Praxis mit Anfallskrankheiten konfrontierten Arzt dürfte die relativ preiswerte Monographie ein wertvoller Helfer sein.

G. Neuhäuser, Gießen

Harnsteinleiden. Ursachen – Diagnose – Therapie. Hrsg.: *Vahlensiek, W.* 1987, 198 Abb., 17 Farbtafeln, XIII, 610 Seiten. 910 g. Gebunden DM 148,—. Berlin–Heidelberg–New York–London–Paris–Tokyo: Springer-Verlag. ISBN 3-540-16295-X

In diesem Buch wird das bisherige Wissen über das Harnsteinleiden umfassend und übersichtlich dargestellt. Auch neuere Erkenntnisse über Kausalfaktoren und neueste Therapieverfahren wie die extrakorporale Stoßwellenlithotrypsie werden vorgestellt. Wo es sachlich gut begründete Meinungsunterschiede gibt, werden diese dem Leser nicht vorenthalten. Der Pädiater findet für die besonderen Probleme seiner Klientel nur wenig Information.

Im Vergleich zu den während der letzten Jahre im gleichen Verlag herausgekommenen Handbuchbänden zum Thema „Urolithiasis" hat die Monographie für Leser in unserem Land den Vorteil der Deutschsprachigkeit und der kompakten Information in einem handlicheren Band.

Das Buch gliedert sich in Kapitel über die Epidemiologie, die Kausal- und Formalgenese, die Diagnostik und Therapie des Harnsteinleidens.

Insgesamt kann ich das Buch Kindernephrologen und Kinderurologen sowie in Kinderkliniken tätigen Pädiatern mit besonderem Interesse an urologischen Problemen empfehlen. Die Ausstattung ist hinsichtlich der Übersichtlichkeit des Drucksatzes und hinsichtlich des sehr reichen und qualitativ durchweg erstklassigen Bildmaterials vorbildlich.

H. Olbing, Essen

Ultraschallgezielte perkutane Punktion und Drainage eines Milzabszesses durch Salmonella heidelberg bei einem Kind

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Zusammenfassung

Bei einem 9¹¹/₁₂ Jahre alten Knaben traten im Anschluß an eine Salmonella-Enteritis Schmerzen im linken Oberbauch auf. Mittels Sonographie wurde die Diagnose Milzabszeß gestellt. Nach ultraschallgezielter Punktion und Drainage sowie entsprechender Chemotherapie kam es zu einer deutlichen Besserung des klinischen Bildes und schließlich zur komplikationslosen Heilung. An Hand einer Literaturübersicht wird auf die Problematik von Milzabszessen und auf die Möglichkeit einer perkutanen Punktionsbehandlung hingewiesen. Bei jeder Erkrankung, die mit Fieber und unklaren Bauchschmerzen einhergeht, sollte man auch an den Milzabszeß denken!

Ultrasonically Guided Percutaneous Puncture and Drainage in a Case of Juvenile Purulent Splenitis Caused by Salmonella Heidelberg

In a boy aged 9 years and 11 months pain developed in the left upper abdomen following Salmonella enteritis infection. Purulent splenitis was diagnosed by sonography. Ultrasonically guided peructaneous puncture and drainage, and appropriate chemotherapy led to a marked improvement in the clinical picture and finally to complication-free remission. In a review of the literature the problems associated with purulent splenitis and the possibility of treating it by percutaneous puncture are pointed out. Purulent splenitis should be considered in all patients with temperatures and unexplained abdominal pain.

Milzabszesse sind sehr selten und stellen immer eine diagnostische und therapeutische Herausforderung dar.

Mit der folgenden, unseres Wissens im deutschen Schrifttum bislang ersten Darstellung einer perkutanen Punktion und Drainage eines Milzabszesses bei einem Kind möchten wir auf die verbesserten diagnostischen und therapeutischen Möglichkeiten, die sich aus der organerhaltenden perkutanen Punktion und Drainage ergeben, hinweisen.

Kasuistik

Der bei Aufnahme 9¹¹/₁₂ Jahre alte Junge erkrankte 6 d vor Aufnahme (gleichzeitig mit seiner Schwester) mit Fieber und Durchfall. Unter symptomatischer Therapie besserten sich die Beschwerden. Schon anfangs bestanden Schmerzen im Mittelbauch, die sich, nachdem Fieber und Durchfall abgeklungen waren, in den linken Oberbauch verlagerten. 4 d nach Erkrankungsbeginn gelang der Nachweis von Salmonella (S.) heidelberg im Stuhl. Bei Aufnahme gab der deutlich krank wirkende Junge Schmerzen unter dem linken Rippenbogen beim Atmen und Druckschmerz in derselben Region an. Fieber bestand nicht.

Klinisch waren keine weiteren pathologischen Befunde zu erheben. Paraklinische Befunde bei Aufnahme:

HK: 0,37, Leukozyten: 3,3 Gpt/l, 0,01 Eosinophile, 0,13 Stabkernige, 0,50 Segmentkernige, 0,27 Lymphozyten, 0,08 Monozyten, 0,01 Lymphoidzellen, ESR: 75/113 mm, 2 Blutkulturen steril.

Na: 141,0 mmol/l, K: 5,0 mmol/l, ALAT: 0,24 nmol/s·L, Gesamteiweiß: 67 g/l, Kreatinin: 68 µmol/l, CRP: 68,5 mg/l. Säure-Base-Werte im Normbereich. Urin: Chemisch, zytologisch und bakteriologisch unauffällig. Yersinien – 0:3-Widal negativ.

Analabstrich: S. heidelberg, resistent gegenüber Ampicillin und Azlocillin, empfindlich gegenüber Cephotiam, Chloramphenicol, Polymyxin B, Gentamicin, Trimethoprim-Sulfonamid.

Röntgen: Leichter Zwerchfellhochstand links mit abgeflachtem Sinus. Magenblase etwas nach medial verdrängt. Die Durchleuchtung zeigt eine verminderte Beweglichkeit des linken Zwerchfells. Lungenzeichnung regelrecht.

Sonografie (Abdomen): 3×3 cm große Raumforderung im oberen Drittel der Milz nahe des Hilus mit echoarmem Rand und echoreichem, unregelmäßig begrenztem Inhalt. Die Milz ist im ganzen deutlich vergrö-

Klin. Pädiatr. 204 (1992) 56–60