UNIVERSITÄTS-
KINDERSPITAL
ZÜRICH

Hypermobilität, Bandlaxität – Bedeutung für die Praxis

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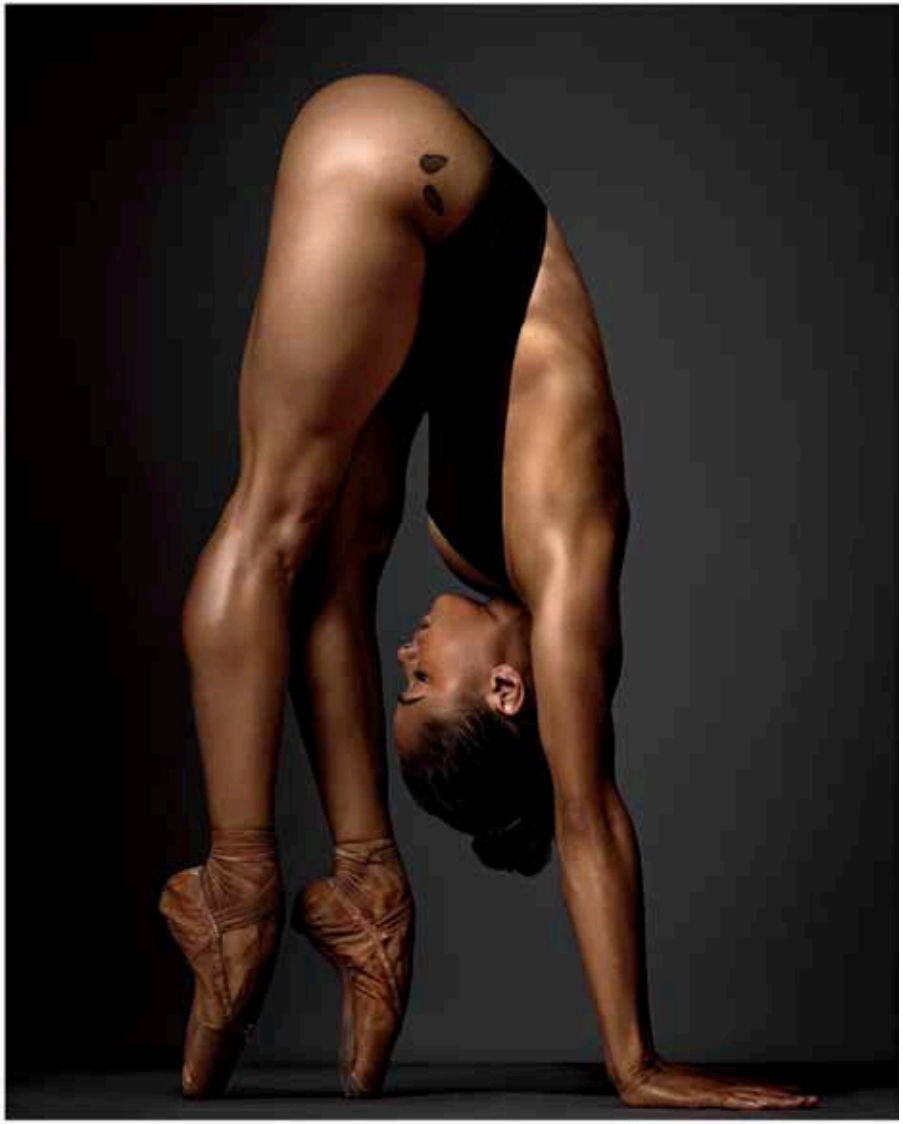
Universitäts-Kinderspital Zürich
Lenggstrasse 30, 8008 Zürich

Das Spital der Eleonorenstiftung

Hyperlaxität

- Hyperlaxität – gibt es das überhaupt? -> Ursachen
- Beighton-Score - ist das noch aktuell? -> wie gemessen, weitere Scores
- Typische Beschwerden? -> weitere Symptome
- Hyperlaxität, was für Abklärungen müssen wann eingeleitet werden? -> Differentialdiagnosen / Bindegewebekrankheiten

Hypermobilität



Misty Copeland,

Hypermobilität-Symptome

- JROM aktiv oder passiv über altersentsprechende Norm bewegbar
- Grenze zwischen normal und hypermobil fließend
- Generalisiert, beidseitig auftretend
- Wirbelsäule mit einbezogen (Haltungsinsuffizienz)
- Verschiedene grosse und kleine Gelenke betroffen
- Keine Schwellung, Überwärmung
- Instabilität, Subluxationen, Luxationen
- Blockaden
- Schmerzen


Ursache

Bindegewebe

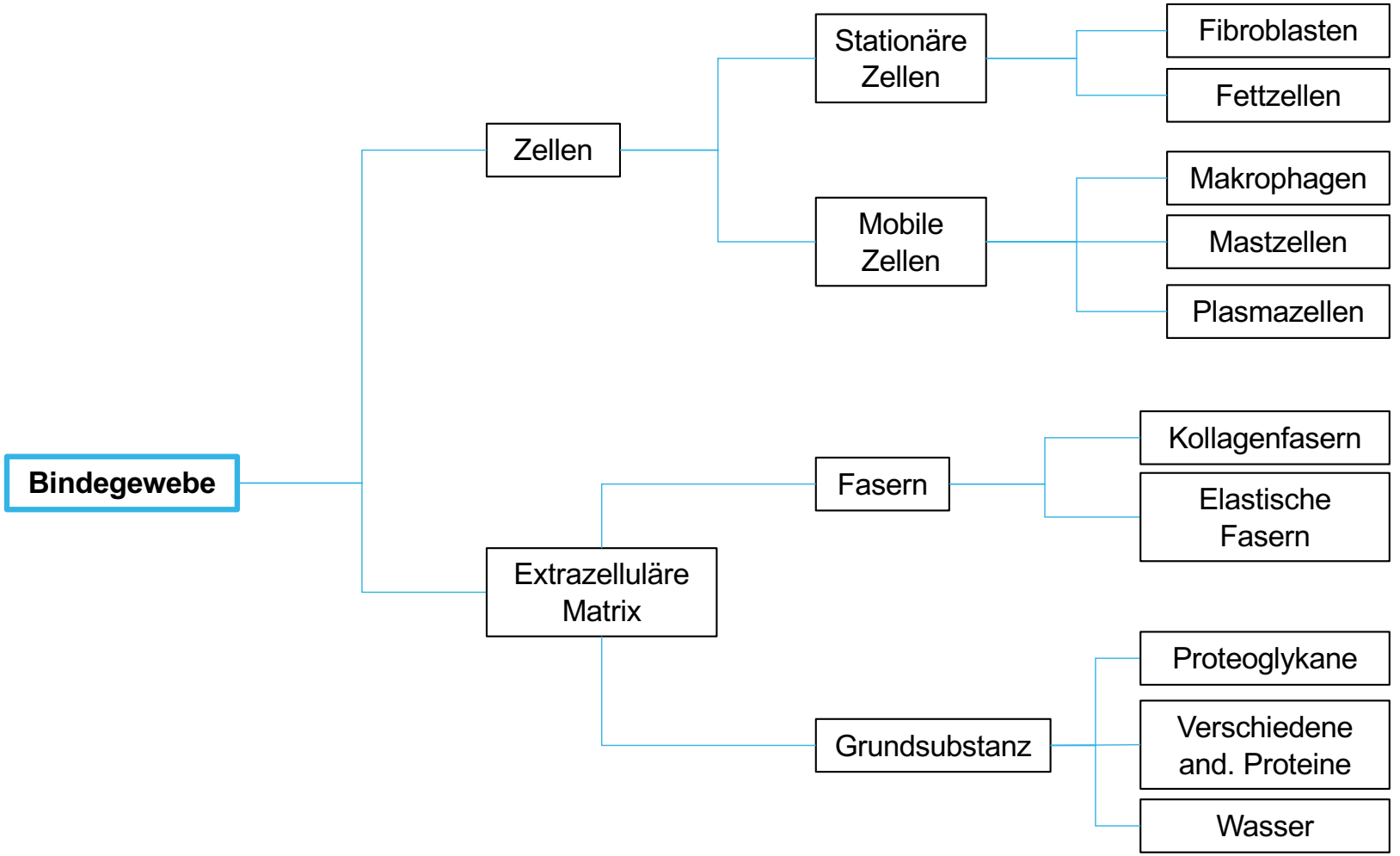
- umfassendes Netzwerk vom Scheitel (Hirnhäuten) bis zur Sohle (Plantarfaszie)

- Binde- und Stützfunktion in allen Strukturen / Organen

- gigantisches Kommunikationssystem (Rezeptoren, Nervenendigungen, etc.)

- gewährleistet harmonisches Zusammenspiel aller Körperteile

Bindegewebeaufbau



Funktionen der Bauteile

• Kollagene	→	Zugfestigkeit
• Elastische Fasern	→	Elastizität
• Proteine	→	Struktur
• Wasser	→	Druckaufnahme
• Zellen	→	Proteinproduktion, Immunität, etc.


Scores

Kriterien hypermobiles Ehlers Danlos Syndrom

The International Consortium on Ehlers-Danlos Syndromes & Related Disorders

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)

This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society.

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 ***and*** 2 ***and*** 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:

- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

Beighton Score: _____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:

- ☐ Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)

- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women without a history of significant gain or loss of body fat or weight
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ Arachnodactyly, as defined in one or more of the following:
  (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: _____/12

Feature B

- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥3 months
- ☐ Recurrent joint dislocations or frank joint instability, in the absence of trauma

CRITERION 3 – All of the following prerequisites MUST be met

1. Absence of unusual skin fragility, which should prompt consideration of other types of EDS
2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired CTD (e.g. Lupus, Rheumatoid Arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted toward a diagnosis of hEDS in this situation.
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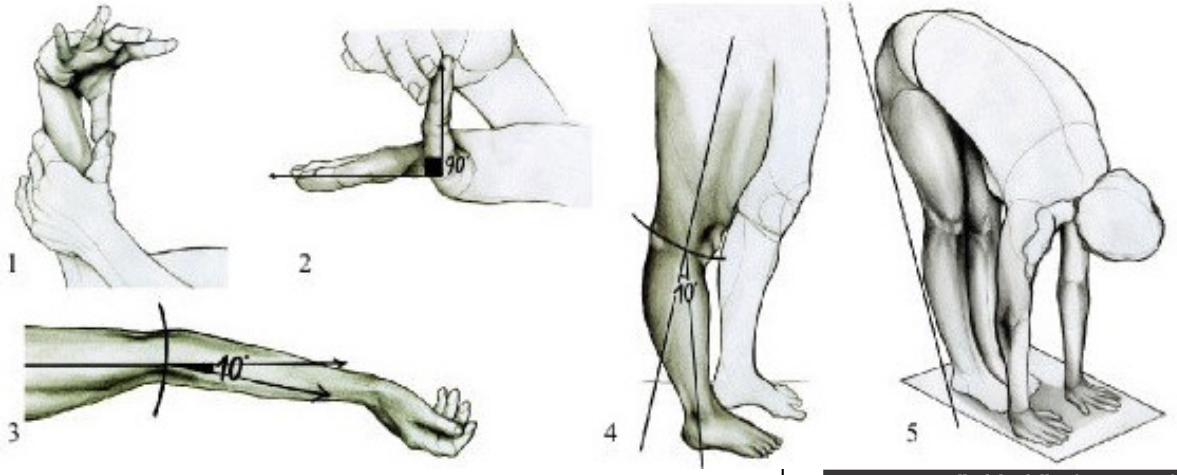
Diagnosis: _____

v9

Kriterien hypermobiles Ehlers Danlos Syndrom

Beighton Score / Hypermobilität



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fat or weight

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Diagnosis: _____

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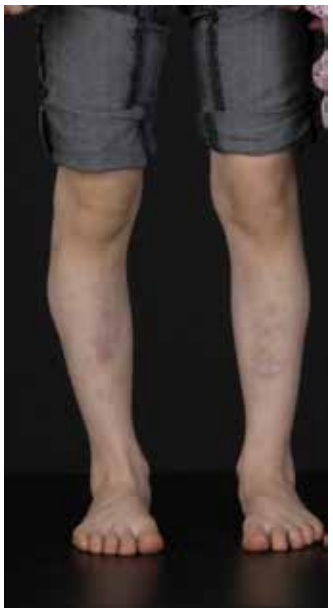
Kriterien hypermobiles Ehlers Danlos Syndrom



HAUT

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- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)

- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women without a history of significant gain or loss of body fat or weight
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
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- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: _____/12

Feature B

- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥3 months
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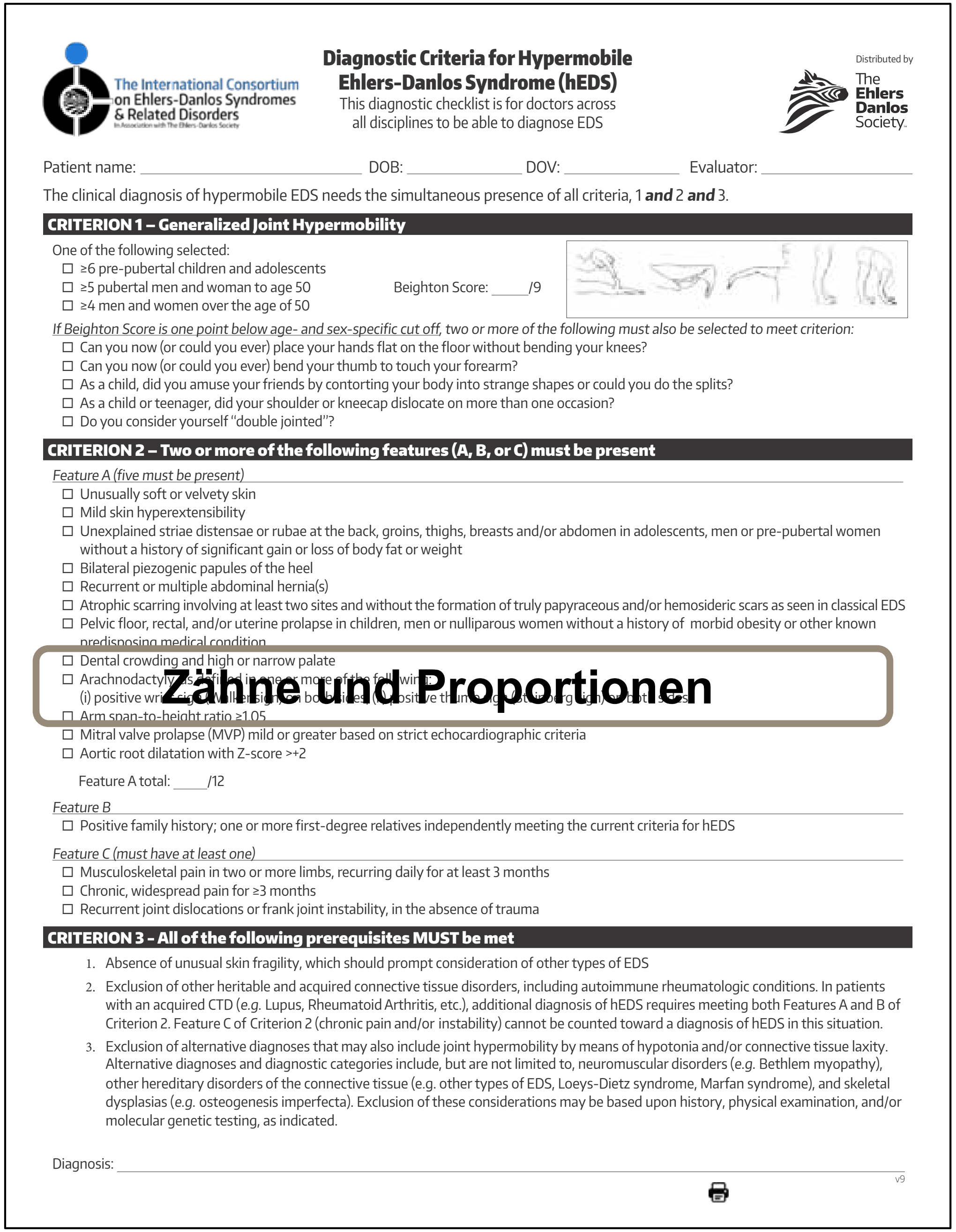
Diagnosis: _____

v9

Kriterien hypermobiles Ehlers Danlos Syndrom



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Feature A total: ____/12

Feature B

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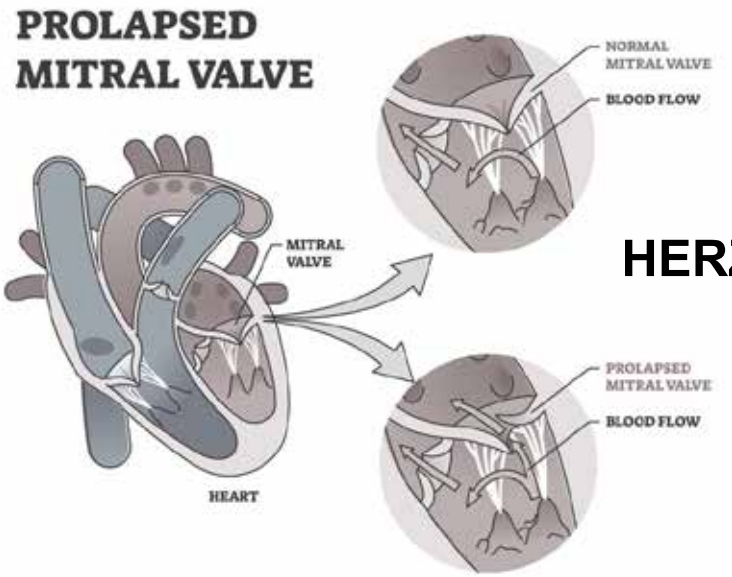
Diagnosis: ______________________

v9

Zähne und Proportionen

Kriterien hypermobiles Ehlers Danlos Syndrom



HERZ

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Feature A total: _____/12

Feature B

- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
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Kriterien hypermobiles Ehlers Danlos Syndrom

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Feature A total: _____/12

Feature B

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Feature C (must have at least one)

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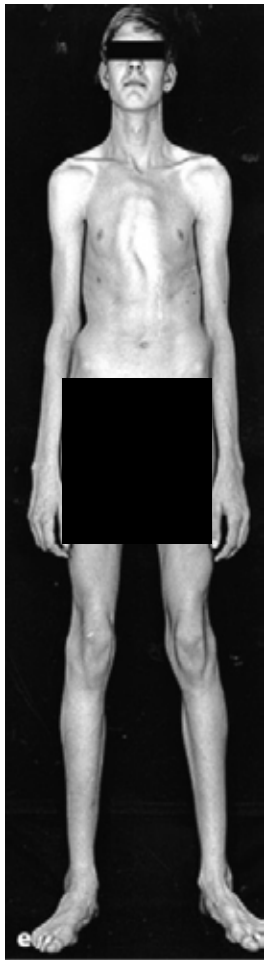
Diagnosis: _____

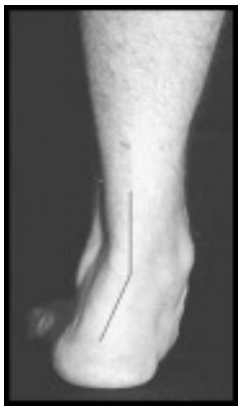
v9

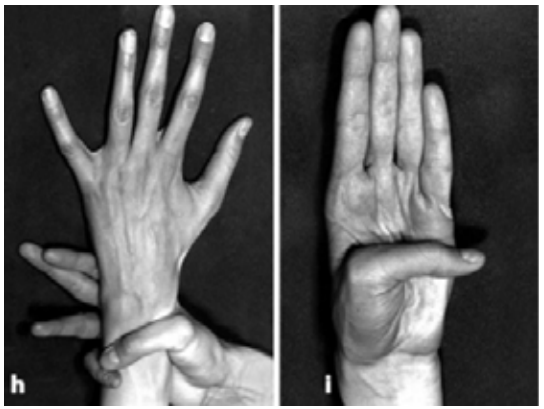
Marfan Score



Scoring of systemic features.

Feature	YES	NO
Wrist AND thumb sign	3	0
Wrist OR thumb sign	1	0
Pectus carinatum deformity	2	0
pectus excavatum or chest asymmetry	1	0
Hindfoot deformity	2	0
Plain flat foot	1	0
Pneumothorax	2	0
Dural ectasia	2	0
Protrusio acetabulae	2	0
Reduced US/LS and increased armspan/height	1	0
Scoliosis or thoracolumbar kyphosis	1	0
Reduced elbow extension	1	0
3/5 facial features	1	0
Skin striae	1	0
Myopia	1	0
Mitral valve prolapse	1	0

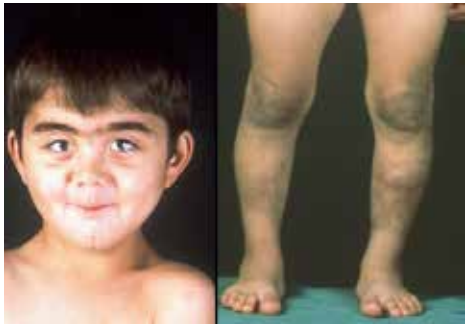





Ehlers-Danlos-Syndrome

Ehlers-Danlos-Syndrome

Sehr heterogene Gruppe



cEDS (COL V)

hEDS (?)

Haut

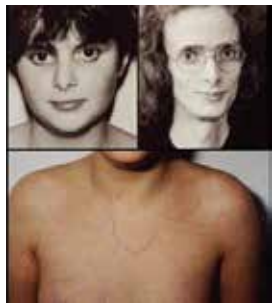
- hyperelastisch
- fragil, Narben, Suffusionen

Gelenke

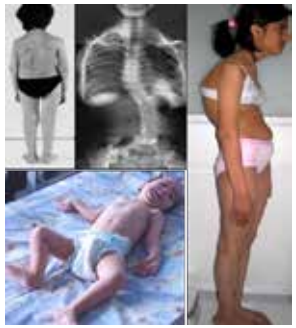
- hypermobil, Luxationen

Herz und Gefässe

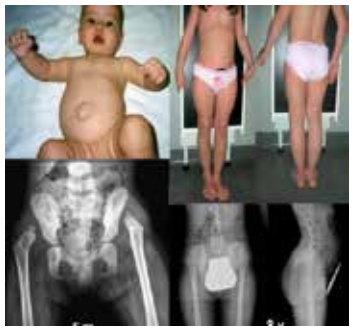
- Hernien, Herzklappeninsuffizienz
- fragile Arterien



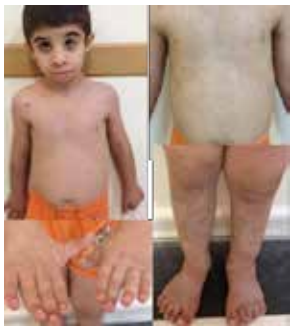
vEDS (COL III)



kEDS (PLODI)



aEDS (COL I)



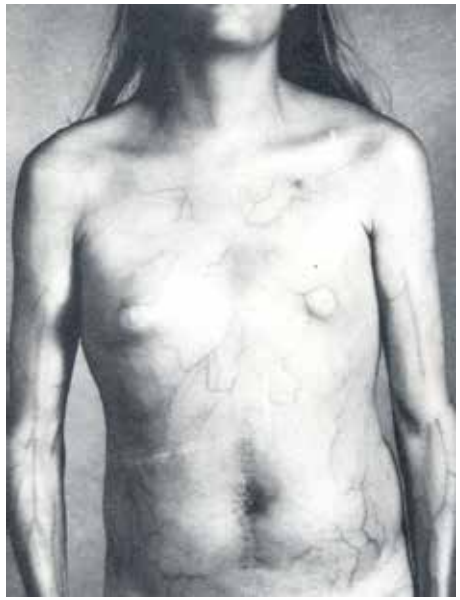
dEDS (ADAMTSL2)

Bilder mit freundlicher Genehmigung von Prof. Dr. M. Rohrbach, Kinderspital Zürich

Vaskuläres Ehlers-Danlos-Syndrom

Hauptkriterien

- Variante in COL3A1, autosomal dominanter Erbgang
- positive Familienanamnese
- Arterielle Rupturen in jungem Alter
- Ruptur von inneren Organen (Colonperforation, Uterusruptur, Carotis-cavernous sinus Fistel ohne Trauma)
- Spontanpneumothorax



Nebenkriterien:

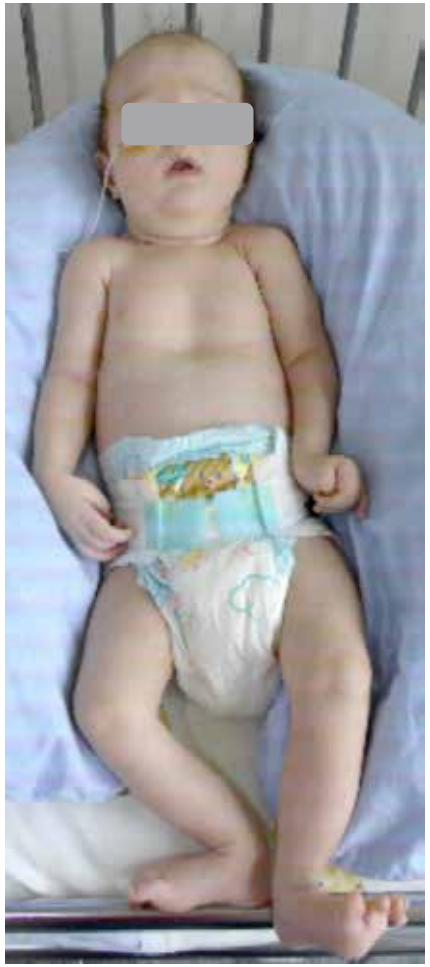
- Dünne Haut mit sichtbarem venösem Geflecht
- Typisches Gesicht
- Hypermobilität

Kyphoskoliotisches Ehlers-Danlos-Syndrom

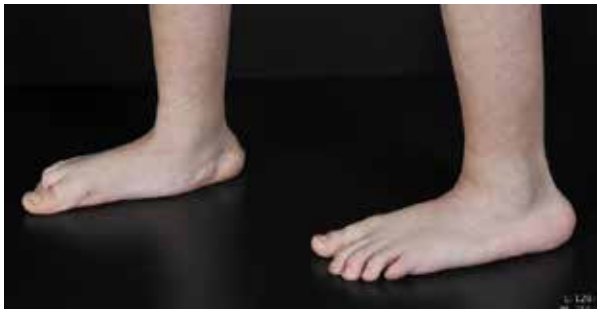
Klinik:
- Hypermobilität, Luxationen
- Gespaltene Uvula
- Muskuläre Hypotonie
- Klumpfüsse
- Pectus excavatum, Skoliose
- Überdehnbarkeit der Haut
- Wundheilungsstörung
- Hörminderung
- Cardiovaskuläre Manifestationen
- Marfanoider Grosswuchs

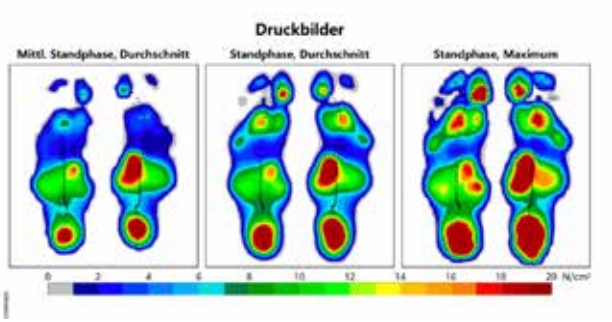
Genetik:
- PLOD1- o. FKBP14-Mutation

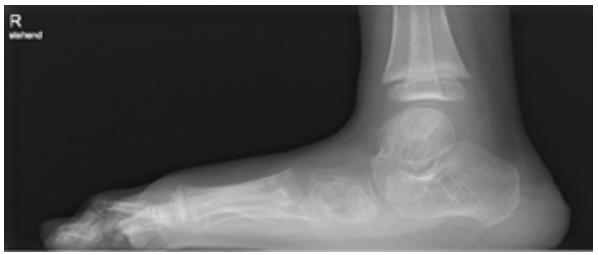
-> Faltung/Stabilisierung Kollagene

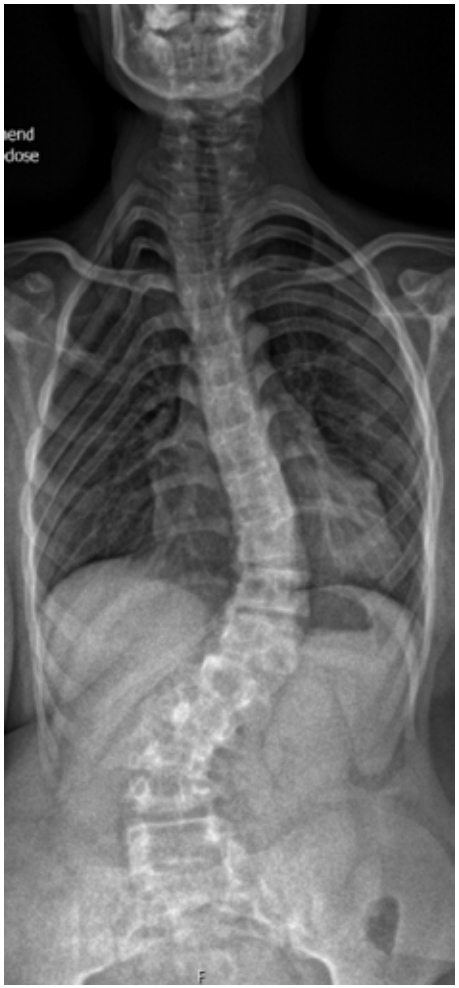


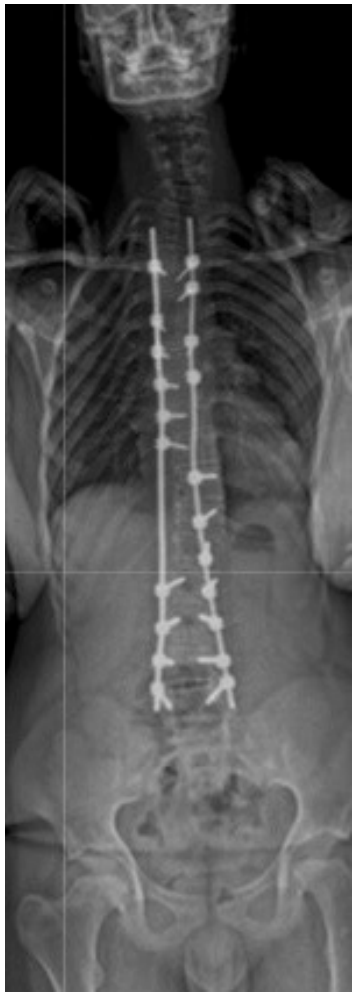
Kyphoskoliotisches Ehlers-Danlos-Syndrom

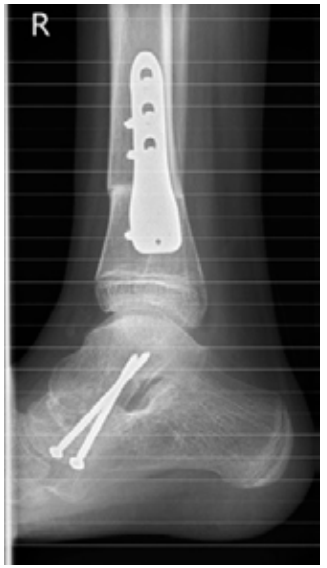












Marfan und Loeys Dietz-Syndrom

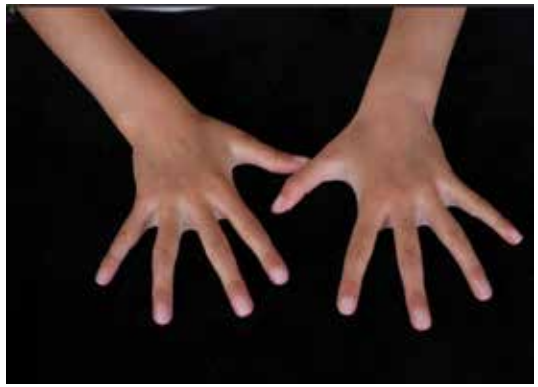
Marfan-Syndrom

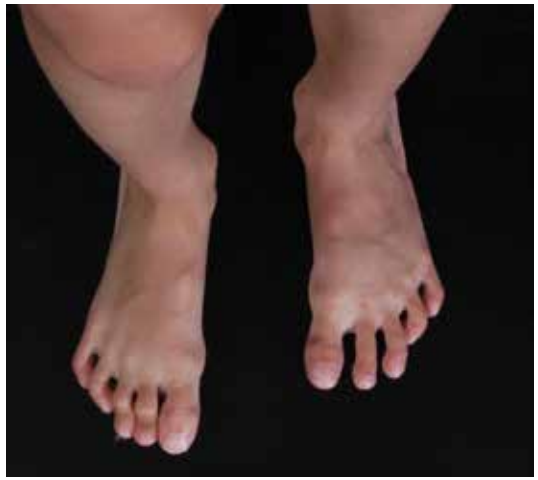
Klinik:

- Sehr variabel
- Grosswuchs, Arachnodaktylie
- Pes planovalgus
- Skoliose, Trichter- / Kielbrust
- Hypermobilität
- Ellebogenbeuegkontraktur
- Herz- und Gefässbeteiligung (Aortendilatation, -dissektion)
- Augenbeteiligung (Myopie)

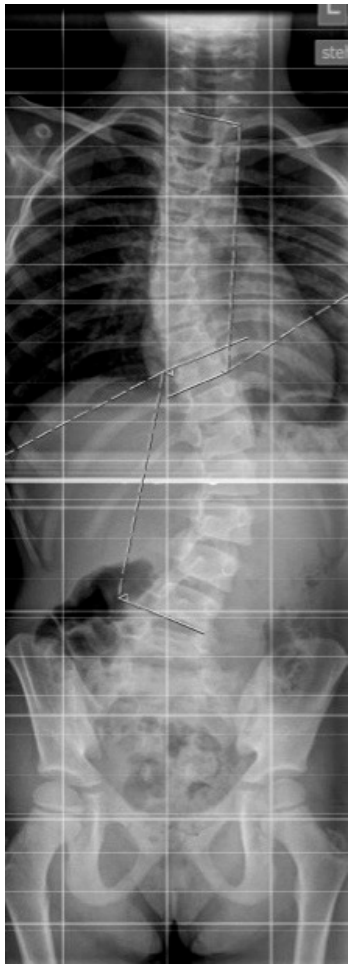
Genetik:

- FBN1-Mutation -> Fibrillin

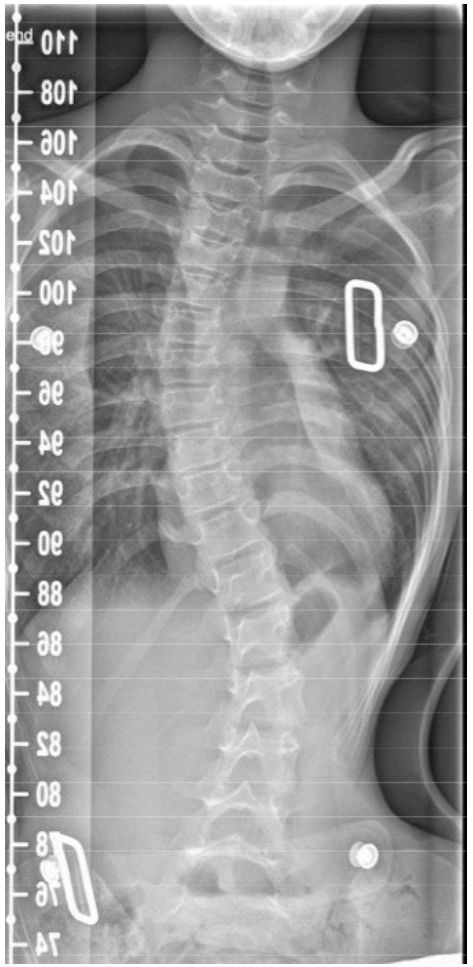




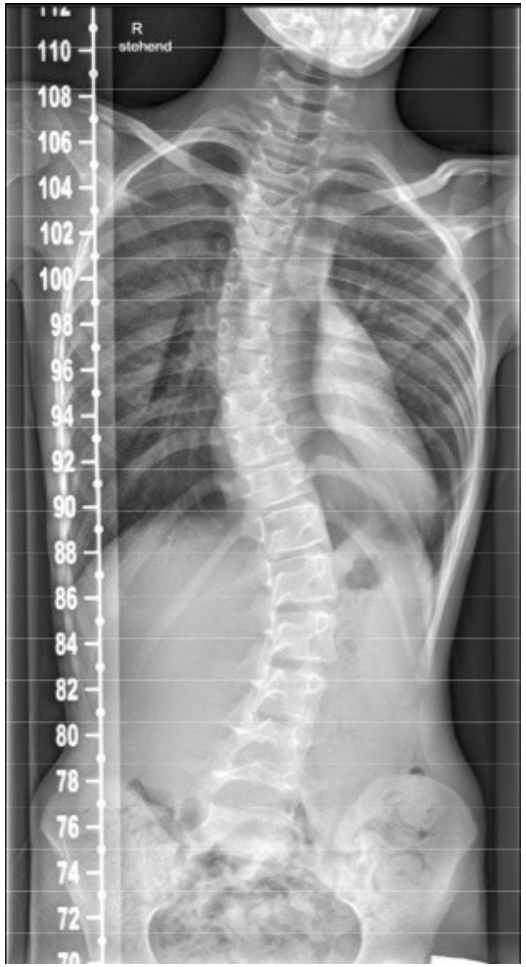
Marfan-Syndrom



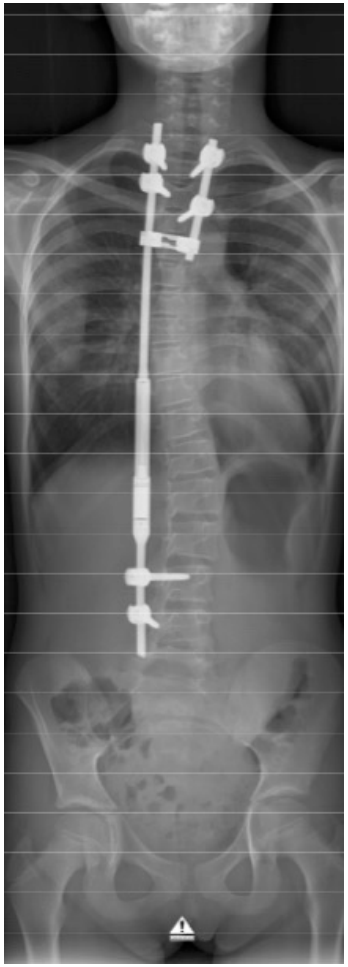
6/08



12/10



3/11



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Loeys Dietz Syndrom

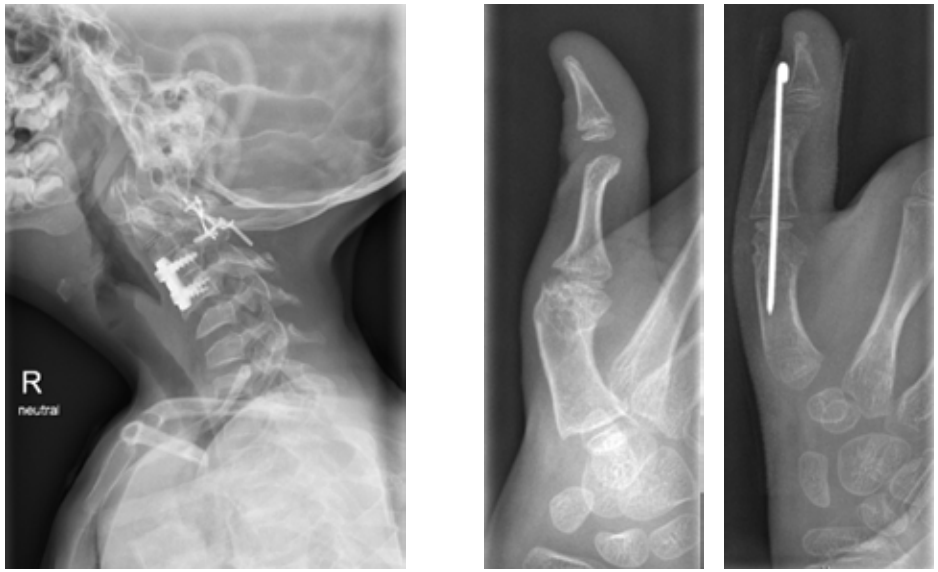
Klinik:

- Sehr variabel
- Auffällige Fazies, Gaumenspalte
- Arachnodaktylie
- Pes planovalgus
- Skoliose, Trichter- / Kielbrust
- Hypermobilität, Luxationen
- Skoliose, Listhese
- Sponatane Bandrupturen
- Herz- und Gefässbeteiligung (Dilatationen, -dissektionen)

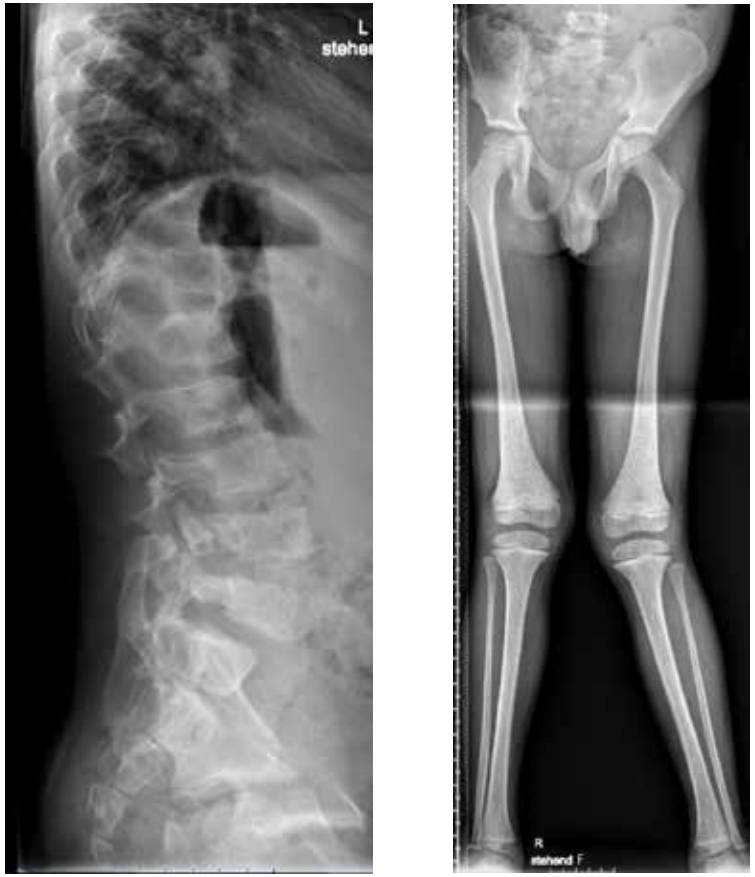
Genetik:

- Heterogen: TGFBR1, TGFBR2, SMAD3, etc. -> growth factor

Loeys-Dietz-Syndrom



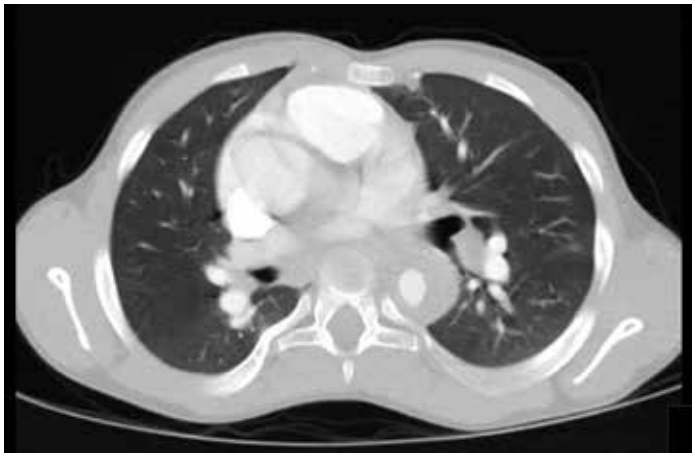
2012-2014

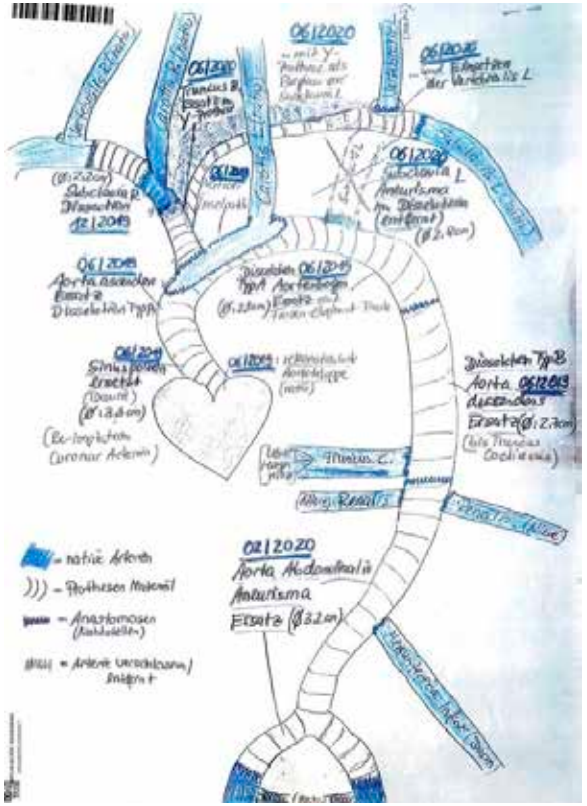


2016

Loeys-Dietz-Syndrom

- Thorakaler Aortenersatz 12.06.2009
- Thorakoabdomineller Aortenersatz 14.06.2019
- Aneurysmaresektion Tr brachiocephalicus 11.12.2019
- Abdominaler Bifurkationsersatz 20.02.2020
- Thorakale Bifurkationsprothese 03.06.2020
- Mitralklappenrekonstruktion 25.01.2022





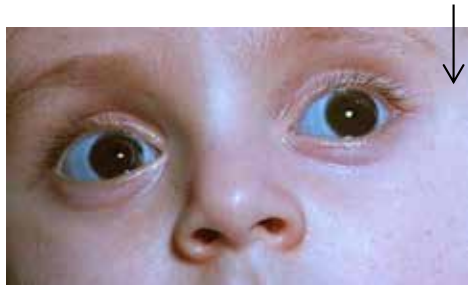
Osteogenesis imperfecta

Osteogenesis imperfecta = Glasknochenkrankheit

Genetik: COL1A, COL1A, FKBP10, PLOD2 -> Kollagen I

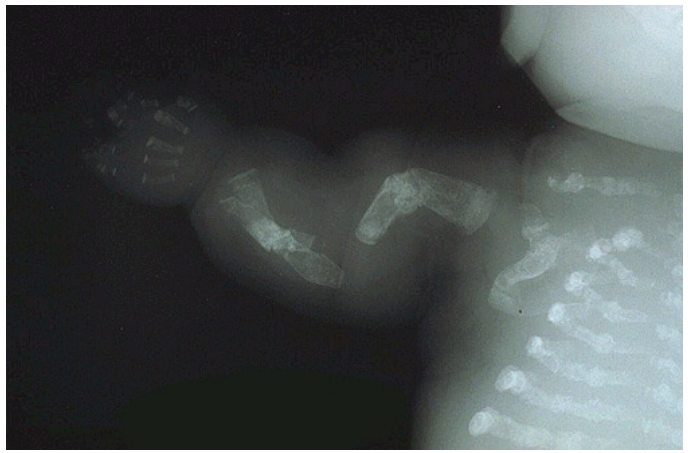
Habitus:

- Blaue Skleren
- Hypermobilität



Knochen:

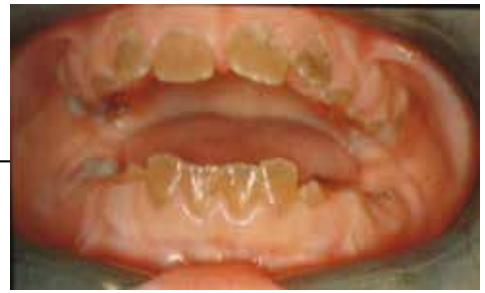
- Osteopenie, Platyspondylie, Frakturen
- Deformitäten, Skoliose



Zähne:

- Schmelzdefekte, Kariesanfälligkeit, Dentinogenesis imperfecta

- Herz / Gefässe: Dilatataionen, Klappenveränderungen

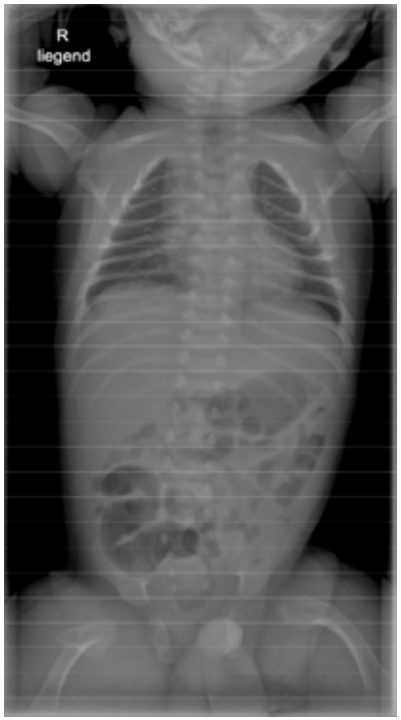


Bilder mit freundlicher Genehmigung von Prof. Dr. M. Rohrbach, Kinderspital Zürich

Osteogenesis imperfecta

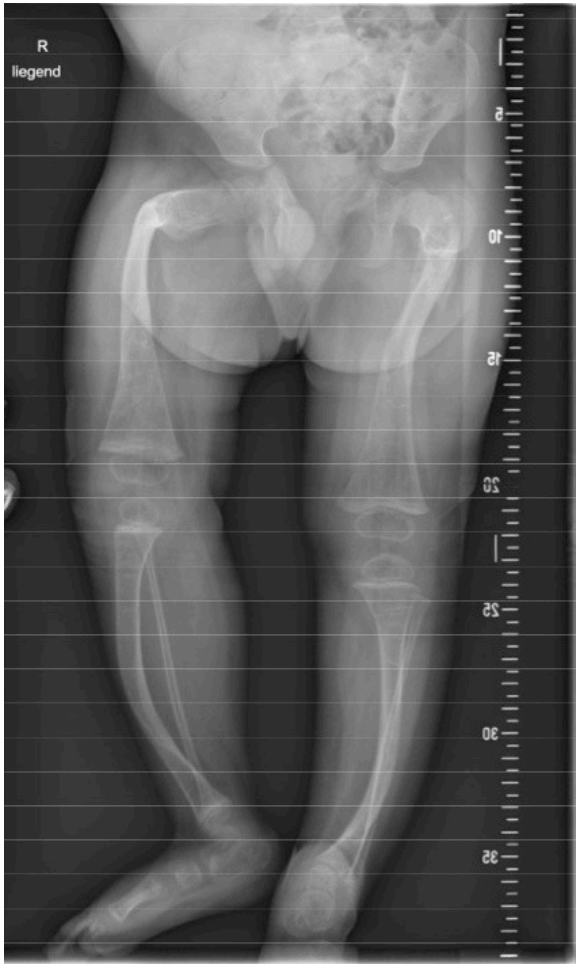
- Mutation in COL1A1

- pränatale Frakturen (distaler Radius rechts, Humerus links, Femurschaft bds)
- postnatale Humerusschaftfraktur rechts, subcapitaler Humerusfraktur rechts, distaler Radiusfraktur links
- proximale Femurfraktur links 10/2012
- Grünholzfraktur Tibia rechts 10/2012
- Femurschaftfraktur rechts 09/2013
- Tibiafraktur links 12/2016
- distale Humerusschaftfraktur links 12/2018
- Refraktur distaler Humerus links 04/2020
- Fraktur Femur und Nagelversagen Tibia rechts 03/2021
- distale Humerusfraktur 03/2022



- Aortendilatation

Osteogenesis imperfecta, N.Z, ♂, 20.04.2012, Neridronat seit 09/2012



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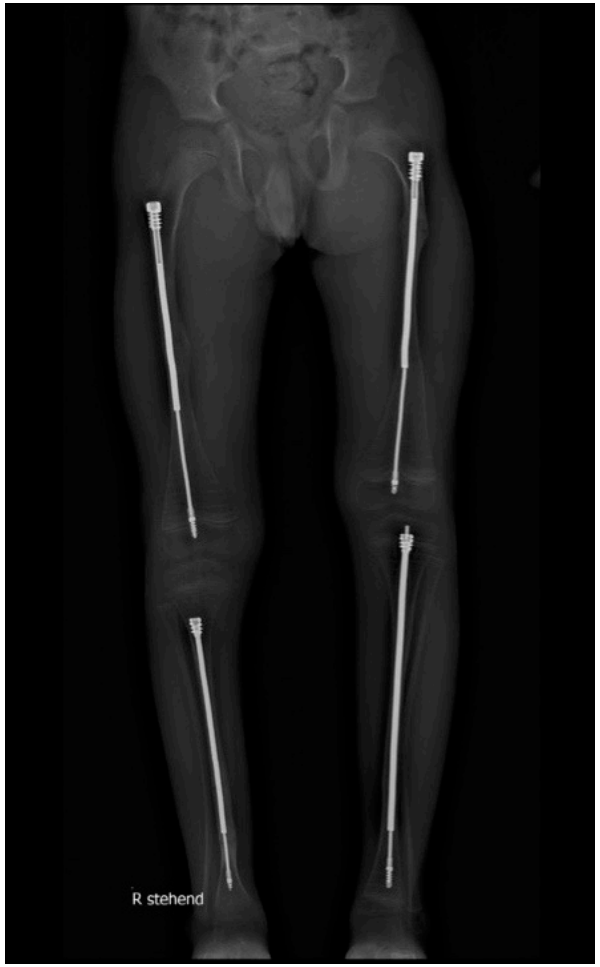

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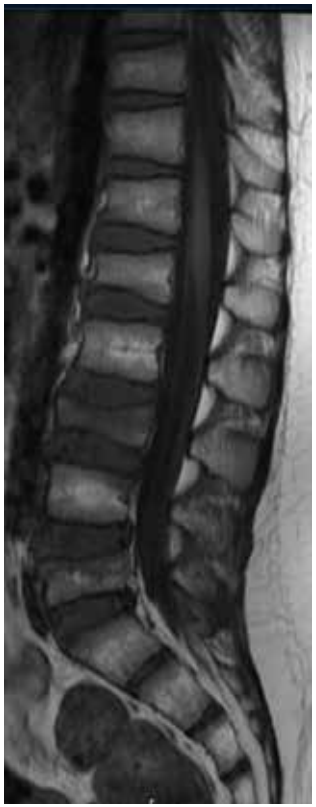
Osteogenesis imperfecta

- Mutation in COL1A1

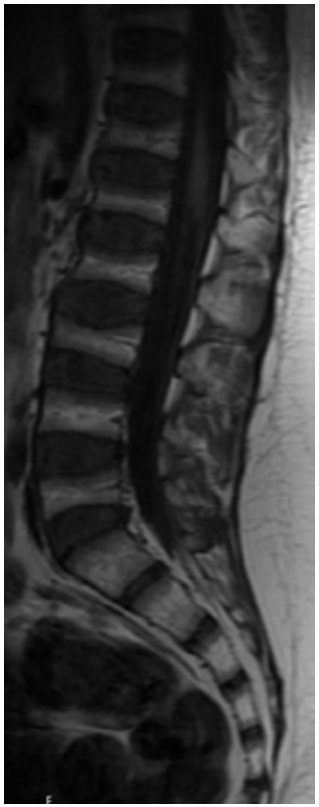
- St.n. Unterschenkelfraktur i. Kindesalter
- St.n. multiplen alten und frischen Wirbelkörperimpressionsfrakturen C7, Th3-12, L1, L3 - L5 nach fraglich adäquaten Traumata ED 10/2016
- St.n. Wirbelkörperfrakturen Th8, Th10, L1 und L2 mit progredienter Sinterung, Deckplattenimpression und Höhenminderung am 30.05.2017
-
- Bisphosphonattherapie seit 07/2017 (Neridronat)

- leichtgradiger Gangunsicherheit bei Grob-Motorik-Störung und Hypermobilität

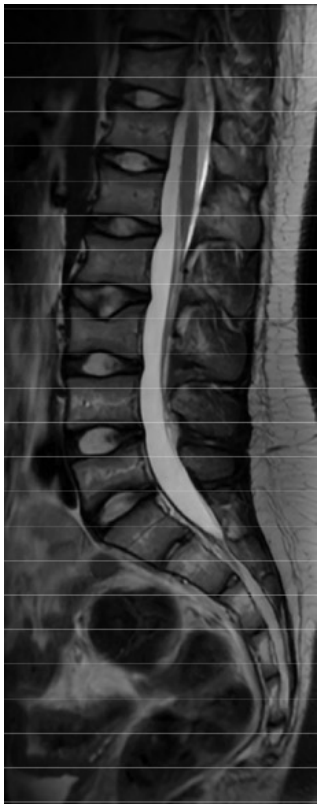
Osteogenesis imperfecta, F.R., ♂, 03.01.2006



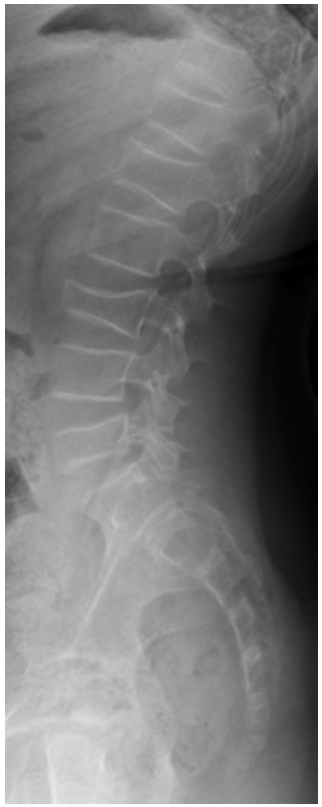
10 J.



13 J.



16 J.



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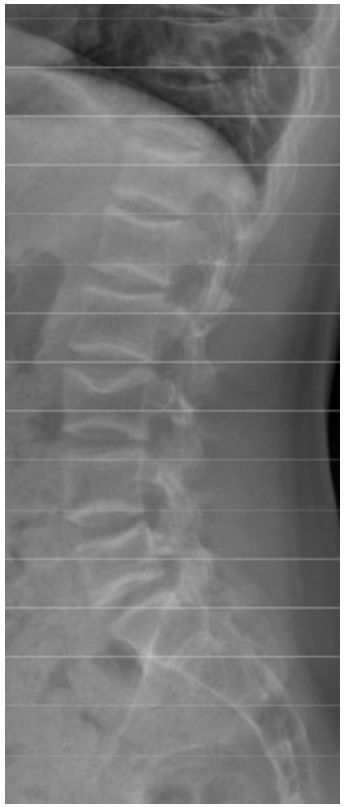

16 J.

Fragen?