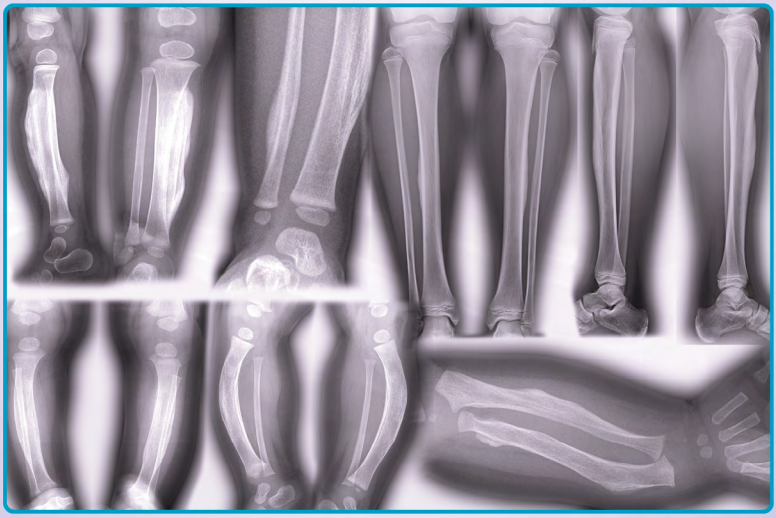
Pohybové ústrojí

Pokroky ve výzkumu, diagnostice a terapii



Vydává
Společnost pro pojivové tkáně ČLS J. E. Purkyně z.s.
Ortopedicko-protetická společnost ČLS J. E. Purkyně z.s.
Ambulantní centrum pro vady pohybového aparátu, s.r.o.

ročník 28 / 2021 číslo 2

EMBASE / Excerpta Medica | Bibliographia medica Čechoslovaca

ISSN 2336-4777

ORLING

ZALOŽENO ROKU 1992

BIOAKTIVNÍ KOLAGENNÍ PEPTIDY REGENERUJÍ

Kolagen je nezbytný pro pohyblivost kloubů, stabilitu kostí, odolnost a pevnost vazů a šlach a také pro zdravé svaly a hojně se vyskytuje i v cévách, meziobratlových ploténkách, hematoencefalické bariéře a rohovce, dentinu a střevní stěně – kolagen je životně důležitá složka celého těla.

Kolagenní peptidy zvyšují syntézu kloubního kolagenu a proteoglykanů

Nejen sportovci jsou ve zvýšené míře náchylní ke kloubním problémům a léčba se u nich nijak neliší od jejího zvládání u běžné populace. Hlavním cílem je minimalizovat bolestivost a zlepšit funkčnost kloubů. Klinická studie provedená v Penn State University testovala účinek kolagenních peptidů na studenty sportovních škol, kteří trpěli kloubními problémy v důsledku mechanické zátěže. V porovnání s kontrolní skupinou došlo u studentů, kteří užívali kolagenní peptidy, k **výraznému snížení kloubních potíží a také ke zlepšení pohyblivosti.** Tyto pozitivní účinky byly patrné zejména u účastníků s problémy kolenních kloubů pocházejících z mechanické zátěže. *(Clark K. , Sebastianelli W., Flechsenhar K., Aukermann D., Meza F., Millard R., Deitch J., Sherbondy P., Affiliations A., 24-Week study on the use of collagen hydrolysate as a dietary supplement in athletes with activity-related joint pain, Curr Med Res Opin, 2008 May;24(5):1485-96)*

Významný je i vliv kolagenních peptidů na hustotu kostí, zejména u osob s osteoporózou či osteopenií, potvrzeno už v roce 2010 pilotní studií s doplňkem stravy Calcidrink®.

V této studii se řešil „Vliv suplementace kolagenními peptidy, vápníkem a vitaminem D, resp. Calcidrinkem® na úbytek kostní hmoty a remodelaci kosti u postmenopauzálních žen s osteopenií“ *(Ortopedie 2010, Gabriela Šimková, Revmatologická ambulance 1. PP Kladno)*. Výsledky byly velmi nadějné. U žádné pacientky se nevyskytly během sledovaného období jednoho roku žádné nové nízkozátěžové zlomeniny. Cílem bylo prokázat účinek pravidelného užívání přípravku Calcidrink (vitamin D, kalcium a kolagenní peptidy) na snížení úbytku kostní hmoty u postmenopauzálních žen s osteopenií. Výsledky studie tento efekt potvrdily.

Doplňky stravy Geladrink® a Calcidrink® s vysokým obsahem ověřených kolagenních peptidů Gelita®

Kolagenní peptidy Gelita® jsou obsaženy ve fyziologicky účinné dávce v originálních produktech české firmy Orling, s využitím všech nových poznatků ohledně jejich působení.

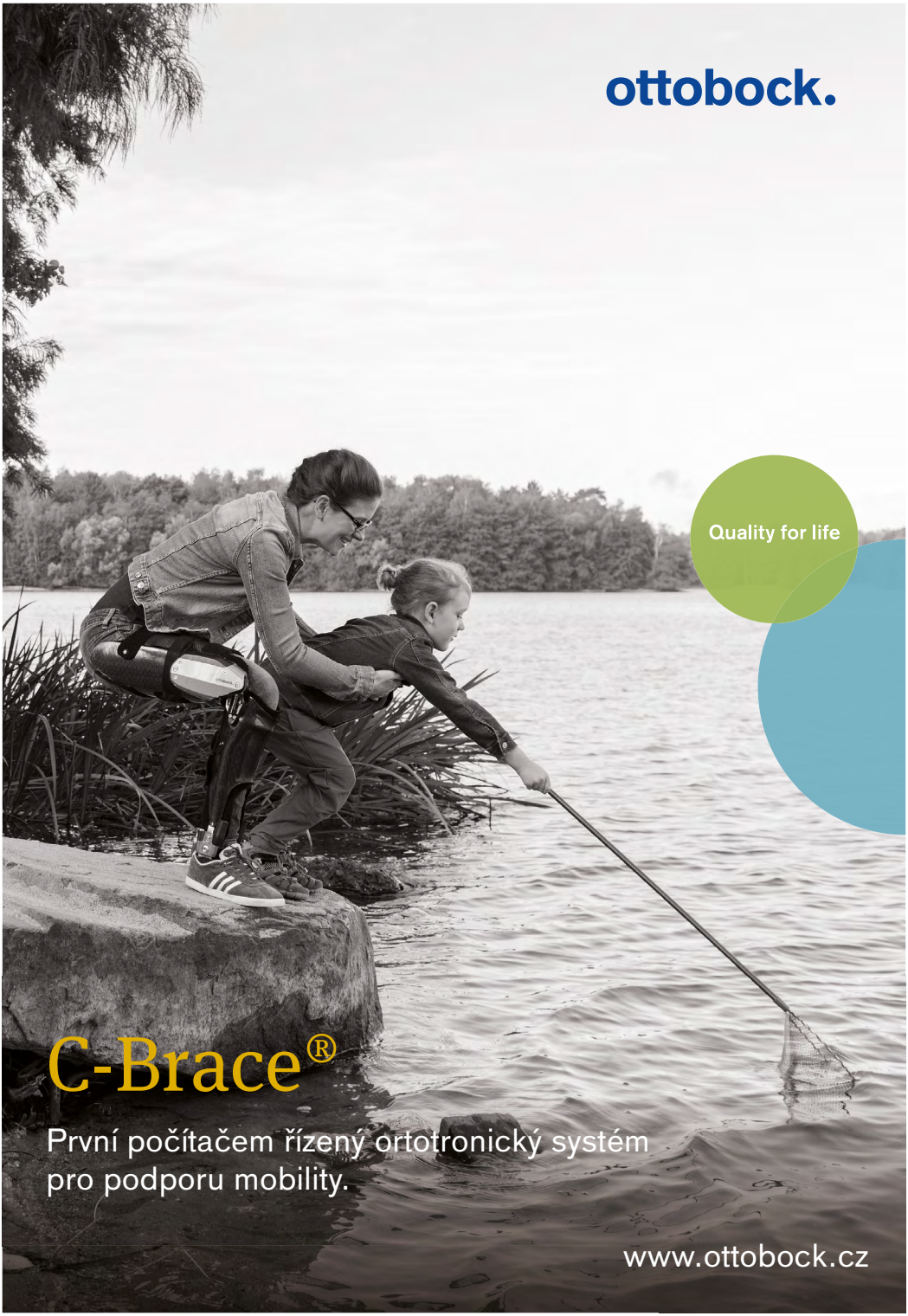
Prof. MUDr. Milan Adam, DrSc. byl první, kdo objevil obrovský potenciál kolagenních peptidů a v průběhu let jej další vědci a lékaři opakovaně prokázali a ještě rozšířili oblasti použití, pro které jsou kolagenní peptidy vhodné.

Prof. MUDr. Milan Adam, DrSc. Dr.h.c.
český revmatolog, zakladatel kloubní výživy Geladrink®

GELADRINK® a CALCIDRINK®
- PRO LEPŠÍ KVALITU ŽIVOTA

VYVINUTO A VYROBENO V ČESKÉ REPUBLICE

Více informací na WWW.ORLING.CZ


ottobock.
Quality for life
C-Brace®
První počítačem řízený ortotronický systém
pro podporu mobility.
www.ottobock.cz

POHYBOVÉ ÚSTROJÍ

ročník 28, 2021, číslo 2 | datum vydání: 22. 03. 2022

Pohybové ústrojí. Pokroky ve výzkumu, diagnostice a terapii.

ISSN 2336-4777 (od roku 2013 pouze on-line verze)
Vydává Společnost pro pojivové tkáně ČLS J. E. Purkyně z.s.
& Ortopedicko-protetická společnost ČLS J. E. Purkyně z.s.
& Ambulantní centrum pro vady pohybového aparátu, s. r. o.
Excerpováno v Excerpta Medica a Bibliographia medica Čechoslovaca.
Návrh a grafická úprava obálky Pavel Lorenc.
Časopis je na Seznamu recenzovaných neimpaktovaných periodik vydávaných v České republice. Dvě čísla časopisu vycházejí v elektronické verzi jako ročník s průběžným vydáváním příspěvků po recenzi.
Při příležitosti sympozií je dvakrát ročně vydáváno supplementum.

Pro současné odběratele časopisu PÚ a další zájemce doporučujeme přihlásit se na http://www.pojivo.cz/en/newsletter/, zadat jméno a e-mailovou adresu, na kterou bude časopis posílán. Na webové doméně SPT ČLS JEP http://www.pojivo.cz/cz/pohybove-ustroji/ naleznete ve formátu PDF všechna jednotlivá čísla a dvojčísla časopisu (včetně Supplement) vydaná od roku 1997.
Rukopisy zasílejte na adresu *profesor MUDr. Ivo Mařík, CSc., Olšanská 7, 130 00 Praha 3, (ambul_centrum@volny.cz)* ve formátu doc. Vydavatel upozorňuje, že za obsah inzerce odpovídá výhradně inzerent. Časopis, jakožto nevýdělečný, neposkytuje honoráře za otištěné příspěvky.

LOCOMOTOR SYSTEM

Advances in Research, Diagnostics and Therapy

Published by The Society for Connective Tissues, Czech Medical Association of J. E. Purkyně, Prague, Society for Prosthetics and Orthotics, Czech Medical Association of J. E. Purkyně, Prague, Czech Republic and Centre for Defects of Locomotor Apparatus Prague, Czech Republic.

Call for papers

Support this journal by sending in your best and most interesting papers. The issue of the journal is published during whole year after proof acceptation of the reviewers. In occasion of the symposia (twice a year) is published the supplement.

Submitted papers: Locomotor System will review for publication manuscripts engaged in diagnostics and interdisciplinary treatment of genetic and metabolic skeletal disorders, limb anomalies, secondary osteoporosis, osteo/spondyloarthritis and another disorders that negatively influence development and quality of locomotor apparatus during human life. Both papers on progress in research of connective tissue diagnostics, medical and surgical therapy of multiple congenital abnormalities of skeleton mainly in the fields of paediatric orthopaedic surgery and plastic surgery, orthotics and prosthetics treatment, and papers dealing with biomechanics, clinical anthropology and paleopathology are appreciated.

The journal has an interdisciplinary character which gives possibilities for complex approach to the problems of locomotor system. The journal belongs to clinical, preclinical and theoretical medical branches which connect various up-to-date results and discoveries concerned with locomotor system. You can find the volumes of Locomotor System journal at http://www.pojivo.cz/cz/pohybove-ustroji/ since 1997 (free of charge). Since 2013 only electronic edition of the journal is available. That is why we recommend to all subscribers and those interested apply at http://www.pojivo.cz/en/newsletter, enter personal data, titles and e-mail address where the journal will be mailed.

Abstracts of presented papers are excerpted in EMBASE/Excerpta Medica (from the year 1994) and in the Bibliographia medica Čechoslovaca (from the year 2010). We prefer the manuscripts to be prepared according to Uniform Requirements for Manuscripts Submitted to Biomedical Journals (Vancouver Declaration, Brit med J 1988; 296, p. 401–405).

28. ročník časopisu Pohybové ústrojí, pokroky ve výzkumu, diagnostice a terapii je věnován jubilantům, členům Společnosti pro pojivové tkáně ČLS J.E. Purkyně a členům redakční rady časopisu

prof. MUDr. Janě Pařízkové, DrSc. – 90 let,
prof. Ing. Janovi Čulíkovi, DrSc. – 85 let,
prof. Ing. Miroslavovi Petrtýlovi, DrSc. – 80 let,
prof. MUDr. Václavovi Smrčkovi, CSc. – 70 let,
RNDr. Daniele Zemkové, CSc. – 70 let,
MUDr. Petrovi Krawczykovi – 55 let,
odb. asist. MUDr. Josefovi Krausovi, CSc. – 70 let
a Ing. Pavlovi Černému, Ph.D. – 60 let.

The 28th volume of Locomotor System journal,
is dedicated to the anniversary of

Professor Jana Pařízková, M.D., DSc. – 90 years,
Professor Ing. Jan Čulík, DSc. – 85 years,
Professor Ing. Miroslav Petrtýl, DSc. – 80 years,
Professor Václav Smrčka, M.D., Ph.D. – 70 years,
RNDr. Daniela Zemková, Ph.D. – 70 years,
Petr Krawczyk M.D. – 55 years,
Assistant Professor Josef Kraus, M.D., Ph.D. – 70 years
and Ing. Pavel Černý, Ph.D. – 60 years

POHYBOVÉ ÚSTROJÍ, 28, 2021, č. 2

Pokroky ve výzkumu, diagnostice a terapii.
Datum vydání: 22. 03. 2022

OBSAH

KASUISTIKY

ZPRÁVY

LOCOMOTOR SYSTEM, 28, 2021, No. 2

Advances in Research, Diagnostics and Therapy

CONTENT

CASE REPORTS

NEWS

SLOVO ČTENÁŘŮM

Vážení čtenáři, autoři a inzerenti!

Děkujeme za Vaši nezastupitelnou pomoc při tvorbě mezioborového odborného recenzovaného časopisu *„Pohybové ústrojí – pokroky ve výzkumu, diagnostice a terapii* (dále PÚ).“

Všechna čísla časopisu (včetně Supplement) vydaná od roku 1997 najdete ve formátu PDF na webové doméně Společnosti pro pojivové tkáně ČLS JEP z.s. http://www.pojivo.cz/cz/ pohybove-ustroji/ (bezplatný přístup).

Časopis PÚ byl v roce 2008 zařazen Radou pro výzkum, vývoj a inovace vlády ČR na Seznam recenzovaných neimpaktovaných periodik vydávaných v České republice. Od roku 2013 je časopis PÚ vydáván pouze v elektronické formě (v roce 2014 bylo přiděleno nové ISSN 2336-4777). V souvislosti se změnou v elektronickou formu vydávání v roce 2013 časopis nedopatřením vypadl z tohoto Seznamu. Od roku 2015 je elektronická forma Pohybového ústrojí opět na Seznamu recenzovaných neimpaktovaných periodik.

Od roku 2016 vydáváme v časopisu PÚ příspěvky přijaté po recenzi v chronologickém pořadí jako číslo 1 a 2, dále dvě samostatná Supplementa s příspěvky ze symposií Kubátovy dny a Prague-Lublin-Sydney-St. Petersburg Symposium.

Nedostatek příspěvků je příčinou zpožděného vydání i tohoto čísla 2, časopisu PÚ, 28, 2021. Koncem roku 2021 v době Omikronové vlny koronavirové pandemie jsme opožděně vydali číslo 1 časopisu PÚ, 28, 2021.

Současně s konáním tradičních sympozií v roce 2021 byla vydána Supplementa 1 a 2 časopisu PÚ, 28, 2021 s abstrakty. Při příležitosti 26. Kubátových dnů (téma Ortopedická protetika – mezioborová spolupráce 5 – adaptace, online přenos 6. března 2021 z Lékařského domu v Praze 2) za účasti pana prof. MUDr. Štěpána Svačiny, DrSc. (předseda České lékařské společnosti J.E.Purkyně z.s.) byla předána ocenění jubilantům paní prof. MUDr. Janě Pařízkové, DrSc. – 90 let, panu prof. Ing. Janovi Čulíkovi, DrSc. – 85 let, panu prof. Ing. Miroslavovi Petrtýlovi, DrSc. – 80 let, panu prof. MUDr. Václavovi Smrčkovi, CSc. – 70 let, paní RNDr. Daniele Zemkové, CSc. – 70 let, panu MUDr. Petrovi Krawczykovi – 55 let, panu odb. asist. MUDr. Josefovi Krausovi, CSc. – 70 let a panu Ing. Pavlovi Černému, Ph.D. – 60 let. Odborné životopisy jubilantů jsou publikovány v Supplementu 1 časopisu PÚ, 28, 2021. V Supplementu 2 časopisu PÚ, 28, 2021 byla uveřejněna nejen abstrakta z The 23rd Prague-Lublin-Sydney-St. Petersburg Symposium (topic: Locomotor Apparatus Adaptation II – Interdisciplinary Aspects, November 20, 2021 Medical House, Sokolská 31, Prague, Czech Republic), ale i odborné životopisy zahraničních kolegů ortopedů – profesora Dr. Hanse Zwippa (Dresden, Germany) a Dr. Pieta van Loona (Deventer, Netherlands), kteří byli oceněni Čestnou medailí České lékařské společnosti J.E.Purkyně z.s. předanou panem prof. MUDr. Štěpánem Svačinou, DrSc., předsedou České lékařské společnosti J.E. Purkyně. Toto

tradiční Symposium se konalo prezenční a online formou s aktivní online účastí našich kolegů z Holandska (Dr. Piet van Loon) a Lublinu (Professor Tomasz Karski, MD, PhD a ass. Professor Jacek Karski, PhD).

Posláním časopisu PÚ je uveřejňovat vědecké práce zabývající se diagnostikou a mezioborovým léčením genetických kostních chorob, vrozených defektů končetin, sekundární osteoporózy, osteo/spondyloartrózy, ale i jiných chorob, které ve svých důsledcích negativně ovlivňují růst, vývoj a kvalitu pohybové ústrojí v průběhu lidského života. Ceněny jsou práce vycházející z výzkumu pojivových tkání na všech úrovních poznání, práce orientované na biochemickou, morfologickou, genetickou a molekulární diagnostiku chorob pohybového ústrojí. Zvláštní pozornost je přikládána pracím z oblasti ortopedické a antropologické biomechaniky, neuroadaptačním změnám skeletu v období růstu, řízené remodelaci pojivových tkání, studiím muskuloskeletálních a neuronálních interakcí v závislosti na léčebných metodách (kalciotropní léky, rehabilitace, ortoticko-protetické a operační léčení) a v neposlední řadě sdělením antropologickým a paleopatologickým. Oceňujeme především interdisciplinárně zaměřené práce. V anglickém jazyce jsou publikována sdělení zahraničních i našich autorů. Žádaným doplněním obsahu časopisu jsou zprávy ze sjezdů a konferencí. V rubrice zprávy zveřejňujeme oznámení o životním výročí členů RR časopisu, SPT ČLS JEP z.s., OPS ČLS JEP z.s. a významných osobností, sdělení o prioritních pozorováních, ze studijních a poznávacích cest aj.

V každém ročníku najdete směrnice pro autory příspěvků, kterým věnujte prosím pozornost při tvorbě Vašich vědeckých sdělení. Souhrny prací publikovaných v časopisu jsou excerpovány v EMBASE / Excerpta Medica (od r. 1994) a v Bibliographia medica Čechoslovaca (od r. 2010).

K prosazení časopisu Pohybové ústrojí mezinárodně přispívá citovat práce publikované v našem časopisu v příspěvcích posílaných do zahraničních impaktovaných časopisů. Pro zvýšení mezinárodního zájmu o časopis PÚ je žádoucí získávat původní kvalitní práce a kazuistiky v angličtině. Souhrny všech prací doporučujeme psát co nejvýstižněji, strukturovaně, česky a anglicky (objectives, methods, results and discussion), s klíčovými slovy.

Těšíme se na Vaši spolupráci a tvůrčí připomínky v roce 2022.

Redakční rada

A WORD TO READERS

Dear readers, authors and advertisers!

Thank you for your indispensable help in the creation of the interdisciplinary peer-reviewed journal *Locomotor System – Advances in Research, Diagnosis and Therapy* (journal LS).

All issues of the journal (including the Supplement) published since 1997 can be found in PDF format on the web domain of the Society for Connective Tissues of the Czech Medical Association J.E. Purkynje http://www.pojivo.cz/cz/ pohybove-ustroji/ (free access).

In 2008, the journal was included by the Council for Research, Development and Innovation of the Government of the Czech Republic in the List of peer-reviewed non-impacted periodicals published in the Czech Republic. Since 2013, the journal has been published only in electronic form (in 2014, a new ISSN 2336-4777 was assigned). In connection with the change to electronic publication in 2013, the journal inadvertently dropped from this List. Since 2015, the electronic form of the journal Locomotor System is again on the List of peer-reviewed non-impacted journals.

Since 2016 we have been publishing in the journal LS the papers accepted after review in chronological order as issues 1 and 2, as well as two separate Supplements with papers from the symposia Kubát´s days and Prague-Lublin-Sydney-St. Petersburg Symposium.

The lack of submissions is the reason for the delay in the publication of this issue 2, of the journal LS, 28, 2021. At the end of 2021 during the Omicron wave of the coronavirus pandemic, we belatedly published issue 1 of the journal LS, 28, 2021

Supplements 1 and 2 of the journal LS, 28, 2021 with abstracts were published simultaneously with the traditional symposia. On the occasion of the 26th Kubát Days (the topic of Orthopaedic Prosthetics - Interdisciplinary Cooperation 5 – Adaptation, online transmission on March 6, 2021 from the Medical House in Prague 2) with the participation of Prof. Štěpán Svačina, MD, DrSc. (President of the Czech Medical Association J.E.Purkyně z.s.), awards were presented to the jubilarians, Professor Jana Pařízková, M.D., DrSc. – 90 years , to Professor Ing. Jan Čulík, DrSc. – 85 years, Professor Ing. Miroslav Petrtýl, DrSc. – 80 years, Professor Václav Smrčka, M.D., Ph.D. – 70 years, RNDr. Daniela Zemková, Ph.D. – 70 years, Mr. Petr Krawczyk M.D. – 55 years, Professor Asst. Josef Kraus, M.D., Ph.D. – 70 years and Ing. Pavel Černý, Ph.D. – 60 years. The professional biographies of the jubilarians are published in Supplement 1 of the journal LS, 28, 2021.

In Supplement 2 of the journal LS, 28, 2021 were published not only the abstracts from The 23rd Prague-Lublin-Sydney-St. Petersburg Symposium (topic Locomotor Apparatus Adaptation II – Interdisciplinary Aspects, November 20, 2021, Medical House, Sokolská 31, Prague, Czech Republic), but also the professional biographies of our foreign orthopaedic colleagues Professor Dr. Hans Zwipp (Dresden, Germany) and Dr. Piet van Loon (Deventer, Netherlands), who were awarded the Honorary Medal of the Czech Medical Society J.E.Purkyně z.s. presented by Prof. MUDr. Štěpán Svačina, DrSc., President of the Czech Medical Society J.E. Purkyně. This traditional

Symposium was held in a face-to-face and online format with active online participation of our colleagues from the Netherlands (Dr. Piet van Loon) and Lublin (Professor Tomasz Karski, MD, PhD and Ass. Professor Jacek Karski, PhD).

The mission of the journal is to publish scientific papers dealing with the diagnosis and interdisciplinary treatment of genetic bone diseases, congenital defects of the limbs, secondary osteoporosis, osteo/spondyloarthritis, as well as other diseases that adversely affect the growth, development and quality of the musculoskeletal system during human life. Works based on research on connective tissues at all levels of knowledge, works oriented on biochemical, morphological, genetic and molecular diagnostics of musculoskeletal diseases are valued.

Particular attention is paid to works in the field of orthopaedic and anthropological biomechanics, neuroadaptive changes of the skeleton during the growth period, controlled remodelling of connective tissues, studies of musculoskeletal and neuronal interactions in relation to therapeutic methods (calciotropic drugs, rehabilitation, orthotic-prosthetic and surgical treatment) and, last but not least, anthropological and paleopathological communications. We particularly appreciate the interdisciplinary work. Communications by foreign and national authors are published in English. Reports from congresses and conferences are a welcome addition to the content of the journal. In the news section, we publish announcements of life anniversaries of members of the editorial board of the journal, Society For Connective Tissues CMA J.E. Purkynje & Society for Prosthetics and Orthotics CMA J.E. Purkynje and important personalities, announcements of priority observations, study and discovery trips, etc.

In each edition, you will find guidelines for authors of papers, which please pay attention to when drafting your scientific communications. Summaries of papers published in the journal are excerpted in EMBASE / Excerpta Medica (since 1994) and in Bibliographia medica Čechoslovaca (since 2010).

The citation of papers published in our journal in papers sent to foreign impacted journals contributes to the promotion of the journal Locomotor System internationally. In order to increase the international interest in the journal of Locomotor System, it is desirable to obtain original high quality papers and case reports in English. Abstracts of all papers are recommended to be written as concisely as possible, structured, in Czech and English (objectives, methods, results and discussion), with key words.

We look forward to your cooperation and creative comments in 2022.

Editorial Board

SOCIETY FOR CONNECTIVE TISSUES J.E.Purkyně PRAGUE • CZECH REPUBLIC

CZECH SOCIETY FOR PROSTHETICS AND ORTHOTICKS J.E. PURKYNĚ PRAHA • CZECH REPUBLIC

CZECH MEDICAL ASSOCIATION J.E. PURKYNĚ

AMBULANT CENTRE for defects of locomotor apparatus limited liability company

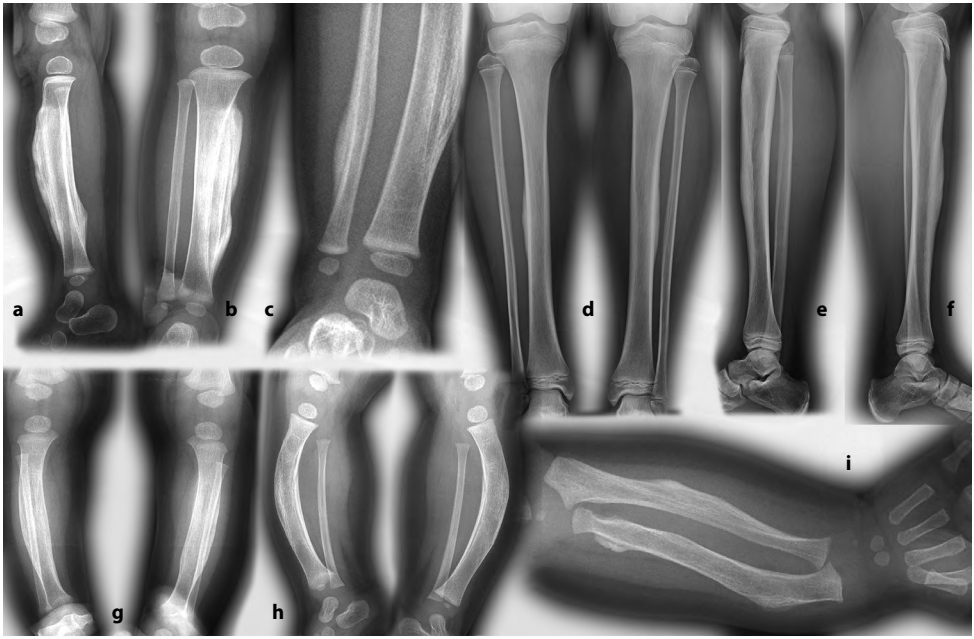
OBRÁZEK NA TITULNÍ STRANĚ ČASOPISU: INFANTILNÍ KORTIKÁLNÍ HYPEROSTÓZA

Obrázek na titulní straně časopisu demonstruje rentgenologicky ověřené projevy infantilní kortikální hyperostózy. O infantilní kortikální hyperostóze jako první referoval G. Roske v roce 1930. V literatuře se užívají synonyma Roskeho-Caffeyova, Caffeyova Silvermanova a deTony-Caffeyova choroba.

Podle *„Nosologie a klasifikace genetických kostních chorob: revise 2019"* je Caffeyova nemoc klasifikována ve 22. skupině Neonatální osteosklerotické dysplazie (OMIM No. 114000, AD/AR dědičnost, gene *COL1A1*, (609577)(609577) ORPHANET code 1310).

Caffeyova choroba (CD) je osteosklerotická dysplazie charakterizovaná akutním zánětem s masivní subperiostální tvorbou nové kosti (viz **obr. 1 a 2**), která obvykle postihuje čelist, lopatky, klíční kosti a diafýzy dlouhých kostí horních a dolních končetin. Vyskytuje se nejčastěji během prvních šesti měsíců života. Prenatálně lze skeletální změny zjistit ultrazvukem.

Obrázek na titulní straně je složen ze snímků abnormálních radiologických skeletálních znaků: Typickým nálezem je ztluštění periostu postižené kosti, které může připomínat nádorový proces. Na dlouhých kostech se proces manifestuje ztluštěním v oblasti diafýz.

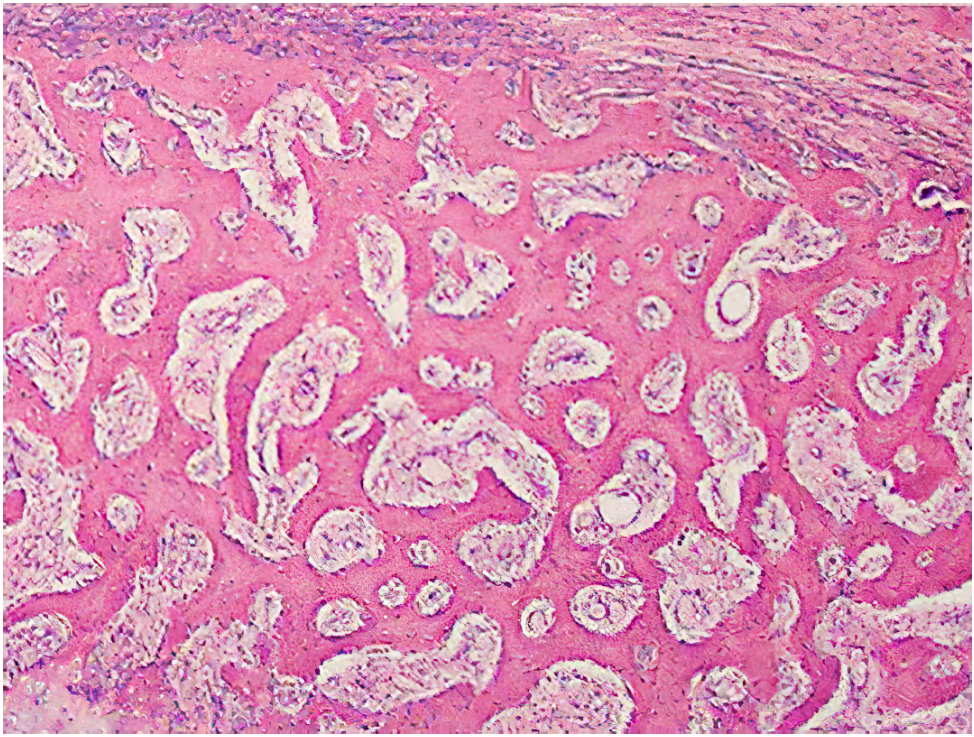


Obr. 1.

V horní polovině obrázku zleva doprava jsou snímky bérců dítěte ve věku 8 měsíců – na diafýze pravé tibie anteromediálně je excesivní subperiostální kortikální ztluštění (1a, b), 16 měs. – subperiostální kostní apozice diafýzy P tibie mediálně a fibuly v distální třetině laterálně (1c) a ve věku 10,5 roku, kdy je na obou tibiích ve střední třetině diafýzy anterolaterální kortikální ztluštění (1d, e, f).

V dolní polovině obrázku zleva doprava jsou snímky obou bérců a pravého předloktí jiného dítěte ve věku 3,5 měs. – je zde nadměrná subperiostální tvorba kortikální kosti diafýz obou tibií, které jsou ventrálně zakřiveny, dřeňový kanál je rozšířen (1g, h). Diafýzy ulny a radia také obklopuje nadměrné množství novotvořené kortikální kosti (1i).

Použity kopie RTG snímků z archivu Ambulantního centra pro vady pohybového aparátu s.r.o., Olšanská 7, 130 00 Praha 3.



Obr. 2. Histologie. V probatorní excizi z oblasti ztluštělého kortexu diafýzy tibie pacienta s Caffeyovou chorobou byly zaznamenány změny charakteru hyperostózy, tvořené trabekulami nově vytvořené kosti s řídkou kolagenní tkání mezi nimi. Barveno HE. S laskavým svolením profesora Ctibora Povýšila, MD, DrSc.

Hlavními **klinickými příznaky** jsou celková podrážděnost, horečka, anorexie, bolestivé otoky měkkých tkání někdy s pseudoparalýzou postižené končetiny. V případech s protrahovaným průběhem se pozoruje zakřivení a/nebo zvětšení délky postižených dlouhých kostí. V těžkých případech je popisován trpasličí vzrůst s krátkými končetinami a perinatální úmrtí; může být přidružen polyhydramnion. Postižení skeletu je charakterizováno nadměrnou tvorbou nové kosti – *hyperostózou* (viz **obr**. **2**).

Hlavní **radiografické znaky**. Kortikální hyperostóza postihuje nejčastěji dolní čelist, klíční kosti, lopatky, žebra a dlouhé kosti.

U vleklých případů lze pozorovat resorpci původní kortikální kosti s rozšířením dřeňového kanálu, diafyzární expanzi a zakřivení a podélné přerůstání. Mezi sousedními kostmi se mohou vytvořit kostní můstky. U těžkých prenatálních forem bývají dlouhé kosti krátké a zakřivené.

Genetický přenos je autozomálně dominantní, ale byla zaznamenána i autozomálně recesivní dědičnost.

Molekulární podstata a patogeneze. U pacientů s prenatální a postnatální formou byla zjištěna chybná mutace v exonu 41 genu kódujícího alfa 1(I) řetězec kolagenu typu I (*COL1A1*). U jiných pacientů s těžkými prenatálními projevy nebyly mutace *COL1A1* zjištěny.

Průběh a prognóza. Těžké, antenatálně postižené případy se mohou narodit mrtvé nebo zemřít při porodu v důsledku respiračního selhání. V akutní fázi onemocnění bývá horečka, dráždivost, lokalizované otoky. Laboratorně se zjišťuje zvýšená sedimentace a CRP, leukocytóza a trombocytóza, anémie. U těžkých forem bývá zvýšení alkalické fosfatázy v krevním séru. Ve většině případů můžeme očekávat úplné klinické a radiografické uzdravení během několika týdnů nebo měsíců po akutním začátku. V některých případech je průběh vleklý s opakovanými remisemi a exacerbacemi, které mohou trvat až do dospívání. U dospělých může být přítomna ochablost kloubů, kýly a zvýšené riziko zlomenin kostí.

Léčba. Intenzivní péče o kriticky nemocné novorozence. Kortikosteroidy účinně zastavují proces během akutní fáze. Nesteroidní protizánětlivé léky poskytují symptomatickou úlevu.

Diferenciální diagnostika. Spranger et al. (2018) uvádějí dobře známé nozologické jednotky, např. osteogenesis imperfecta, osteoektázie s hyperfosfatázií, Camuratiho-Engelmannova choroba, Pachydermoperiostóza, Syndrom týraného dítěte, Hyperostóza-hyperfosfátémie, Goldbloomův syndrom a Sekundární hyperostóza doprovázející infekce (vč. syfilis), kurděje, křivici, hypervitaminózu A, podávání prostaglandinu E, chronické plicní nebo srdeční onemocnění a leukémii. Tyto kostní choroby a poruchy se rozlišují podle anamnézy, věku prvních příznaků, průběhu onemocnění, přidružených klinických, laboratorních, molekulárně genetických a radiografických projevů.

THE FIGURE ON THE TITLE PAGE OF THE JOURNAL: INFANTILE CORTICAL HYPEROSTOSIS

The figure on the title page of the journal demonstrates the abnormal radiological manifestations typical for the diagnosis of Infantile Cortical Hyperostosis. Infantile cortical hyperostosis was first reported by G. Roske in 1930. The synonymous names Roske-Caffey, Caffey-Silverman and deTony-Caffey disease are used in the literature.

According to the "Nosology and Classification of Genetic Bone Diseases: 2019 Revision", Caffey's disease is classified in Group 22 Neonatal Osteosclerotic Dysplasia (OMIM No. 114000, AD/AR inheritance, *COL1A1* gene, ORPHANET code 1310).

Caffey disease (CD) is an osteosclerotic dysplasia characterized by acute inflammation with massive subperiosteal new bone formation (**see figure 1 and 2**) usually involving the jaw, shoulder blades, collar bones, and shafts of long bones in the arms and legs. It occurs most often within the first six months of life. Prenatally skeletal changes may be detected by ultrasound.

The picture is composed from films of abnormal radiological skeletal features: A typical finding is thickening of the periosteum of the affected bone, which may resemble a tumour process. On long bones, the process manifests itself by thickening in the diaphyseal region.

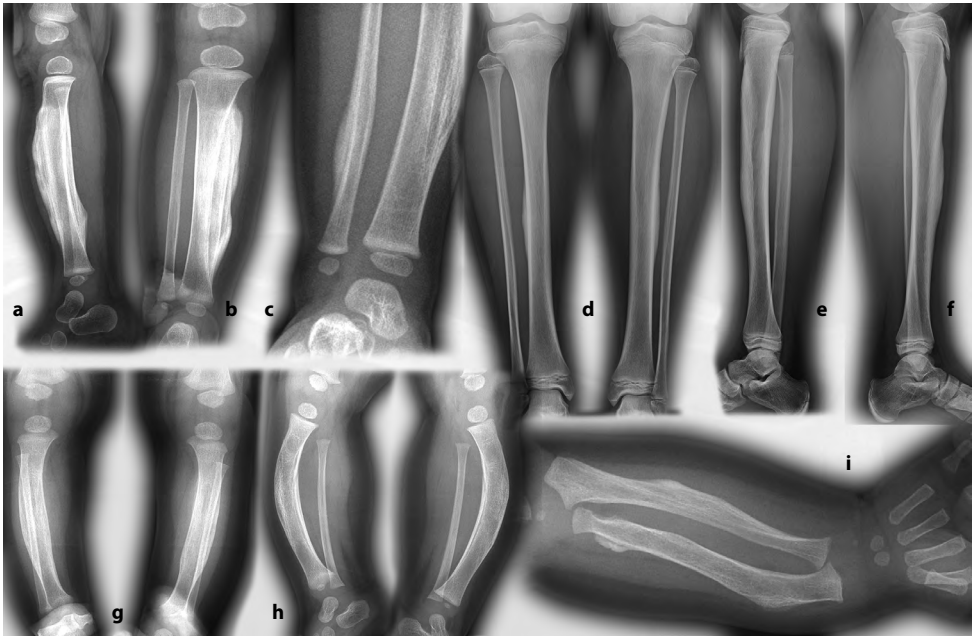


Figure 1.

In the upper half of the picture, from left to right, are the images of the tibiae of the child at the age of 8 months – there is an exaggerated subperiosteal cortical thickening on the diffusion of the right tibia anteromedially (1a, b), on the image at 16 months – subperiosteal bony apposition of the diaphysis of the P tibia medially and the fibula in the distal third laterally (1c), and at 10.5 years of age, when there is anterolateral cortical thickening on both tibiae in the medial third of the diaphysis (1d, e, f).

In the lower half of the image from left to right are images of both tibias and the right forearm of another child at 3.5 months of age – there is excessive subperiosteal cortical bone formation of the diaphyses of both tibias, which are curved ventrally, and the intramedullary canal is dilated (1g, h). The diaphyses of the ulna and radius are also surrounded by an excessive amount of newly formed cortical bone (1i).

Copies of radiographs from the archive of the Centre for Defects of Locomotor apparatus, Olšanská 7, 130 00 Praha 3 were used.

The **main clinical signs** are general irritability, fever, anorexia; painful soft tissue swelling sometimes with pseudoparalysis of the affected limb. In cases with a protracted course, curvature and/or increase in length of the affected long bones is observed. In severe cases, dwarfism with short limbs and perinatal death are described; polyhydramnios may be associated. Skeletal involvement is characterized by excessive new bone formation – hyperostosis (see **Figure 2**).

Main radiographic features. Cortical hyperostosis most commonly affects the mandible, clavicles, scapulae, ribs and long bones.

In protracted cases, resorption of the original cortical bone with enlargement of the medullary canal, diaphyseal expansion and curvature, and longitudinal overgrowth can be observed. Bone bridges may form between adjacent bones. In severe prenatal forms, the long bones tend to be short and curved.

Genetic transmission is autosomal dominant, but autosomal recessive inheritance has also been reported.

Molecular basis and pathogenesis. A missense mutation in exon 41 of the gene encoding the alpha 1(I) chain of type I collagen (*COL1A1*) has been identified in patients with prenatal and postnatal forms. In other patients with severe prenatal manifestations, *COL1A1* mutations were not detected.

Course and prognosis. Severe, antenatally affected cases may be stillborn or die at birth due to respiratory failure. In the acute phase of the disease there is fever, increased sedimentation rate and C-reactive protein, leukocytosis and thrombocytosis, anaemia. In severe forms, there is an increase of serum alkaline phosphatase. In most cases, complete clinical and radiographic recovery can be expected within weeks or months after acute onset. In some cases, the course is protracted with

repeated remissions and exacerbations that may last into adolescence. Joint laxity, hernias and increased risk of bone fractures may be present in adults.

Treatment. Intensive care of critically ill newborns. Corticosteroids effectively stop the process during the acute phase. Non-steroidal anti-inflammatory drugs provide symptomatic relief.

Differential diagnosis. Spranger et al. (2018) list well-known nosological entities, e.g., osteogenesis imperfecta, osteoectasia with hyperphosphatemia, Camurati-Engelmann disease, Pachydermoperiostosis, Abused Child Syndrome, Hyperostosis-hyperphosphatemia, Goldbloom syndrome, and Secondary Hyperostosis accompanying infections (incl. syphilis), scurvy, rickets, hypervitaminosis A, prostaglandin E administration, chronic lung or heart disease and leukaemia. These bone diseases and disorders are classified according to history, age of first symptoms, course of disease, and associated clinical, laboratory, molecular genetic, and radiographic manifestations.

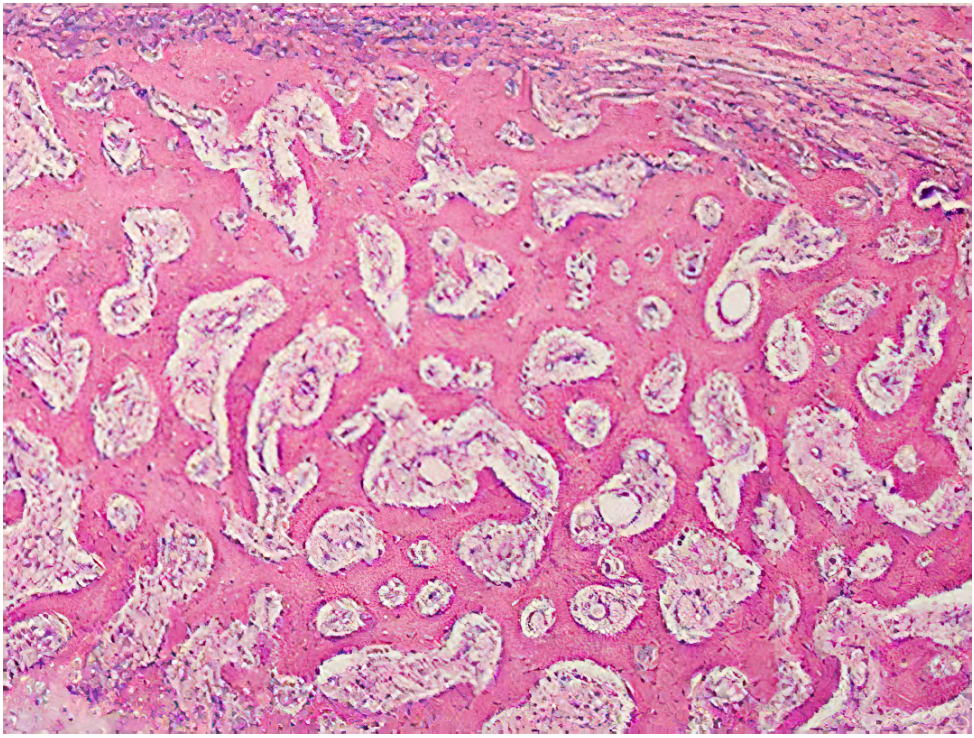


Figure 2. Histology. In a probatorial excision from the region of thickened cortex of the diaphyseal tibia from a patient with Caffey's disease, changes of the character of hyperostosis were noted, consisting of trabeculae of newly formed bone with sparse collagenous tissue between them. Coloured by HE. Courtesy to Professor Ctibor Povýšil, MD, DSc.

References

CAFFEY J, SILVERMAN W. (1945) Infantile cortical hyperostosis, preliminary report on new syndrome. Am. J Roentgen 54; p. 1–16.
DEJONG G, MULLER MM (1995) Perinatal death in Caffey disease. Am J Med Genet 59; p. 134–138.
KAMOUN-GOLDRAT A, MARTINOVIC J, SAADA J, et al. Prenatal cortical hyperostosis with *COL1A1* gene mutation. Am J Med Genet 146A; p. 1820–1824.
MAŘÍK I, MAŘÍKOVÁ A, POVÝŠIL C. Kostní genetické choroby. In: Ctibor Povýšil et al. Patomorfologie chorob kostí a kloubů, Galén, 2017: p. 84.
MORTIER GR, COHN DH, CORMIER-DAIRE V, HALL Ch, et al. Nosology and classification of genetic skeletal disorders: 2019 revision. Am J Med Genet Part A. 2019; 1–27. https://doi.org/10.1002/ajmg.a.61366
ROSKE U (1930) Über eine eigenartige Knochenerkrankung im Säuglingsalter. Mschr Kinderheilk 47; p. 385–400.
SPRANGER JW, BRILL PW, HALL CH, NISHIMURA G, SUPERTI-FURGA A, UNGER S. Bone Dysplasias An Atlas of Genetic Disorders of Skeletal Development. 4th ed. Oxford University Press, 2018: 623–626.

Professor Ivo Marik, MD, PhD & Radek Myslivec, MD
Centre for Defects of Locomotor Apparatus l.l.c.
Olšanská 7, 130 00 Prague, Czech Republic
E-mail: ambul_centrum@volny.cz

KLASIFIKACE NÁSLEDKŮ KOMPARTMENT SYNDROMU (CS) NEBO POSTISCHEMICKÉHO SYNDROMU (PIS) DOLNÍ KONČETINY A NOHY U DOSPĚLÝCH, ADOLESCENTŮ A DĚTÍ

CLASSIFICATION OF SEQUELAE OF COMPARTMENT SYNDROME (CS) OR POSTISCHEMIC SYNDROME (PIS) OF THE LOWER LEG AND FOOT IN ADULTS, ADOLESCENTS AND CHILDREN

ZWIPP HANS, Emeritus

Center of Orthopedics and Traumatology of the University Hospital Carl Gustav Carus at the Technical University of Dresden, Germany

ABSTRACT

The sequelae of an undiagnosed, an insufficiently treated or unpreventable (by crush injury) compartment or postischemic syndrome, most often after lower leg fracture or popliteal artery rupture, are caused by necrosis and contracture of the extrinsic foot muscles. The more harmless hammer toes due to contracture of the intrinsic muscles follow most often an untreated isolated foot CS induced e. g. by a calcaneal fracture. In combined CS of the lower leg and the foot or in some isolated cases of mainly one involved muscle we see different types of deformity (Types 1–5). The different types are also altered by involvement of nerve damages. Due to the involved and the amount of scarred muscles we observe flexible or contract hammer toes, claw toes, hallux flexus, hallux valgus, foot equinus or the severest form of a postcompartment or postischemic pes equinovarus. Less common (16 of 66 cases) in our seria of corrections were the deformities caused by an isolated CS, such as necrosis of the anterior tibialis, long extensor or the peroneal muscles. Most surprisingly have been few cases of a functional hallux flexus or a severe hallux valgus in a boy.

Keywords: lower leg and foot compartment syndrome – postischemic syndrome – hammertoes and claw toes – hallux valgus – pes equinovarus

INTRODUCTION

There are many reports about acute CS or PIS how to detect and how to treat, but only few about isolated and/or combined sequelae due to neglected acute CS or PIS of the lower leg which terms are often used synonymously. It was Sir Henry John Seddon [**11**] who had introduced the worldwide classification of nerve damage and who described in 1966 very precisely the typical foot and ankle deformity when the deep flexor compartment of the lower leg was involved by fracture or popliteal artery rupture producing a contract pes equinovarus. Others reported [**3–10, 12–15**] after Seddon about different deformities as sequelae of CS or PIS of the lower leg. In the international literature is reported about different operative corrections of sequelae due to insufficiently treated or overlooked CS/PIS of the lower leg including below knee amputation as a salvage [**4**]. But no classification related to different sequelae after CS or PIS was published until 2008, but written in German [**14**]. Therefore it is not well known and presented here again. Additionally to a previous report in 1994 [**15**] of a hallux valgus deformity caused by CS a second case in a child is presented

Definitions

A. Sequelae of **Compartment Syndrome (CS)** are caused by posttraumatic muscle necrosis and/or fibrosis due to an unsufficiently treated acute compartment syndrome (too high pressure in a closed compartment) producing different deformities depending on which extrinsic or intrinsic muscles of the lower leg and/or foot have had infarction
B. Sequelae of **Postischemic Syndrome (PIS)** are caused by too late arterial repair and/or unreleased compartments after arterial repair (ischaemia > 6h needs principally splitting of all four compartments of the lower leg [**10**] producing same or even stronger deformities as CS
C. Both groups (A, B) often are combined with nerve damages according to different types of Seddon's classification

Epidemiology

Lower leg compartment syndrome is seen in 17% of all lower leg fractures [**2**]. Urgent release of all 4 lower lower leg compartments was necessary in 75% of all cases of a popliteal artery rupture, but only in 6% due to a tibial head fracture [**10**].The likelihood therefore to develop a posttraumatic pes equinovarus seems to be much more higher after too late or insufficient splitting of the deep flexor compartment of the lower leg caused by popliteal artery rupture and repair than by a fracture of the tibial shaft. Regarding all different compartment syndromes of the skeleton the lower leg CS is seen most often in 72.7 %, whereas the isolated foot CS equals only 3.3 % [**1**]. The less frequent combined CS of the lower leg together with the foot (38% of foot CS) is as much caused by arterial injury as by fracture [**12**]. Acute foot compartment syndrome was reported in 21 patients after fractures and fracture-dislocations of Lisfranc (58%), Chopart (16%), talus (8%), calcaneus (8%), serial metatarsals (8%) and in another 8 patients as a combined lower leg and foot compartment syndrome due to vascular ruptures in 50% [**12**].

Pathology

Regarding the four lower leg compartments different deformities can be observed:

1. In case of an overlooked superficial flexor compartment syndrome a fixed equinus of the foot with mild varus results.
2. In case of insufficiently treated extensor compartment syndrome a loss of dorsiflexion of foot and toes like in palsy of the N. peroneus profundus is seen.
3. An overlooked or incorrectly released peroneal compartment syndrome causes loss of active pronation
4. The sequelae of a deep flexor compartment syndrome are due to the most often overlooked, insufficiently released or even completely unreleased deep flexor compartment of the lower leg. This leads to muscle necrosis, fibrosis and pes equinovarus with claw toes and often with numbness of the planta pedis
5. Often combinations of 1 to 4 are seen.

According to Seddon [**11**] most often infarction and necrosis is found in the deep flexor muscles. Seldomly a postraumatic pes equinovarus is caused by a N. ischiadicus palsy or a chronic infection of the lower leg after fracture [**13, 15**].

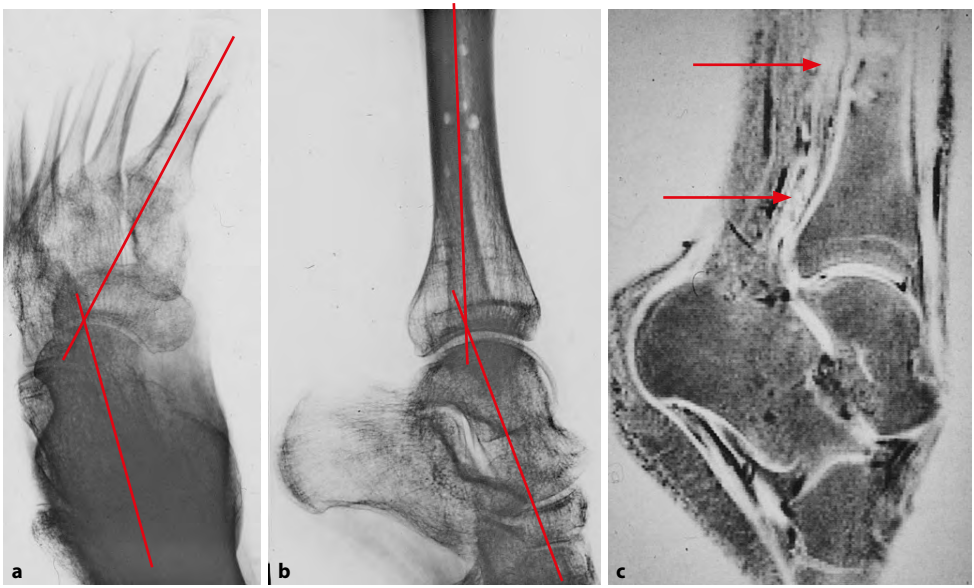


Fig. 1a–c: Diagnostic steps are shown in case of a severe Type 5 after PIS in a young man. **a, b**: shows an abnormal contract foot equinus of 60° with adduction of the forefoot of 40° **c**: CT shows necrotic areas in the posterior tibialis muscle.

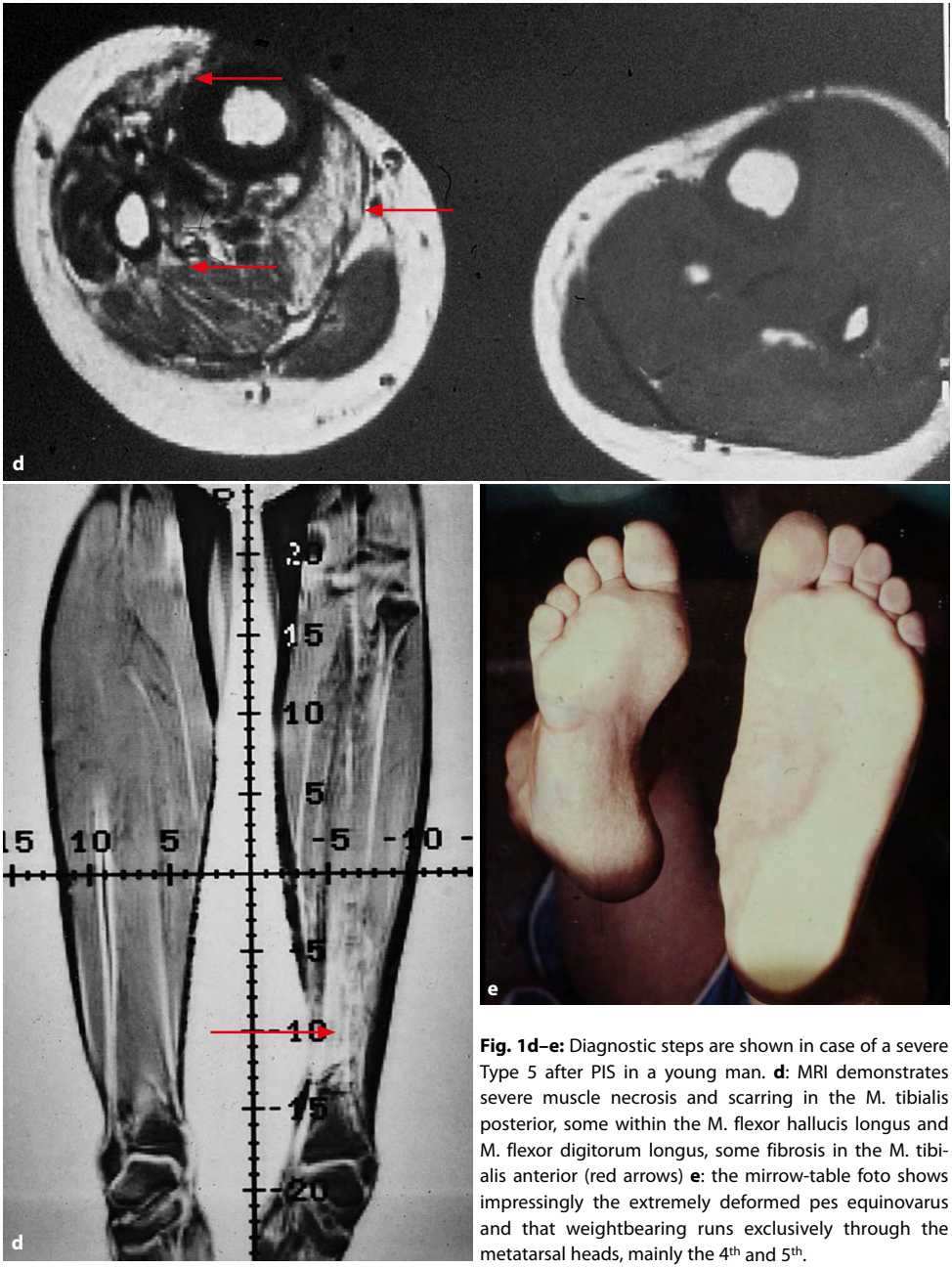


Fig. 1d–e: Diagnostic steps are shown in case of a severe Type 5 after PIS in a young man. **d**: MRI demonstrates severe muscle necrosis and scarring in the M. tibialis posterior, some within the M. flexor hallucis longus and M. flexor digitorum longus, some fibrosis in the M. tibialis anterior (red arrows) **e**: the mirrow-table foto shows impressingly the extremely deformed pes equinovarus and that weightbearing runs exclusively through the metatarsal heads, mainly the 4th and 5th.

Examination for Classification

First of all anamnesis and clinical examination are necessary including clinical tests like active knee bowing to exclude functional sequelae of CS (see example in **Fig. 3**). Standard weight bearing x-rays in two planes including a hindfoot projection are necessary to measure the amount of forefoot adduction and hindfoot varus. MRI of both lower legs and feet shows much better than CT-scanning necrosis of the involved muscles. In cases of neurological deficits a neurological examination including electromyography (EMG) and electroneurography (ENG) both sided are necessary in case of needed muscle transfer for substitution of a paralyzed muscle. Sometimes doppler-sonography or distraction-subtraction-angiography (DSA) is needed preoperatively in cases of critical blood supply (**Fig. 1**).

Patients

During the period of 1994 to 2007 a total of 66 patients were treated operatively at the Trauma Department of the University Hospital Carl Gustav Carus at the Technical University of Dresden, Germany. Isolated sequelae of the lower leg were seen ten patients (15%), isolated sequelae of the foot in 26 patients (39%), combined sequelae of the lower leg and foot in 30 patients (46%).The distribution and different corrective operations are shown in **Table 1**, the classification of 5 Types in **Table 2**.

In our series of 24 treated cases of pes equinovarus 5 cases (21 %) by a postischemic syndrome in patients all treated initially in peripheral hospitals and sent later for repair of the sequelae.

a) isolated lower leg compartment syndrome	n
M. triceps surae (percutaneous Achilles tenotomy)	2
M. tibialis anterior (M.tibialis posterior – transfer	2
M. flexor hallucis longus (open lengthening)	5
Mm. peronei (M. tibialis posterior – transfer)	1
b) isolated foot compartment syndrome	26
Mm.flexores breves (percutaneous tenotomy of D2–D5)	
c) combined lower leg and foot compartment syndrome	
M. flexor hallucis l. + M. flexor digitorum l. + Mm. flexores breves	6
(open lengthening of long flexors + percutaneous tenotomy of short flexors)	
M. triceps surae + M. tibialis posterior + M. flex. dig. l. +.M. flex. hall. l.	24
(triple 4, midfoot-fusion 2, complex soft tissue reconstruction without fusion 18)	

Table 1: Reconstructions after isolated or combined CS or PIS (n=66)

Type 1	Lesser toes	a) hammer toes
		b) claw toes
Type 2	Big toe	a) functional hallux flexus
		b) contract hallux flexus
Type 3	Lesser toes + big toe	a) contract hallux flexus + hammer/claw toes D2–D5
		b) contract hallux flexus + abductus + claw toes D2–D5
Type 4	Pes equinus	a) contract pes equinus without nerve damage
		b) palsy of M.tibialis anterior, M. extensor hallucis/digitorum longus and/or palsy of N. peroneus communis
		c) Necrosis of Mm. peronei and palsy of N. peroneus communis
Type 5	Pes equinovarus	a) + defect of N. peroneus communis (motoric + sensitive)
		b) + defect of N. tibialis (motoric + sensitive)
		c) + combined defect of N.peron.c..+ N.tib.(motoric+sensitive) + numb sole

Table 2: Classification of the sequelae of compartment or postischemic syndrome of the lower leg and foot related to different deformities caused by contractions of muscles and tendons or nerve damages [**14**]

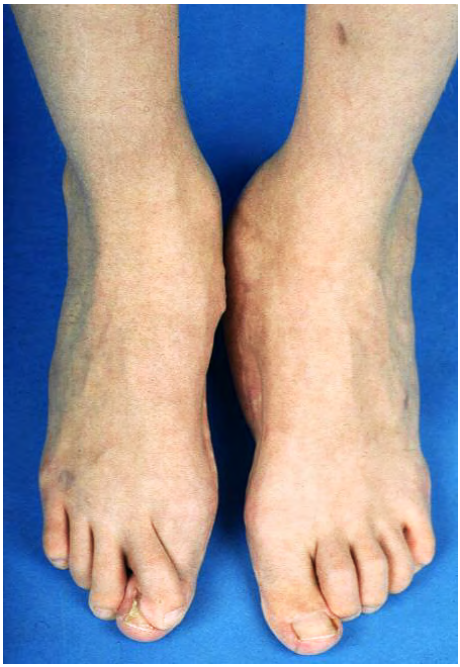


Fig. 2: Example of Type 1 a+b (left foot) and Type 3 b (right foot) as result of an untreated complex foot and ankle CS both-sided in a 14 years old boy who had sustained ten months before a severe polytrauma after fall from the fifth floor. Beneath life threatening injuries and multiple fractures he had additionally complex calcaneal fractures both-sided with a 3° degree soft tissue injury without any release. After referral to our unit ORIF of both calcaneal fractures was done three weeks after injury requiring additionally a free flap for the medial hindfoot left-sided due to severest posttraumatic skin necrosis. The boy suffers now from a painful hallux valgus right-sided, little left-sided, but as well from painful contract hammer and claw toes needing early correction.

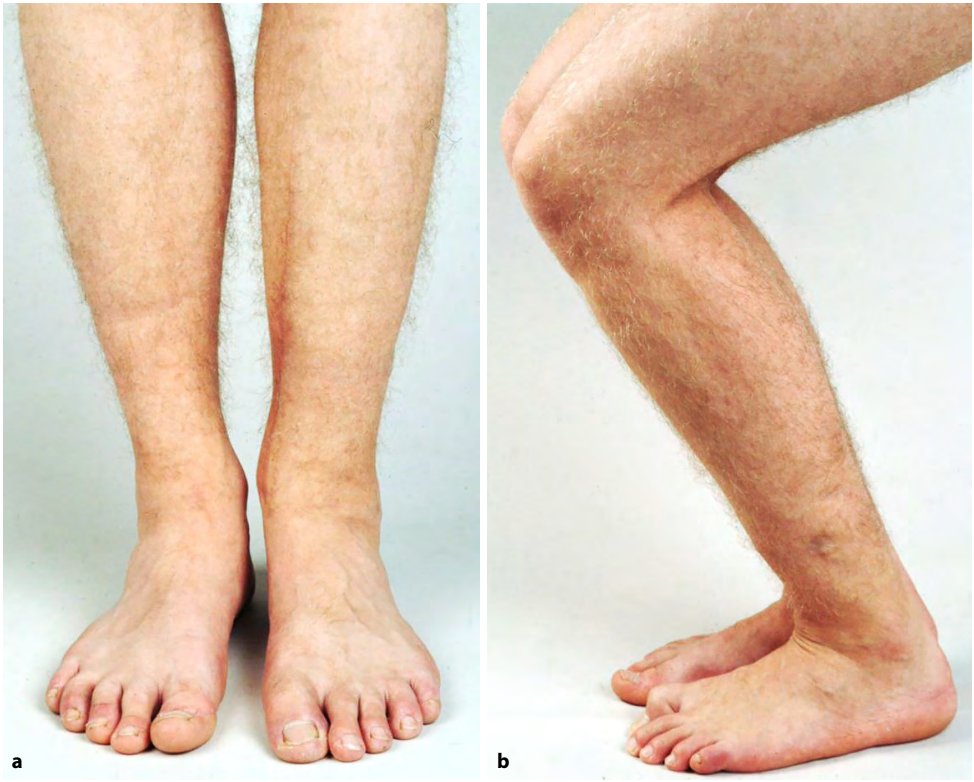


Fig. 3: Type 2 a deformity of a "functional hallux flexus" in a 25 years old plumber being unable to work with bowed knees in shoes due to significant pressure and pain **a**: while standing upright the left big toe is only very little bowed **b**: as soon as bowing knees already in 70° of knee flexion the left big toe is extremely bowed.

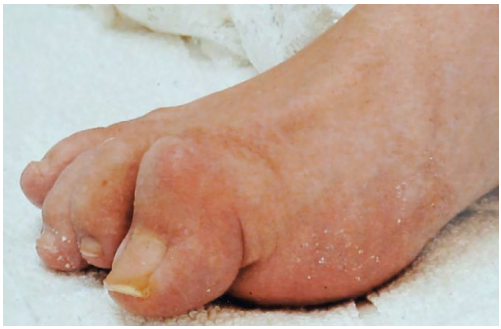


Fig. 4: Painful contract hallux flexus and contracted claw toes D2–D4, hammer toe 5 (Type 3a) two months after ACL-repair and unrecognized postoperative CS in a 22 years old female student, complaining of pain in shoes and not being able to wear high heels.

Typical examples

The most common derformity is the development of hammertoes D2 to D5 after calcaneal fracture with unrecognized or untreated CS of the foot. This is all due to contractures of the intrinsic short flexors of the toes. As soon as the extrinsic long flexors are involved by a lower leg CS we will see as sequelae contract claw toes. Depending on the amount of muscle necrosis some flexibility or rigidity of clawing of the lesser toes is seen. Depending on the amount of necrosis within the long flexor hallucis belly we can observe a flexible, a functional contract or absolutely contract hallux flexus. When necrosis occurred additional in the abductor hallucis muscle we see a hallux valgus with claw or hammer toes of the lesser ones which is caused by CS (**Fig. 2**).

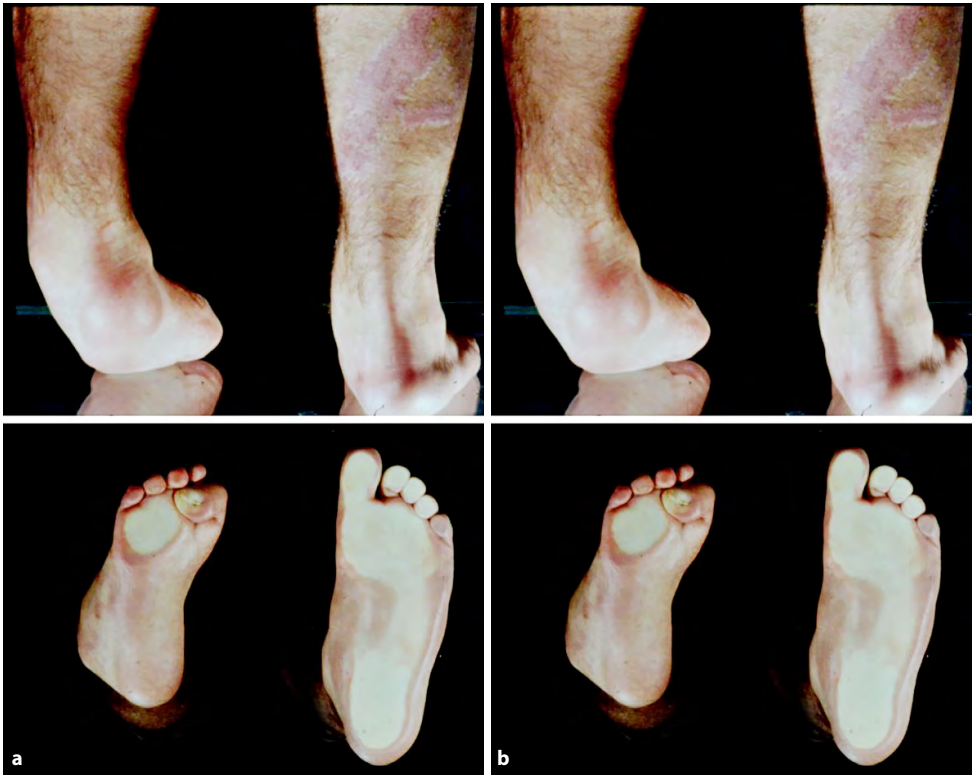


Fig. 5: This Type 5 b equals a severe pes equinovarus after PIS in a 16 years old boy caused by rupture of the popliteal artery but missed splitting of the lower leg compartments after the vascular repair. On the mirror-table one sees from behind (**a**) how flexed the foot is with a lifted heel of about 5 cm caused by contracture of the M. triceps surae, the M. tibialis posterior, the M. flexor digitorum longus and the M. flexor hallucis longus with extreme overpull of the latter one. One sees as well from in front (**b**) how strongly the heel is pulled into varus and how significantly the foot is supinated and adducted by the shortened M. tibialis posterior muscle due to necrosis and scarring in the muscle and in the extrinsic flexors as well.

Big toe deformity was observed as well in three cases as a Type 2 a deformity which equals an isolated and only partial scarring of the flexor hallucis longus muscle. This entity is only detectable by watching the big toe during active knee bowing (**Fig. 3 a, b**).

DISCUSSION

As far as seen in literature no other classification of sequelae due to CS or PIS of the lower leg, the foot or the combination of both was reported until 2008 [**14**]. The advantage of a classification is considered for having by this a base for comparing cases observed and probably treated differently for correction. Manoli et al. [**8**] recommended excision of scarred muscles after ischemic contracture following Seddon's way [**11**]. In contrast, in our series resection was not done in any case. We always had a chance by lengthening the shortened tendons to correct the malformation even in severe cases of a pes equinovarus. Furthermore we had learned by the time in in treating 24 cases of the severe Type 5 that triple arthrodesis as recommended by Hansen [**2**] was seldom necessary (4 of 24). Instead of triple fusion and resection of tendons we established an own standardized complex soft tissue procedure (n=18). Seeing by this procedure equal and reliable long term results [**9, 11**].

Literature

1. Echtermeyer V (1985) Das Kompartment-Syndrom. Hefte Unfallheilkunde 169: 1–120. Springer Heidelberg New York Tokyo
2. Echtermeyer V, Muhr G, Oestern HJ, Tscherne H (1982) Chirurgische Behandlung des Kompartment-Syndromes. Unfallheilk 85: 144–152
3. Hansen ST jr (1991) Chronic Sequelae of Leg Compartment Injury. In: Jahss. Disorders of the Foot & Ankle. Vol III, p. 2586–2590
4. 4. Karlström G, Lönnerhold T, Olerud S (1975) Cavus deformity of the foot after fracture of the tibial shaft. JBJS (Am) 57: 893–900
5. Kikuchi S, Hasue M, Watanabe M (1978) Ischemic contracture in the lower limb. Clin Orthop 134: 185-192
6. Lintz F, Colombier JA, Letenneur J, Gouin F (2009) Management of Long-Term Sequelae of Compartment Syndrome involving the Foot and Ankle. FAI 9: 847–853
7. Manoli A, Smith DG, Hansen ST jr (1993) Scarred Muscle Excision for the Treatment of Established Ischemic Contracture of the Lower Extremity. Clin Orthop Rel Res 292: 309–314
8. Matsen FA (1975) Compartmental syndromes: a unified concept. Clin Orthop 113: 8–14
9. Rammelt S, Zwipp H (2008) Reconstructive surgery after compartment syndrome of the lower leg and the foot. Eur J Trauma Emerg Surg3: 237–248
10. Scola E, Zwipp H (1991) Das Kompartmentsyndrom bei Verletzung der A. poplitea. Unfallchirurg 94: 254–256
11. Seddon HJ (1966) Volkmann`s ischaemia in the lower limb. JBJS(Br) 48: 627–636
12. Swoboda B, Scola E, Zwipp H (1991) Operative Behandlung und Spätergebnisse des Fußkompartmentsyndroms. Unfallchirurg 94: 262–266
13. Zwipp H, Sabauri G, Amlang Ml (2008) Zur chirurgischen Behandlung des Pes equinovarus als Folge eines Kompartment- und/oder Postischämie Syndroms der tiefen Flexorenloge des Unterschenkels. Unfallchirurg 111: 785–795

14. Zwipp H (2008) Wiederherstellende Chirurgie der Folgezustände nach Kompartmentsyndrom des Unterschenkels und/oder Fußes – Vorstellung einer neuen Klassifikation. Unfallchirurg 111: 776–784
15. Zwipp H (1994) Chirurgie des Fußes. p. 221.Springer, Vienna New York

Author's address:
Prof. em. Dr. med. Hans Zwipp
e-mail: hans.zwipp@t-online.de

TEST FLEXE PRSTŮ K ROZPOZNÁNÍ PATOLOGIE NOHY. KLINICKÉ PŘÍZNAKY. PROFYLAXE. TERAPIE

FLEXIONS TOES TEST TO RECOGNIZE THE PATHOLOGY OF THE FOOT. CLINICAL SYMPTOMS. PROPHYLAXIS. THERAPY

Karski Jacek[1], Karski Tomasz[2], Pyrc Jarosław[3], Slowińska Beata[4], Boryga Bartosz[4]

[1] Jacek Karski MD PhD – Medical University in Lublin, Poland, jkarski@vp.pl

[2] Prof. Tomasz Karski MD PhD – Vincent Pol University in Lublin, Poland, tmkarski@gmail.com

[3] Pyrc Jarosław MD PhD – Center of Orthopedic and Trauma Surgery Elblandkliniken, Radebeul, German, j.pyrc@web.de

[4] Mgr Slowinska Beata – Rehabilitation Department of Military Hospital in Lublin, Poland, beataslowinska2016@gmail.com

[4] Mgr Boryga Bartosz – Rehabilitation Department of Military Hospital in Lublin, Poland, b.boryga@wp.pl

ABSTRACT

The feet of infants and children go through different developmental stages during life. Sometimes we can see incorrect anatomy, sometimes various deformities – congenital, neurological, connected with "Syndrome of Contracture and Deformities" according Prof. Hans Mau and other disorders. In adults – foot deformities and pain syndromes can be a result of restricted movement of the foot joints. In the article we present the deficit of the toes flexion in metatarsal – phalangeal joint and examples of the "pain syndromes" as the result of this pathology. The problem was discovered in 1971 and from this time many cases have been observed throughout the many years of all the author's professional activities. The first publication about this problem was in the Polish Orthopedic Journal in 1971, the next in the German Orthopedic Journal in 1985.

Key words: foot anatomy, foot physiology, foot pathology, toes flexion test

INTRODUCTION

The feet – part of the human body – which are important for standing, walking, running, jumping and other activities, at work and in sports (**References 1–29**). Due to the specific anatomical structure of the bones of the feet and the full range of motion in all parts of the foot – we can function without difficulty, in every situation and in every period of our lives.

The correct and unrestricted movement of the ankle joint, the subtalar joint and the metatarsal – phalangeal joints is particularly important. The latter is very important when walking, running or jumping.

Functional anatomy of the foot

Proper foot anatomy and full, unrestricted movement at each joint of the foot is a prerequisite for normal function. Especially important is the correct range of motion at the ankle joint - dorsal flexion, in other words "extension" and plantar flexion – when walking up stairs, climbing a mountain and jumping. Any rotational movement in this joint is pathological and can cause instability, swelling and pain. We have described these disorders in articles in the USA (3 publications), India (1 publication), and Czech Republic (1 publication) in 2017–2021 (**References 19–29**).

Other, important joints in the feet are the metatarsal – phalangeal joints of all the toes. Proper of plantar flexion range of the toes is crucial for daily activities. Proper, pain free walking is only possible when the plantar flexion of the toes is unlimited. Every limitation of plantar flexion of the toes is the cause of "foot pathology" and this problem is described on many cases in this paper (**Fig. 3a, 3b, 3c, 4a, 4b, 5a, 5b, 5c, 6a, 6b, 6c, 7a, 7b**).

Toe flexion test to identify the state of foot function. The first publication about this problem was in the Polish Orthopedic Journal **(Chirurgia Narządów Ruchu i Ortopedia Polska)** in 1971 and next in the German Orthopedic Journal **(Beitrage zur Orthopädie und Traumatologie)** in 1985 (**Fig. 1**) as well as in many others publications (**References 4–26**). The foot has two important parts – tarsus – "standing part" and forefoot – "active part" – important in walking (**Fig. 2**). We have found, that "plantar flexion" of the toes in metatarsal – phalange joints is different in children, different in youth and specially in adults. (**Fig. 3a, 3b, 3c, 4a, 4b, 5a, 5b, 5c, 6a, 6b, 6c, 7a, 7b**). We observed in the following years of ours life, that the range of this movement is more and more restricted and in many patients significantly small. The range of this movement is age related – bigger in children, smaller in adults.

In the paper we inform also about two other forms of feet pathology connected with limitation of the plantar flexion of the toes. There are patients with the Friedreich syndrome and patients with the Köhler II disease. The Köhler II disease is an aseptic necrosis of the metatarsal bone head – mostly second, and mostly connected with using of improper shoes, which disturb the blood circulation in the frontal part of the feet (**Fig. 8, 9a, 9b**).

MATERIAL

In our orthopedic activity – T. Karski since 1961 and J. Karski since 1989, J. Pyrc since 1996 we have treated many thousands of people of various age – children, youth and adults with feet problems. In this paper we present examples of the cases of feet pathology connected with limited toe flexion.

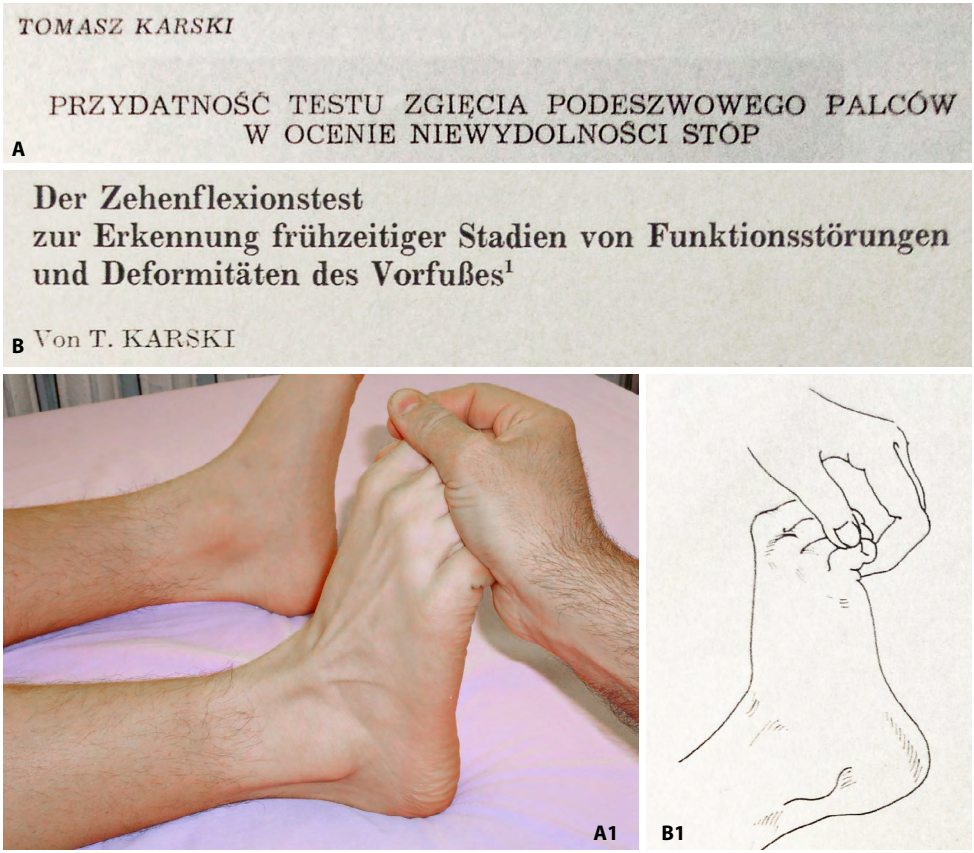


Fig. 1. History of the discovery of the 'toe flexion test'. Exercises in the therapy of 'painful feet'. (A) – publication in Poland (1971), (B) – publication in Germany (1985 – with the help of Dr F. Lettow, Neuruppin), (A1) (B1) Example of exercises performed by a physiotherapist.

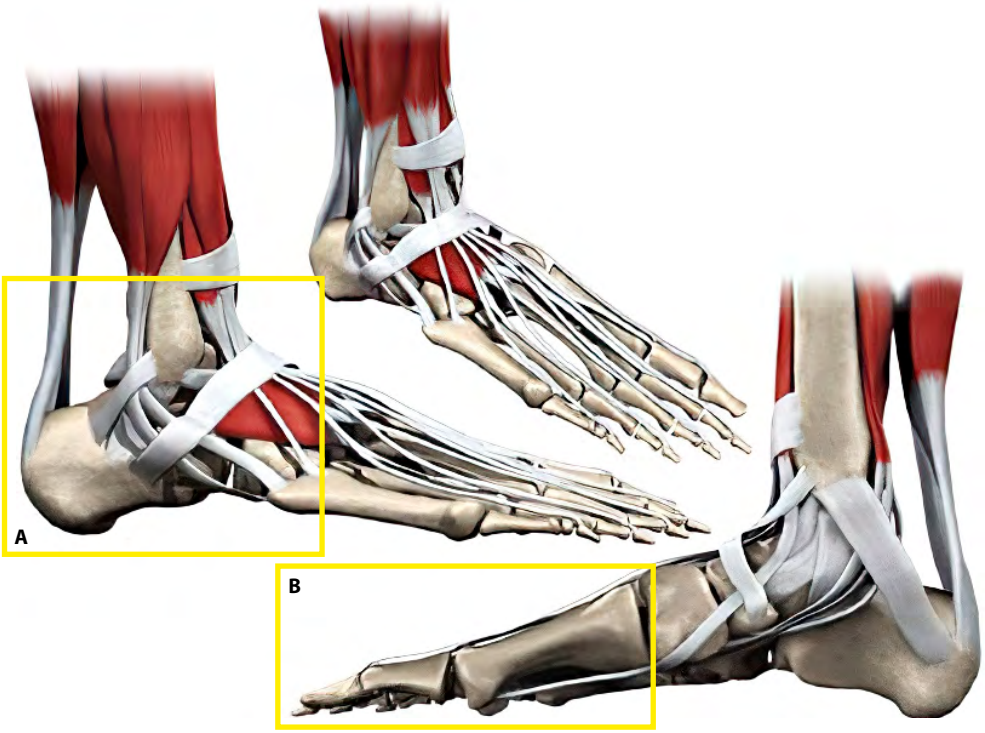


Fig. 2. Foot anatomy (picture from the Internet): 26 bones, 33 joints, over 100 muscles and tendons. Part (A) important for standing. Part (B) important for walking. Full flexion in metatarsal – phalangeal joints allow normal activity – walking, running, jumping, working, every kind of sport.

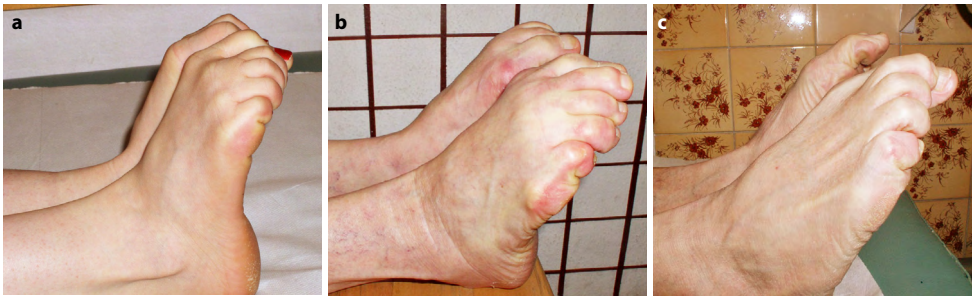


Fig. 3a, b, c.
a: Full flexion in metatarsal – toe joints. No problems with walking.
b: Limited flexion in metatarsal – toe joints. Problems with walking. Pain.
c: Maximally limited flexion in metatarsal – toe joints. Extremely big problems with walking. Pain in every step.

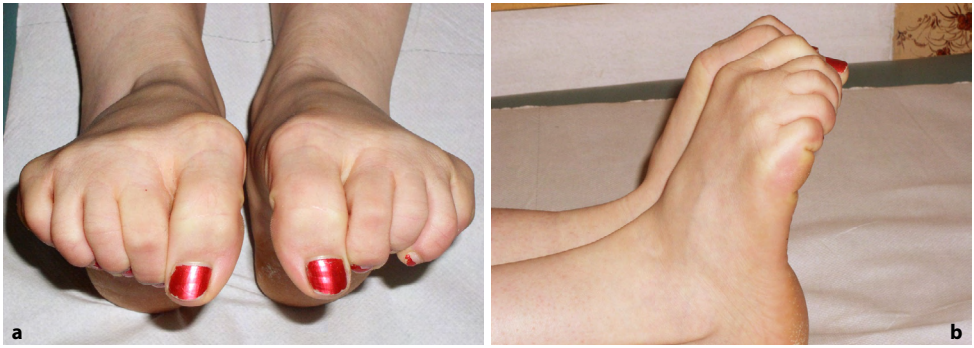


Fig. 4a, b. Female, 19. Student. Toe flexion test – fully physiological. Active flexion of toes of both feet till 50°. No problems with walking and with daily activities. No pain.

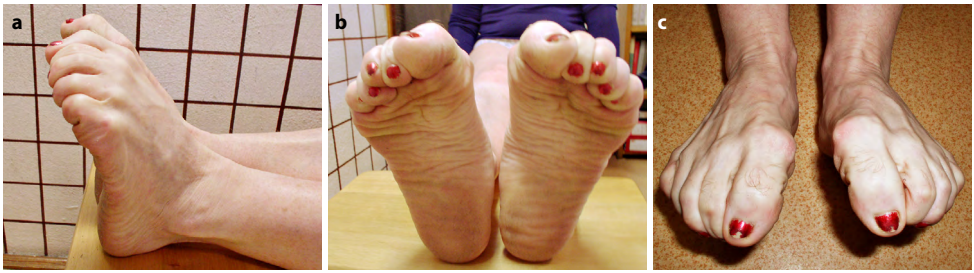


Fig. 5a, b, c. Female, 54, gardener. Problem with feet. Difficulties in walking. Pain after walking longer distances. Partially limited plantar flexion of the toes Flexion test – 30° flexion in metatarsal – phalanges joints. Additionally, valgus of knees and so-called idiopathic scoliosis in the new classification – 2A group – left convex lumbar curve. In therapy – flexion exercises for the toes.

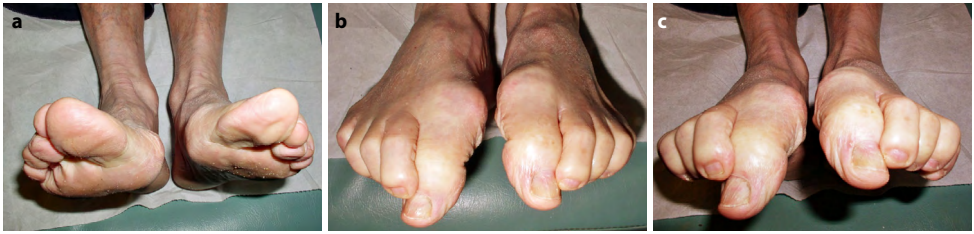


Fig. 6a, b, c. Man, 66, architecture engineer. Problem with feet. Difficulties in walking. Pain in every step. Maximally limited plantar flexion of the toes. Flexion test – maximal pathology – zero flexion in metatarsal – phalanges joints. Additionally, varus of shanks and so-called idiopathic scoliosis – in the new classification – 3rd group. Totally stiff spine. No curves. In therapy – flexion exercises for the toes.

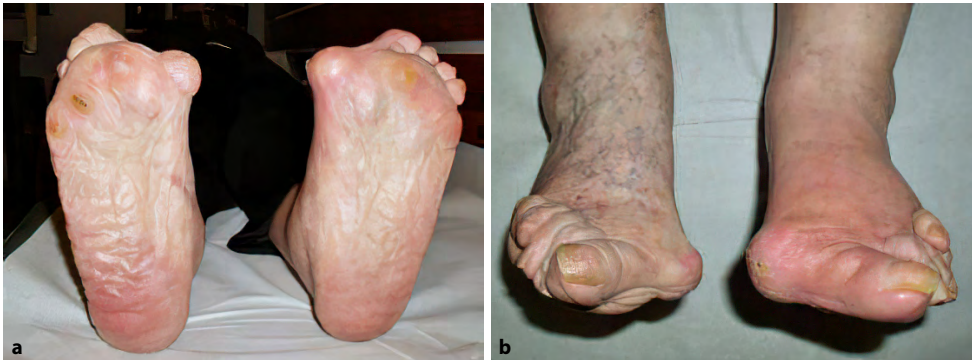


Fig. 7a, b. Female, 70. Maximal deformation of the feet. Pedes plani. Halluces valgi. The cause of hallux valgus on both sides and full hyperextension position of toes – wearing improper shoes in the youth. Flexion test – maximal pathology. Extensive pain in feet.
Fully unable to walk. Range of flexion of toes in metatarsal – phalange joints – hyperextension of toes 50°.

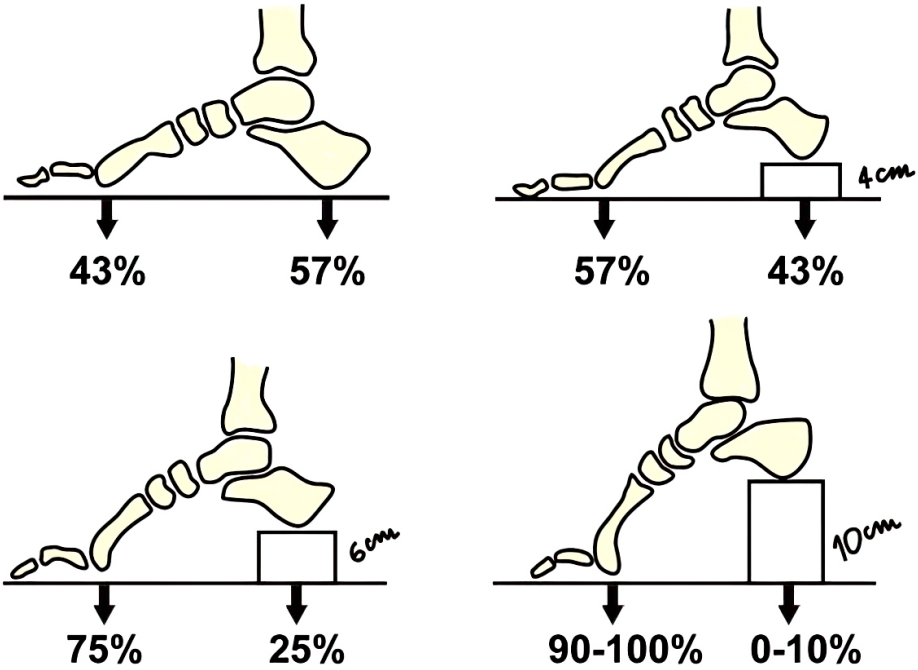


Fig. 8. Picture form the Internet. Loading of the foot depending on the type of shoes. Pathologic role of high heels. In result – disturbing of the gait, toe deformity, disturbing of blood circulation and in result – Morbus Köhler II or Morton metatarsalgy.

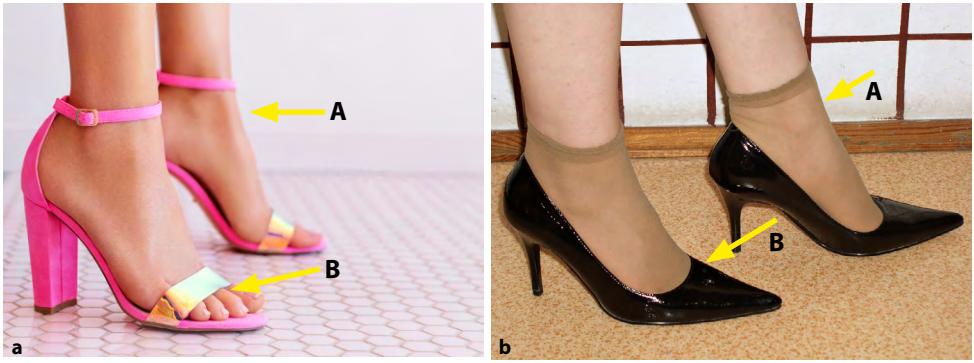


Fig. 9a, b. Improper shoes (Fig 9a – picture taken from the Internet). Heels too high. (A) Feet in plantar flexion. With the time limited dorsal flexion. (B) Toes in dorsal flexion. Normal propulsion in gait is impossible, walking disturbed.

PHYSIOTHERAPY

Good experience in this therapy have co-authors of this paper – Mgr. B. Slowinska and Mgr. B. Boryga – both physiotherapists. When a patient come to us with a problem of pain in the front part of one foot or both feet, we should check the range of plantar flexion of the toes. When the plantar flexion of the toes is small – for example only 5 or 10 degrees, or 0 degree or even "contracture in hyperextension" this mean – the pain is connected with "disturbed function of the front part of the foot or feet" and additionally with deformities of the foot. When plantar flexion of the toes is limited, kinesiotherapy (**Fig. 10**), i.e., toe flexion exercises in warm water, or another form of therapy should be recommended. Exercises should be passive, performed with the help of physiotherapists, and also active, performed by the patient alone over a long period of time, even many months or years. Physiotherapists should also inform the patients about using the proper shoes. The treatment is long-term, it can take months or even years, but a positive effect we have to see in all patients. Good results have been observed also in patients with deformities like hallux valgus, "hammer toes", valgus of the feet (**Fig. 7a, 7b**) – because the "first problem" is not connected with deformity but with "insufficiency of function".

DISCUSSION

Knowledge for physiotherapists, rehabilitation doctors and general physicians. People suffer, mostly because of pain syndromes of spine, of the hips, knees and shoulder. Nonetheless, many suffer because of feet deformities (**References 1–19, 28, 29**). This group of patients come to the doctor because of "pain syndromes" in the frontal part of the feet and difficulties in waking. This pathology is related with "restriction of the toes flexion" in the metatarsal – phalangeal joints. Limited flexion movement of toes disturb walking and other forms of daily activities. Very often in these patient groups the skin on the plantar surface of the feet is "pathologically changed" in the form of "thickness", "corn", footprint", "toe prints". In any form of deformity, such as hallux valgus, pain is the first

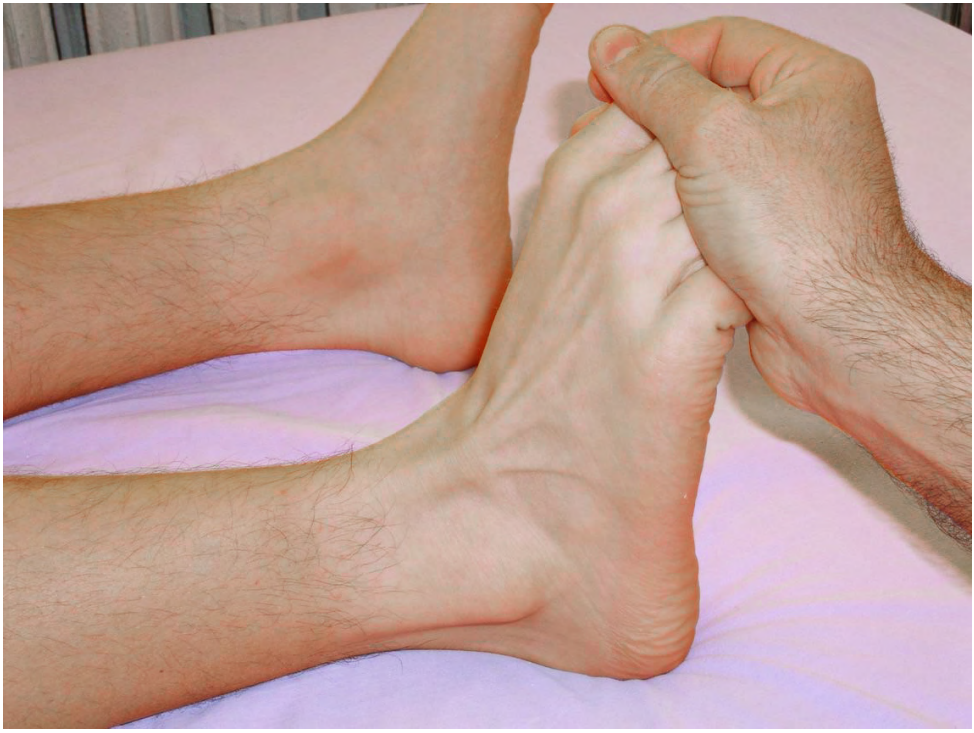


Fig. 10. Kinesiotherapy. Methods of the therapy – on the basis of publications from 1971 and 1985. Thermotherapy increases the effectiveness of exercises considerably.

priority. In the opinion of the patients, it is not the deformity but the limited plantar flexion of the toes that causes the difficulty. Passive and active plantar flexion exercises of the toes are important in therapy.

Another group of patients with foot problems are young girls with symptoms of "necrosis of the metatarsal head", the so-called Köhler II disease. Also in this group – physiotherapy, toe flexion exercises and thermotherapy bring good results. Over the years – in observation of all the authors (see Literature) – the methods of therapy – to receive full flexion of toes is sole proper treatment.

CONCLUSION

1. For proper function and normal activity of everyone it is necessary to have the proper anatomy of the bones, muscles and joints, full range of movements of joints and good blood circulation, nerve system function, as well proper function of all internal organs.

2. Every limitation of joint movement is the cause of pain syndromes. These "pain symptoms" we observe in the spine, in the hips, knees, shoulders and also in the feet.
3. The problem of instability of the ankle joints with symptoms of pain, or swelling in the tarsus region, disturbances of walking is described in articles in the USA, India, Czech Republic in 2017 – 2021 (see references).
4. The limited plantar flexion in the metatarsal – phalangeal joints of the feet is the cause of pain, disturbance when walking and in other daily activities.
5. The limitation of plantar flexion of the toes is caused by wearing a "too narrow improper shoes" in the early period of life.
6. Too "narrow improper shoes" are also the causes of deformities like hallux valgus, "hammer toes", necrosis of the head of metatarsal bones (Köhler II disease) and various skin pathologies.
7. In the paper we present the "toe flexion test", examples of pathology and we give advice for physiotherapy.

Acknowledgment

Many thanks for correction of the English in the article to David Poynton - Lublin, Poland and to Honorata Menet – Caen, France.

REFERENCES

1. ARROYO-HERNÁNDEZ M, MELLADO-ROMERO M, PÁRAMO-DÍAZ P, GARCÍA-LAMAS L, VILÀ-RICO J. Chronic ankle instability: Arthroscopic anatomical repair. Revista Española de Cirugía Ortopédica y Traumatología (English Edition). 2017; 61(2): p. 104–10.
2. HURST B, BRANTHWAITE H, GREENHALGH A, CHOCKALINGAM N. Medical-grade footwear: the impact of fit and comfort. J Foot Ankle Res. 2017;10(1): p. 1–7.
3. KARSKI T., WARDA E [1970] „Typowe zniekształcenia statyczne stóp kobiet noszących niefizjologiczne obuwie" Polski Tygodnik Lekarski, XXV, 17, 1970.
4. KARSKI T. [1971] „Przydatność testu zgięcia podeszwowego palców w ocenie niewydolności stóp. Chirurgia Narządów Ruchu i Ortopedia Polska, XXXVI, 6, 1971.
5. KARSKI T. [1980] „Testy przydatne w ocenia zagrożeń stóp" „Biomechanika i profilaktyka statycznych zniekształceń stóp" Materials from the Scientific Session. Materiały z Sesji Naukowej, Lublin, 4-5, 12, 1979, PZWL, 1980, p. 113–116.
6. KARSKI T. [1973] „Niektóre problemy stóp dziecięcych" Pediatria Polska, XLVIII, 5, 1973: p. 609–612.
7. KARSKI T. [1979] „Testy przydatne w ocenia zagrożeń stóp" „Biomechanika i profilaktyka statycznych zniekształceń stóp" Materiały z Sesji Naukowej, Lublin, 4–5, 12, 1979, PZWL, 1980, p. 113–116.
8. KARSKI T. [1985] „Der Zehenflexionstest zur Erkennung Frühzeitiger Stadien von Funktionsstörungen und Deformitäten der Vorfusses" Beitr. Orthop. Traumat. 32/1985/, H. T., p. 305–307.
9. KARSKI T. [1990] Wrodzone i nabyte wady stóp u dzieci, Ortopedia, traumatologia i rehabilitacja narządów ruchu, pod redakcją Prof. St. Piątkowskiego, PZWL, Warszawa 1990, p. 217–220.

10. KARSKI T., W. KONERA, M. MALICKI [1990] Statische und jatrogene Knieckplattfusdeformitäten bei Kinder. Erklerung der Erscheinungen und Möglichkeiten der Therapie Szekesfehervar, 1990, October, 4–6, Dni Ortopedyczne Węgierskiego Towarzystwa Ortopedycznego.
11. KARSKI T. [1990]: Działalność Zespołu Podologicznego Lubelskich Klinik Ortopedii. Pod kierunkiem Prof. St. Piątkowskiego. Chir. Narz Ruchu i Ortop. Polska. LV, 1990, 3, p. 179–181.
12. KARSKI T., KARSKI J, SNELA S, OSTROWSKI J [1995] Knickfussdeformitäten bei Kindern mit spastischer Verkürzung der Achillessehne. Internationaler Gemeinschaftskongress, 1995, 15–17 Juni, Berlin, Kurzfassungen – Block 6
13. KARSKI J., KARSKI T., SNELA S., OSTROWSKI J [1996] Fussschäden bei Kindern mit spastischer Verkürzung der Achillessehne. Orthopädieschuhtechnik 9/1996, Zeitschrift für Prävention und Rehabilitation, p. 24–26.
14. KARSKI T., FRELEK-KARSKA M., PAPIERKOWSKI A., TACZAŁA J. [1996] Foot deformities with children with cerebral palsy Annual Meeting of the Hungarian Orthopedic Association, Abstracts, 20-21, Nyiregyhaza 20-22 June 1996.
15. KARSKI J., KARSKI T. [2014]: "Syndrome of contractures and deformities" according to Prof. Hans Mau as the primary cause of motoric deformities in children. Case studies including deformities of hips, neck, shank and spine. Archives of Physiotherapy and Global Researches, 2014, Vol.18, No 2. Formerly "Zeszyty Naukowe", Copyright 2014, Vincent Pol University In Lublin, Poland, p. 15–23.
16. KARSKI T., KARSKI J. [2015] „Syndrome of Contractures and Deformities" according to Prof. Hans Mau as Primary Cause of Hip, Neck, Shank and Spine Deformities in Babies, Youth and Adults. American Research Journal of Medicine and Surgery, Vol. 1, No. 2, 2015.
17. KARSKI J., KARSKI T., PYRC J., KULKA M. [2016]: Deformations of the Feet, Knees, Hips, Pelvis in Children and Adults with Minimal Brain Dysfunction. Causes. Treatment. Prophylaxis. Locomotor System, 23, 2016, No 2, p. 20–31.
18. KARSKI T., KARSKI J., PYRC J. [2017]: Mozna pricina bolestiveho Syndromu kloubu dolni koncetiny – popis a kasuistiky, A possible cause of the "Pain Syndrome of Lower Extremity Joints". Description and case reports. Locomotor System, 24, 2017, No 1, p. 10
19. KARSKI J., KARSKI T., KARSKA K., PYRC J. [2017]: Ankle Joint Pathology of Car Drives and Passengers. Case Report. American Research Journal of Medicine and Surgery. www.arjonline.org 2017, Volume 2, p. 1–12.
20. KARSKI T., KARSKI J. [2017]: Ankle Joint, Knee, Hip Distortion Syndrome Connected with the Using of Small Cars. Crimson Publishers, Ortho Res Online, Research Article, 2017, p. 1–4.
21. KARSKI T. [2017]: Physiotherapy – Correct, or Incorrect, Based on 'Wrong Principles of Treatment'. Example for Spine, Hip, Knee, Shank and Feet. Crimson Publishers [2017]: Ortho Res Online, Review Article, 2017, p. 1–5.
22. KARSKI T., KARSKI J., KARSKA K., KARSKA K. and MENET H. [2018]: Pediatric Prophylaxis Program of Motor System Deformations and Illnesses in Children. Problems of Spine, Hips, Knees and Feet, EC PAEDIATRICS, Review Article 7.7 (2018).
23. KARSKI T., KARSKI J., KARSKA K., KARSKA K. and MENET H. [2018]: Prophylactic Rules for Newborns, Babies, Children and Adults in problems of Hip, Knee, Shank, Feet and Spine, Online Journal CRIMSON PUBLISHERS, USA, Published: March 12, 2018.
24. KARSKI J., KARSKI T., KARSKA K., MENET H., PYRC J. [2020]: Pathology of Ankle Joint and Knee as a Result of Permanent Distortion Syndrome in Car Drives and Passengers, Clinical Surgery Journal, India-UK, 2020, p. 1–5.
25. KARSKI J., KARSKI T., KARSKA K., PYRC J. [2021] Rotation Distortion Syndrome of Ankle Joint and Knee in Car Drivers and Passengers, International Journal of Orthopedic Research, USA / Kansas, 4 (1), p. 17–21.
26. KARSKI T., KARSKI J. [2021] Toes Flexions Test to Recognize the Functional Status of the Foot: Examples of Pathology: Knowledge from 1971. Journal of Orthopaedic Science and Research, India / UK. 2021, 2 (2): p. 1–14.

27. MENZ HB. [2016] Chronic foot pain in older people. Maturitas Sep; 91: p. 110-4: /2016/j.maturitas.2016.06.011. Review.
28. VAILLANT J, REVILLET P, SEVENIER AM, JUVIN R.[2016]: Impact of fatigue on postural control in quiet standing in fibromyalgia. Ann Phys Rehabil Med. 2016 Sep; 59S:e124-e125. doi: 10.1016/j.rehab.2016.07.280.
29. www.ortopedia.karski.lublin.pl

Corresponding authors:
Jacek Karski
jkarski@vp.pl

Tomasz Karski
tmkarski@gmail.com

EXTENZOROVÉ LÉZE V ZÓNĚ III – BOUTONNIEROVA DEFORMITA

EXTENSOR LESIONS IN ZONE III – BOUTONNIER'S DEFORMITY

Jaroš Martin[1,2], Bejvlová Jarmila[2], Smrčka Václav[1,2]

[1] 1st Medical Faculty, Charles University in Prague, Czech Republic

[2] ESME s.r.o.

SOUHRN

Úvod
Léze ve III. zóně šlachy extenzorů nazýváme Boutonnierova deformita. Hlavní příčina postižení je disrupce středního pruhu extenzorů nad proximálním interfalangeálním kloubem. Výsledkem poranění je semiflektované postavení v PIP kloubu a zároveň hyperextenční postavení v DIP kloubu.

Materiál a metodika
Účelem této retrospektivní studie bylo stanovit úspěšnost konzervativní léčby Boutonnierovy deformity. Náš soubor zahrnoval 59 pacientů s Boutonnierovou deformitou. Během retrospektivní studie jsme vyšetřili deset pacientů.

Výsledky
Při hodnocení ztráty extenze PIP kloubu mělo 80 procent pacientů výsledky dobré až výborné.

Diskuze
V porovnání s výsledky studie zabývající se chirurgickou léčbou byly naše výsledky mírně lepší.

Klíčová slova: Boutonnierova deformita, proximální interfalangeální kloub, šlacha extenzorů

SUMMARY

Introduction
Zone III lesion of the extensor tendon is called Boutonniere deformity. The leading cause is disruption of the middle band of the extensor above the proximal interphalangeal joint. The result of injury is a semiflexed lag of the PIP joint and simultaneously hyperextended lag in the DIP joint.

Material and methods
The purpose of this retrospective study was to determine the success of conservative treatment of Boutonniere deformity. Our file included 59 patients with Boutonniere deformity, and we examined ten patients within retrospective study.

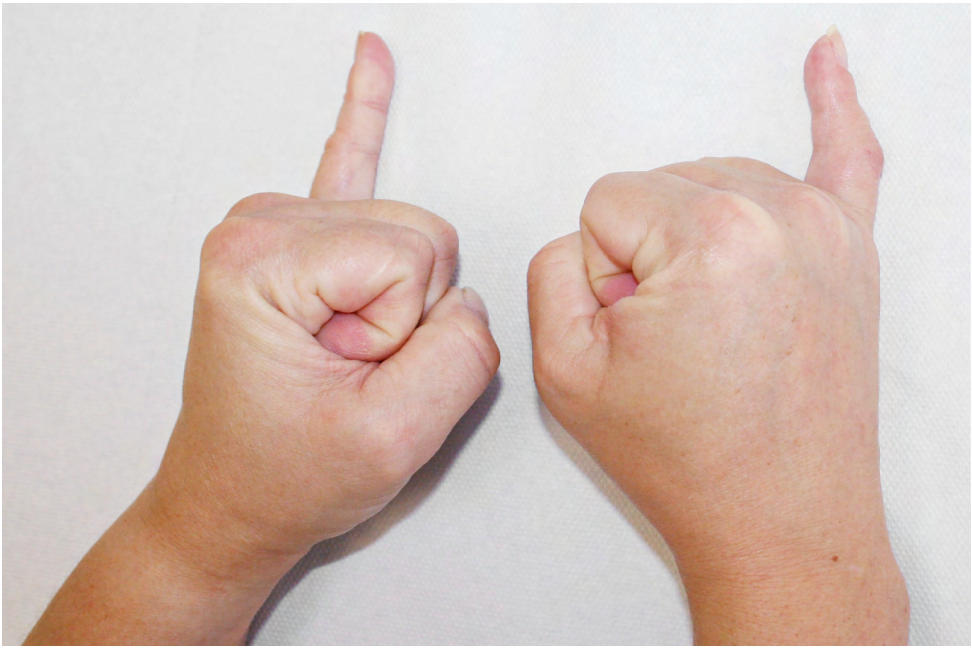
Results
In evaluating the loss of extension in the PIP joint, 80 percent achieved excellent/very good/good results.

Discussion
Our results were a little better compared to the study results dealing with surgical treatment.

Key words: Boutonniere deformity, proximal interphalangeal joint, extensor tendon

ÚVOD

Jako Boutonnierovu deformitu (knoflíkový prst) (**Obr. 1**) můžeme označit lézi se semiflekčním postavením v PIP kloubu a zároveň hyperextenčním postavení v DIP kloubu (**2, 15**). Hlavní faktor vzniku



Obr. 1 Boutonnierova deformita (pravá ruka)

Boutonnierovy deformity je přerušení středního extenzorového pruhu nad proximálním interfalangeálním kloubem. Mezi dalšími faktory, podílejícími se na vzniku, může být dysbalance postranních extenzorových pruhů a krátkých svalů, interoseů a lumbrikálů (**7, 8**).

Vznik knoflíkového prstu může zapříčiňovat celá řada mechanismů. Nejčastěji vzniká na podkladě úrazu, kdy dochází k poranění středního pruhu – discize, ruptura, lacerace, zavřené poranění. Doba od vzniku poranění po vytvoření Boutonnierovy deformity je přibližně 10–14 dní. Mezi další mechanismy vzniku patří např. popáleniny dorsa prstů, revmatoidní artritida (až polovina pacientů s revmatoidní artritidou vyvine Boutonnierovu deformitu), Dupuytrenova kontraktura a vrozené vady (**6**). V rámci sportu jsou ve zvláštním riziku vyvinutí této deformity hráči volejbalu a basketbalu (**11, 14, 17**), ale určit skutečnou incidenci mezi sportovci je složité.

Jedním ze základních vyšetření je Elsonův test (**5**). Vyšetřující ohne vyšetřovaný prst v PIP kloubu do 90 st. flexe, přičemž se pacient pokouší o aktivní extensi v DIP kloubu. Při disrupci středního extenzorového pruhu se u pacienta objeví hyperextenční postavení v DIP.

Závažnost Boutonnierovy deformity je možné klaifikovat dle různých klasifikačních stupnic, např. dle Soutera (**19**), dle Burtona (**3**) a dle Tubiany (**21**).

Důležité je odlišit Boutonnierovu deformitu od pseudo-Boutonnierovy deformity (**12, 16**), kde je sice semiflekční postavení v PIP kloubu, ale chybí hyperextenční postavení v DIP kloubu. Při pseudo-Boutonnierově deformitě není poškozen střední pruh extenzoru, ale při hyperextenčním poranění jsou postiženy palmárně umístěné měkké tkáně, které se jizví.

Chirurgická terapie

Chirurgická léčba u akutní uzavřené Boutonnierovy deformity je doporučována jen v případě, kdy byl střední pruh vytržen i s kostním fragmentem, který volně leží nad PIP kloubem, při nestabilitě PIP kloubu se ztrátou aktivní extense, u mladých pacientů při dlouho trvající zavřené Boutonnierově deformitě a též při selhání konzervativní léčby (**14**). Pro chirurgickou léčbu existuje celá řada technik, mezi nejvíce využívané techniky k omezení hyperextenze patří tenodéza FDS (flexor digitorum superficialis) (**13**), k dalším řadíme rekonstrukci dle Aiche, Barskyho a Weinera (**1**).

Konzervativní terapie

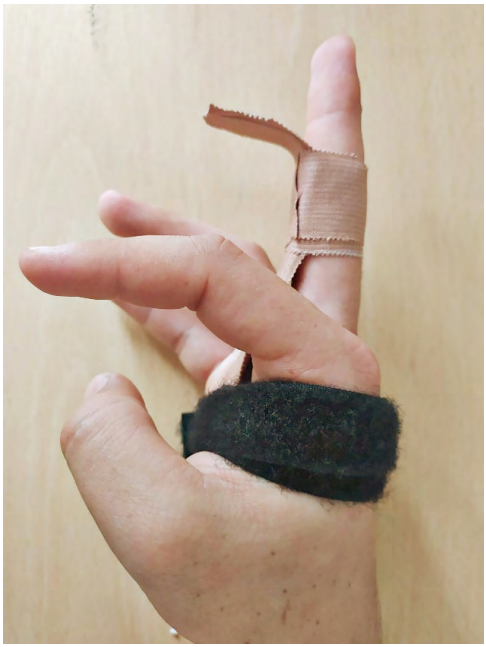
Základním principem konzervativní terapie je dlahování (**10, 11**). Užíváme dvě dlahy, polohovací dlahu na noc a Levameho dlahu na cvičení. Dlahy jsou vyrobeny z dvoumilimetrového nerezového plechu. Dlahy nelze modelovat manuálně, je nutné je upravovat a tvarovat na ponku a poté krýt náplastí. K prstům jsou fixovány náplastí a v oblasti zápěstí suchým zipem. Stresová deprivace z imobilizace má negativní biomechanický a biochemický vliv na šlachu, vaz i kloubní chrupavku (**9**). Časné zařazení aktivního pohybu do rehabilitace je důležité v prevenci adheze šlach a škodlivého efektu imobilizace. Proto se snažíme v prvních 4 týdnech o aktivní flexi v DIP kloubu za pomocí

Levameho dlahy (**Obr. 2**), která umožní repozici laterálních pruhů. Navíc, pokud je v PIP kloubu flekční postavení cca 60 stupňů a víc, je nutné polohovat prst na rovné dlaze i přes noc (**Obr. 3**). S aktivní flexí v DIP pokračujeme i dále ve čtvrtém až osmém týdnu, zároveň ale přidáváme cvičení aktivní flexe v PIP kloubu. Aktivní extenzi v PIP kloubu přidáváme mezi osmým a desátým týdnem. Cvičení s odporem a běžná činnost je doporučena od desátého týdne. Zlepšení flekčního postavení v PIP kloubu během léčby by se zpravidla mělo pohybovat asi 10 stupňů za měsíc. Pokud tomu tak není, je vhodné se zamyslet nad compliancí pacienta.

Retrospektivní studie – soubor pacientů

Hlavním cílem naší studie bylo stanovit úspěšnost konzervativní terapie Boutonnierovy deformity. Celý soubor pacientů s Boutonnierovou deformitou obsahoval 59 pacientů, z nichž se k vyšetření dostavilo 10, kteří byli v rámci studie hodnoceni. K hodnocení výsledků studie nám posloužil námi vytvořený dotazník. Měření úhlu defektu extenze a flexe prováděla jedna a táž osoba pomocí kruhového goniometru. Zároveň stejné úhly zjišťovala i na odpovídajícím zdravém prstu na kontralaterální ruce. Všichni pacienti podstoupili konzervativní léčbu, jeden zároveň i léčbu operační.

Nejčastější mechanismus (70 procent) vzniku Boutonnierovy deformity v našem vzorku byl úraz. U jednoho pacienta deformita vznikla po artritidě, u dalšího byla příčina vrozená a u jednoho se nepodařila určit.



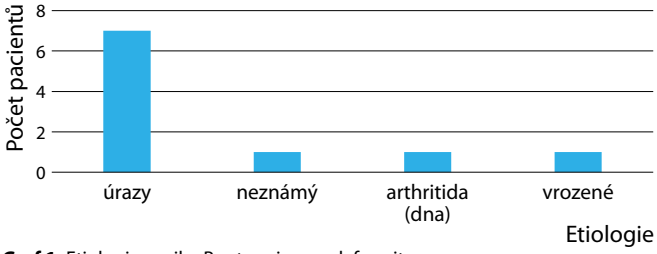
Obr. 2 Levameho dlaha – přes den

Obr. 3 Polohovací dlaha – na noc

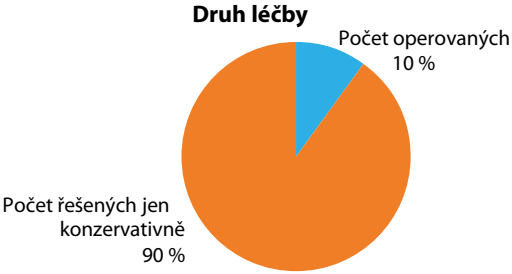
90 procent pacientů byli muži. Věkový rozptyl hodnocených byl mezi 10 až 88 lety. 70 procent defektů vzniklo na levé ruce a zároveň 70 procent bylo na ruce nedominantní.

U pacientů s úrazovou etiologií vzniku začala léčba nejdříve po týdnu, a nejdéle po čtyřech měsících po úrazu.

Každý z pacientů užíval alespoň jednu dlahu. Všichni užívali Levameho dlahu, v rozpětí od jednoho měsíce do 18 měsíců a 8 pacientů z našeho vzorku zároveň i rovnou dlahu, a to v délce od jednoho měsíce do 24 měsíců.



Graf 1: Etiologie vzniku Boutonnierovy deformity

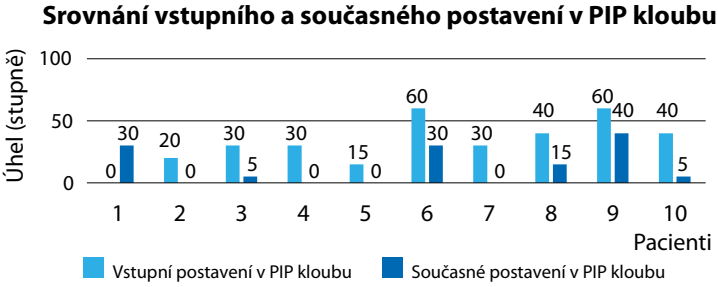


Graf 2: Druh léčby Boutonnierovy deformity

VÝSLEDKY

Hodnotit závažnost Boutonnierovy deformity můžeme pomocí různých klasifikačních stupnic, například podle Soutera (**19**), dle Burtona (**3**) a dle Tubiany (**21**). V naší studii jsme použili modifikovanou klasifikaci dle Soutera. Klasifikace hodnotila 3 různé kategorie po pěti stupních. V první kategorii jsme posuzovali ztrátu extenze v PIP kloubu. První stupeň (výborný) byl 0 stupňů, druhý (velmi dobrý) byl méně než 10 stupňů, třetí (dobrý) méně než 20, čtvrtý (slušný) méně než 40 a pátý (špatný) více než 40. Druhá kategorie hodnotila vzdálenost špičky prstu od distální dlaňové řasy. Stupně byly první 0 cm, druhý méně než 2 cm, třetí méně než 2,5 cm, čtvrtý méně než 4, pátý více

než 4 cm. Třetí kategorie hodnotí flexi v PIP kloubu. První stupeň je normální flexe, druhý více než 90 stupňů, třetí více než 80, čtvrtý více než 75 a pátý menší než 75 stupňů. Stejně jsme hodnotili i prst na kontralaterální straně.



Graf 3: Srovnání vstupního a současného postavení v PIP kloubu

Pacient	Defekt exten. PIP (stup.)	Vzdálenost špička-dist. dl. řasa (cm)	Flexe v PIP
1	Dobré	Velmi dobré	Velmi dobré
2	Výborné	Výborné	Výborné
3	Velmi dobré	Výborné	Výborné
4	Dobré	Výborné	Slušné
5	Výborné	Výborné	Výborné
6	Slušné	Velmi dobré	Dobré
7	Výborné	Výborné	Výborné
8	Dobré	Velmi dobré	Velmi dobré
9	Špatné	Výborné	Výborné
10	Velmi dobré	Výborné	Výborné

Tabulka 1: Hodnocení výsledků léčby u jednotlivých pacientů ve 3 kategoriích dle Soutera

	Kategorie dle Soutera		
Stupně dle Soutera	**Defekt exten. v PIP (stup.)**	**Vzdálenost špička-dist. dl. řasa (cm)**	**Flexe v PIP**
Výborné	30 %	70 %	60 %
Velmi dobré	20 %	30 %	20 %
Dobré	30 %	0 %	10 %
Slušné	10 %	0 %	10 %
Špatné	10 %	0 %	0 %

Tabulka 2: Přehled jednotlivých kategorií a stupňů dle Soutera. V sloupcích jsou procentuálně výsledky léčby pacientů v každé z kategorií.

DISKUZE

Naše výsledky jsem porovnal s výsledky dosaženými chirurgickou léčbou a publikovanými v roce 2003 v retrospektivní studii Catalanem (**4**). Jejich vzorek obsahuje 12 pacientů s věkovým průměrem 41 let a úrazovým mechanismem vzniku deformity. Principem chirurgické léčby byla tenodéza FDS. V rámci studie bylo mimo jiné měřeno vstupní a výstupní postavení. Průměrné vstupní postavení stanovené Catalanem činilo 31 stupňů, výstupní 12. Tyto výsledky jsem porovnal s naším vzorkem, z kterého jsem použil osm pacientů, když jsem předem vyřadil pacienta zároveň podstoupivšího i chirurgickou léčbu a pacienta s vrozenou etiologií. Naše průměrné vstupní postavení bylo cca 33,125 stupňů, výstupní cca 8,75 stupňů. Z těchto výsledků je vidět, že i když vstupní postavení v PIP kloubu bylo mírně horší v našem vzorku pacientů, po absolvování terapie bylo výsledné postavení v PIP kloubu mírně lepší u pacientů s konzervativní léčbou.

ZÁVĚR

U 80 procent pacientů se dosáhlo výborných až dobrých výsledků extenze v PIP kloubu. Při měření flexe v PIP kloubu jsme zjistili ještě lepší výsledky, a to výborné až dobré, u 90 procent pacientů. 100 procent pacientů prst dlahovalo, 10 procent navíc podstoupilo i operační léčbu. Průměrná doba užívání Levameho dlahy bylo 6,1 měsíců, rovné dlahy 5,6 měsíců.

LITERATURA

1. AICHE A., BARSKY A. J., WEINER D.L.: Prevention of the boutonniere deformity. *Plast Reconstr Surg.* 46, 1970, č. 2: 164–7.
2. BINSTEAD J. T., TAFTI D., HATCHER J. D.: Boutonniere Deformity. In: StatPearls [Internet]. Treasure Island (FL): *StatPearls Publishing*; 2021 Jan-.
3. BURTON R.I.: Extensor tendons-late reconstruction, in Green D.P., ed: *Operative Hand Surgery.* New York, Churchill Livingstone, 1988, 2073–2116.
4. CATALANO, L. W., SKARPARIS A. C., GLICKEL S. Z. et al.: Treatment of chronic, traumatic hyperextension deformities of the proximal interphalangeal joint with flexor digitorum superficialis tenodesis. *The Journal of hand surgery* [online]. 28, 2003, č. 3: 448–452
5. ELSON R. A.: Rupture of the central slip of the extensor hood of the finger. A test for early diagnosis. *J Bone Joint Surg Br.* 68, 1986, č. 2: 229–31
6. GRANDIZIO L. C., KLENA J. C.: Sagittal band, boutonniere, and pulley injuries in the athlete. *Curr Rev Musculoskelet Med*. 10, 2017, č. 1: 17–22
7. GRAU L., BAYDOUN H., CHEN K. et al.: Biomechanics of the Acute Boutonniere Deformity, *JHS*, 43, 2018, č. 1: 80.e1–80.e6
8. HANZ K., SAINT-CYR M., SEMMLER M. et al.: Extensor Tendon Injuries: Acute Management and Secondary Reconstruction, *Plastic and Reconstructive Surgery, 121*, 2008, č. 3: 109e–120e.

9. JOHNSON C., SWANSON M., MANOLOPOULOS K.: Treatment of a zone III extensor tendon injury using a single relative motion with dorsal hood orthosis and a modified short arc motion protocol A case report. *Journal of hand therapy,* 34, 2021, č. 1: 135–141
10. KNAP O., BEJVLOVÁ J., SMRČKA V.: Rehabilitace extenzorových lézí: *Pohybové ústrojí,* 27, 2020, č. 1: 46–55
11. MARIANO J. T., LOURIE G. M.: Boutonnière and pulley rupture in elite athletes. Hand Clin. 28, 2012 č. 3: 437–45
12. McCUE F. C., HONNER R., GIECK J. H. et al.: A pseudo-boutonniere deformity. *The Hand*, *7,* 1975, č. 2: 171–174
13. MCKEON K. E., LEE D. H.: Posttraumatic Boutonnière and Swan Neck Deformities. *Journal of the American Academy of Orthopaedic Surgeons.* 23, 2015, č. 10: 623–632
14. OWEN, J. M., WATTS A. C.: (iii) Extensor tendon injuries. *Orthopaedics and trauma*. 28, 2014, č. 4: 214–218
15. RAO K., MCCARTEN G.: Extensor Tendon Injuries. In Rostam F., Bulstrode N., Mehrara B.J. et al., Plastic Surgery – Principles and Practice, Philadelphia: Elsevier 2021, s. 750–754
16. ROTHWELL A. G.: The pseudo-boutonniere deformity. N Z Med J. 89, 1979; č. 628: 51–4
17. SMITH D. W.: Boutonnière and pulley rupture in elite basketball. *Hand Clin.* 28, 2012, č. 3: 449–50
18. SMRČKA V., DYLEVSKÝ I., MAŘÍK I. Extenzory ruky. [1. vyd.]. Brno: Institut pro další vzdělávání pracovníků ve zdravotnictví, 1998, s. 58–69
19. SOUTER WA.: The problem of boutonniere deformity. *Clin Orthop Relat Res.* 104, 1974, č. 104: 116–33
20. STRAUCH R. J., Extensor Tendon Injury. Wolfe S. W., Pederson W. C., Kozin S. H. et al. In: Green's Opertaive Hand Surgery E-book, Philadelphia: Elsevier, 2016
21. TUBIANA R., VALENTIN P.: The Physiology of the Extension of the Fingers. *Surgical Clinics of North America*: 44, 1964, č. 4: 907–918

Kontaktní adresa:
MUDr. Martin Jaroš
email: martin.jaros@seznam.cz

DUÁLNÍ EMISNÍ RENTGENOVÁ ABSORPCIOMETRIE PRO STANOVENÍ HMOTNOSTI AMPUTOVANÉ ČÁSTI KONČETINY PO TRANSTIBIÁLNÍ AMPUTACI

DUAL-EMISSION X-RAY ABSORPTIOMETRY FOR DETERMINING THE WEIGHT OF AN AMPUTATED LIMB PART AFTER TRANSTIBIAL AMPUTATION

Krawczyk Petr[1,2], Macháčková Jitka[3], Kutáč Petr[3], Uchytil Jaroslav[3], Zemková Dana[4,6], Mařík Ivo[5,6], Bužga Marek[3]

1 PROTEOR CZ s.r.o., Ostrava, Czech Republic

2 Department of Anatomy and Biomechanics, Faculty of Physical Education and Sport, Charles University, Prague, Czech Republic

3 Department of Human Movement Studies, Human Motion Diagnostic Centre, University of Ostrava, Ostrava, Czech Republic

4 Department of Paediatrics, Charles University Hospital Motol, Prague, Czech Republic

5 Faculty of Medical Studies, West Bohemia University, Pilsen, Czech Republic

6 Centre for Defects of Locomotor Apparatus l.l.c., Prague, Czech Republic

Correspondence to:

Petr Krawczyk, MD., email: petr.krawczyk@seznam.cz

ABSTRACT

Background

Most studies addressing the composition and volume of an amputated limb in lower extremity amputations have focused on determining the amputation stump volume. Examination of the weight of the amputated part of the limb is important for guiding the application of a definitive prosthesis.

Objectives

To assess the theoretical value of the weight of the amputated part of the lower limb in patients after transtibial amputation.

Study design
Cross-sectional study.

Methods
14 patients with transtibial amputation were enrolled, along with 14 matching controls. We assessed limb weight using dual-emission X-ray absorptiometry (DXA) and compared these values to those derived using anthropometric models (per Zatsiorsky, Osterkamp, and Mozumdar).

Results
The lowest average (± standard deviation) weight determined for the hypothetical distal part of the amputated limb by DXA was –3.11±1.13 kg. Compared to DXA, anthropometric models provided significantly higher values. The highest value (4.85 ± 0.77 kg) resulted from using calculations according to Osterkamp, and the model proposed by Mosumdar gave the lowest value (3.43±0.45), which was closest to that derived using DXA.

Conclusion
DXA is generally considered the gold standard for measuring body composition. Our comparison of data measured with DXA to that from anthropometric models shows that calculations of amputation mass using selected anthropometric models can significantly overestimate the weight of an amputated limb.

Keywords: body segment parameter, lower limb, amputation, DXA, anthropometryze

BACKGROUND

Dual-emission X-ray absorptiometry for determining the weight of an amputated lower limb part after transtibial amputation

Proper and timely fitting of a prosthetic device is essential in the care of patients after lower limb amputations. The reduction of limb stump oedema, prevention of contractures, and loss of physical condition are all important factors to consider in ensuring success with prosthetic and rehabilitation care. Another issue is the type of prosthetic replacement that is best for an individual patient.

Loss of the lower limb or part of it leads to serious changes and damage to the musculoskeletal system. The correct choice of prosthesis attracts most of the emphasis with regard to cognitive function and physical activity. A recent general trend has been toward development of prostheses that are aslight, highly functional, and comfortable as possible. However, the literature offers no reports comparing the weights of the amputated limb and prosthesis to healthy limb weight. Furthermore, few studies have examined the effect of prosthesis weight on the energy demands of gait in patients with transtibial amputation (TTA).[1,2,3] Anthropometric methods have been proposed as a way to determine the expected weight of the amputated part of the limb.[4]

This gap is an important one to address because patients have approximately the same weight in both lower limbs until amputation, and post-amputation weight and length asymmetries can have negative biomechanical consequences. Pertinent issues in orthopaedic prosthetic management include ensuring consonance betweenthe prosthesis weight and the weight of the amputated part.

Several approaches are available for calculating the weight and volume of a limb, and a number of methods also are available for accurate measurements of body segments. We can classify these as direct methods, using cadavers or imaging in living persons, and indirect methods, including anthropometric models.[5] Extrapolation of results to the general population is relatively difficult because body fluid ratios and muscle mass volume change with age and somatotypes differ among generations (a secular growth trend).[6] Direct measurements in volunteers and patients using imaging tend to be closer to exact values. Initially, the most common modality was X-rays, to be followed by computed tomography and magnetic resonance imaging.[7] Despite their applicability, however, these methods have limitations, including price, complexity, length of examination, and radiation exposure (X-ray, computed tomography). A further important limitation is complexity of the resulting data, which precludes their full use in standard clinical practice. Therefore, these methods typically apply only in the research setting.

An available non-invasive and direct reference method for body examination is dual-emission X-ray absorptiometry (DXA).[8] DXA, which allows for the examination of bone density and soft tissues, has become the gold standard for precise evaluation of body composition. [9] The low radiation dose (approximately 1 mSv/scan) is an undeniable advantage, falling below natural background radiation, as compared to >10 mSv for a whole-body image using computed tomography.[10]

In addition to direct methods, other methods are available that are based on the relationship between anthropometric parameters and the actual weight and distribution of body tissues.[11] The advantage of anthropometric models is their low demand, low cost of measuring equipment, and absence of radiation exposure. On the other hand, these methods have several pitfalls. Anthropometric measurement requires a certain routine and a degree of individual accuracy and thus always carries the burden of "human error." Another difficulty is the calculation of the measured values in a regression equation. Specific population groups form the basis for regressions, which may not fully correspond to the group of patients measured.[12]

This study is the first to involve measurement of the missing part of an amputated limb using DXA and to compare the results to those obtained with standard anthropometric models. Our aim was to evaluate the weight of the amputated part of the lower limb in patients undergoing TTA. Our secondary goal was to compare the weight of the amputated limb and prosthesis with the weight of a healthy limb.

METHODS

Participants

From May 2017 to October 2018, we examined patients after TTA and performed measurements in a control group of volunteers at the same time. The group with TTA had a total of 14 persons (13 men and 1 woman) with a mean age of 51.5 years and a mean BMI (±standard deviation) of 27.7 ± 3.6 kg/m^2. The average weight of the prosthetic equipment was 2.39 ± 0.21 kg, whereas the weight of the amputation stump according to DXA was 1.18 ± 0.47 kg. The mean weight of the healthy shank lower limb in patients was 4.66 ± 0.72 kg.

Table 1 provides the basic information about the group of patients after TTA. Patients in this group were selected at a regional prosthetic centre. Inclusion criteria were unilateral transtibial amputation, willingness to cooperate with the examination, and a limb amputation at least 12 months previously. Exclusion criteria were oedema of the amputated limb, cachexia, and obesity. Indications for TTA were peripheral vascular complications in the absence of diabetes (n = 1), traumatic amputations (n = 9), malignancies (n = 1), and infections (n = 3). Average time after amputation was 13.9 ± 11.6 years.

A total of 96 people were included as control participants, divided into three groups. The first group consisted of participants matched to patients for somatic features and age (*n* = 14, mean age was 47.5 ± 12.2 years with mean BMI of 26.6 ±2.1 kg/m^2). The second group consisted of non-obese individuals who engaged in regular physical activity but did not have a dominant lower limb (*n* = 63). Members of the third control group were all high-jump or long-jump athletes, so that one of their lower limbs was dominant (the lead leg for jumping), and all met the inclusion criterion of specialisation in jumping training for at least 2 years (*n* = 19). **Table 1** lists the somatic parameters of the participants in the three control groups.

Group	n	Age (y)	Height (cm)	Weight (kg)	BF (kg)	BF (%)
ALL M	13	51.5 ± 12.9	176.5 ± 5.9	83.5 ± 12.1	23.7 ± 7.9	27.3 ± 5.8
ALL F	1	47.8	16.5	65.9	23.4	35.0
CG1 M	13	47.5 ± 12.2	177.7 ± 6.6	84.0 ± 8.6	22.2 ± 5.1	26.2 ± 4.7
CG1 F	1	50.1	157.5	63.5	22.1	34.9
CG2M	38	22.6 ± 2.9	180.6 ± 6.5	77.1 ± 8.6	13.5 ± 3.5	17.5 ± 3.9
CG2F	25	21.4 ± 2.0	168.0 ± 6.5	58.2 ± 7.0	15.3 ± 3.0	26.2 ± 4.1
CG3M	10	18.7 ± 3.2	181.0 ± 4.9	69.2 ± 4.9	9.9 ± 1.8	14.3 ± 2.3
CG3F	9	17.4 ± 2.3	172.4 ± 9.4	61.7 ± 6.5	15.4 ± 2.3	25.1 ± 2.2

ALL M – amputated lower limb, male; ALL F – amputated lower limb, female; CG1 M – matched control group, male; CG1 F – matched control group, female; CG2 M – control group 2, male (healthy without sports); CG2 F – control group, female (healthy without sports); CG3 M – control group 3, male (active athletes); CG3 F – control group 3, female (active athletes); n – frequency; BF – body fat

Table 1. Descriptive characteristics of control groups used for the comparison of lower limb symmetry (mean ± SD)

All study participants signed an informed consent. All procedures performed in the study were in accordance with the ethical standards of the 2013 Declaration of Helsinki.

All study participants underwent initial measurement of somatic parameters and lower limb weight. We used certified gauges to take weight and height measures of participants in their under wear. In patients with amputation, weight with and without the prosthesis and of the prosthesis it self was measured. Likewise, we used a standardised band gauge to measure limb circumferences and examined lower limb weight using a bone densitometer (Discovery A; Hologic, Waltham, MA, USA) calibrated according to the manufacturer's recommendations with an established precision error.[13] To calculate the weight of the lower limb based on bone densitometer data, we summed bone mineral content, fat free mass, and body fat. Using the sub-region function, we defined the regions in the obtained densitometric data as "thigh" and "calf" and summed them to obtain the total weight of the lower limb. We chose this procedure because of an overlap of parts of individual lower limbs (right and left) when defining the measurement of the sub-region in the entire lower limb. To verify the symmetry of lower limb weights in healthy participants, we compared the weights of the right and left lower limbs in all three control groups.

Calculation of the weight of the distal part of the amputated limb using anthropometric methods

To compare weight measurements achieved with DXA, we used established and generally known calculation methods with regression equations. For this comparison, we used equations of anthropometric models according to both the original and modified equations of Zatsiorski[14,15] and the equations of Osterkamp[16] and Mozumdar[17].

Calculation of the hypothetical weight of the distal part of the amputated limb using DXA

For calculating weight, we used methods for determining the weight of individual segments of the lower limb, which are standardised DXA measurements provided by the device manufacturer. To

Group	LLEG (kg)	Sign.	RLEG (kg)	Sign.
CG1 (n = 14)	13.31 ± 1.63	0.095	13.59 ± 1.69	0.087
CG2 (n = 63)	12.60 ± 2.10	0.504	12.72 ± 2.20	0.599
CG3 (n = 19)	12.80 ± 1.52	0.277	12.93 ± 1.55	0.301

Group	LLEG (kg)		RLEG (kg)	
CG1 (n = 14)	4.70 ± 0.72	0.181	4.59 ± 0.65	0.098

CG1 – matched control group; CG2 – physically active without a dominant lower limb; CG3 – physically active with a dominant lower limb (jumpers); LLEG – left limb; RLEG – right limb; n – frequency; Sign. – statistical significance

Table 2. Evaluation normality data of weight symmetry of lower limbs Shapiro-Wilxoc test

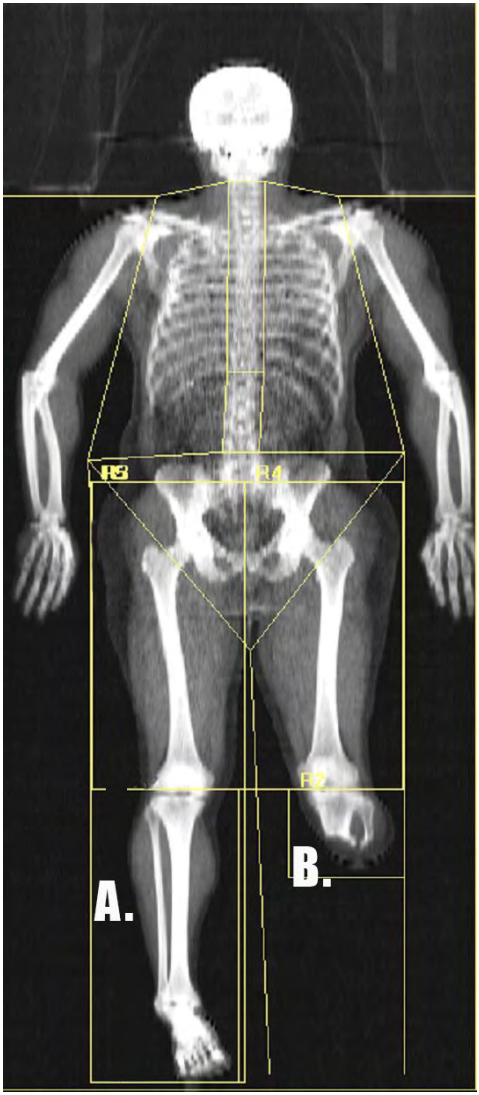


Figure 1. DXA scan of a patient after left-side TTA, with a marked region of interest for the calculation of the amputated part of the left lower limb.

determine the weight of the amputated limb, we took the difference between the weight of the shank of the healthy limb (measured from the knee to the distal end of the toes) and the preserved part of the shank of the contralateral amputated lower limb (measured from the knee to the distal end of the stump). **Figure 1** depicts the individual parts of the area of interest for the examination of the lower limb using DXA.

We performed comparative measurements in healthy controls to validate the calculation of the weight of the amputated lower limb and to compare the symmetry of the weights of the lower limbs using DXA. The weights of the right and left limbs from the first matched control group were compared to evaluate weight asymmetry of the distal part of the lower limb in healthy individuals. To evaluate the validity of anthropometric models for calculating the weight of the amputated part of the limb, we used the concept of criterion validity. We then used theresults measured via DXA as an empirical criterion for anthropometric models.

Statistical analysis of data

For statistics, we characterised the measure of central tendency (mean) and the measure of variability (standard deviation). For verifying the normality of data distribution for the various weights (as determined by DXA and anthropometric models), we used the Shapiro–Wilk test, and we applied independent t-tests to assess the statistical significance of differences in the means. (**Table 2**). For all tests, we set a significance level of $\alpha = 0.05$. We used the effect size according to Cohen (Cohen's *d*) to assess the general significance of differences in means. A *d* value at the level of 0.2 indicates a minor change, 0.5 an intermediate change, and 0.8 a major change.[18] To assess empirical validity, we used the Pearson correlation coefficient (*r*)

and standard error of estimation. We performed all statistical processing of results using IBM SPSS Statistics (Version 21; IBM, Armonk, NY, USA).

RESULTS

Comparison of control groups

We first compared groups of patients with TTA to the age- and somatically matched control group. Both groups were very similar in terms of somatic parameters. The control group of healthy non-obese participants without sports activity and the group of active athletes included younger participants with a lower BMI. However, the difference in age and BMI did not play a role in the study results, so we applied the data for these latter two control groups to verify the hypothesis of lower limb symmetry. All parameters of interest in all monitored groups had normal distribution, so we were able to use the parametric independent t-test to assess the statistical significance of differences in means. **Table 1** gives the data for the examined groups.

Comparison of symmetry of lower limbs

To test lower limb symmetry among the three control groups, we used Cohen's test. The age- and somatically matched control group did not show a difference in weight between the right and lower limbs (Cohen's $d = 0.17$). The weights of the right and left limbs also did not differ in the participants without obesity who were not athletes (Cohen's $d = 0.06$), and even in the third group of athletes who favoured one leg over the other for their sport, these weights did not differ (Cohen's $d = 0.08$). Thus, the right and left limbs did not differ in weight in any of the three control groups. (**Table 3**). Due to the fact that the normality of the data was not disturbed, Student's t-test was used.

Group	**LLEG (kg)**	**RLEG (kg)**	**Diff**	***P* (95% CI)**	***d***
	Whole lower limb				
CG1 (n = 14)	13.31 ± 1.63	13.59 ± 1.69	-0.28^{NS}	0.660 (-1.568; 1.009)	0.17
CG2 (n = 63)	12.60 ± 2.10	12.72 ± 2.20	-0.12^{NS}	0.744 (-0.883; 0.632)	0.06
CG3 (n = 19)	12.80 ± 1.52	12.93 ± 1.55	-0.13^{NS}	0.791 (-1.145; 0.879)	0.08
	Shank of the lower limb				
Group	**LLEG (kg)**	**RLEG (kg)**	**Diff**	***P* (95% CI)**	***d***
CG1 (n = 14)	4.70 ± 0.72	4.59 ± 0.65	0.11^{NS}	0.670 (-0.422; 0.646)	0.16

CG1 – matched control group; CG2 – physically active without a dominant lower limb; CG3 – physically active with a dominant lower limb (jumpers); LLEG – left limb; RLEG – right limb; n – frequency; Diff – difference; p – statistical significance; 95% CI – confidence interval; d – Cohen's d; ^{NS} non significant (t-test)

Table 3. Evaluation of weight symmetry of lower limbs (mean ± SD)

Comparison of shank symmetry

The evaluation of shanks was performed using Cohen's test in the age-matched control group. The result of Cohen's test on the shank part of the limb showed that the weight of the right and left limbs was symmetrical (d = 0.16; **Table 3**).

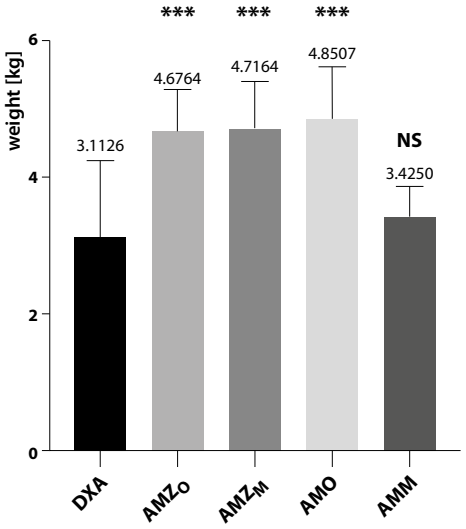


Figure 2. Weight of the amputated part of the lower limb

Evaluation of methods for measuring the weights of amputated limb parts in patients after TTA

With DXA, the lowest weight of the hypothetical distal part of the amputated limb was – 3.11 ± 1.13 kg. Compared to DXA, anthropometric models based on the regression equation showed significantly higher weights of the amputated part of the limb. Osterkamp's equation gave the highest weight, at 4.85 ± 0.77 kg, and Mosumdar's equation gave the lowest value at 3.43 ± 0.45 kg (**Figure 2**).

We found no significant differences between values measured using DXA and those calculated with the Mosumdar model. In the other cases of comparison between DXA and the anthropomorphic models, however, the differences were statistically significant ($p < 0.001$) and otherwise significant (Cohen's $d > 0.8$).

	Diff (kg)	d	r	$S_{y/x}$ (kg)	d_{max} (kg)
AMZ_O	1.58***	1.74	0.31	1.12	2.24
AMZ_M	1.61***	1.72	0.41	1.07	2.14
AMO	1.74***	1.80	0.37	1.09	2.18
AMM	0.32^{NS}	0.37	0.42	1.07	2.14

AMZO – anthropometric model, Zatsiorsky original; AMZM – anthropometric model, Zatsiorsky modified; AMO – anthropometric model, Osterkamp; AMM – anthropometric model, Mosumdar; Diff – difference; d – Cohen's d; r – Pearson's correlation coefficient; ***p < 0.001; Sy/x (SEE) – standard estimation error; dmax – threshold estimation error (±2 Sy/x); NS non significant

Table 4. Differences in means and characteristics of empiric validity to the DXA method

The Pearson's correlation coefficients indicated little if any correlation with the values measured by DXA. The values of Pearson's coefficient explained only 9.6%–17.6% of the variance. The individual anthropometric models did not differ in standard estimation error or limit of observational error. **Table 4** shows differences in means and characteristics of the empirical validity of anthropometric models for DXA.

DISCUSSION

Technological advances in the prosthetics of lower limbs in the last decade have led to improved function for patients after limb amputation.[19] An essential goal after lower limb amputation is to be able to engage inactivities of daily life, adjust the parameters of the prosthetic so to comfortably and stably transfer the biomechanical load, and avoid overloading the musculoskeletal system.

Most studies addressing composition and volume in patients after amputation of the lower extremities have focused on determining the volume of the residual limb part.[20] The length of the amputate and composition of soft tissues both show extensive changes, especially during the first year after amputation.[21] A number of studies have addressed the volume of residual limb and the stabilisation of volume and composition during the maturation process.[22,23] Examination of residual volume is an essential clinical examination, especially in terms of the strategy of application of definitive prosthetic equipment.[25]

Volume, soft tissue composition, and muscle mass are of substantial significance. In recent years, several studies have shown loss of muscle mass after amputation.[25,26] Sherck et al. described this loss in 12 patients after transfemoral and transtibial amputation compared to the unaffected limb.[27] Similarly, Renström et al. described muscle atrophy, documenting the loss of muscle mass in both the affected and unaffected limbs.[28] An increase in the proportion of adipose tissue in inverse association with the loss of muscle mass also has been documented.[29]

Several recent studies have focused on the use of anthropometric methods and prediction equations to determine the body composition of lower limbs, especially in athletes and young populations[30,31], so that the results are not necessarily applicable to the general adult population. However, some studies have measured this composition in adults[32,33] and this composition has a substantial effect on cardiovascular health, in particular. Individuals with a higher BMI have a higher risk of both cardiovascular disease and type 2 diabetes and other serious lifestyle diseases. According to Wong et al., patients need to have a correction of body weight calculations after amputations for appropriate dietary measures to prevent obesity and diabetes.[34] This correction requires information not only about body weight but also about adjusted weight from the missing amputated part of the limb.[35]

Anthropometric models are available for calculating the weight of the missing part of the limb. However, these methods entail several methodological problems. The initial calculations of the weight of individual segments relied on a small number of individuals, mostly white men, without

consideration of changes in body composition in later generations[16]. For a long time, models based on cadaver measurements have been used for calculations.[11] Zatsiorsky and colleagues were the first to use imaging techniques to determine body segment parameters in a group of 115 young students (100 men and 15 women)[14,15] and their model is still in use. Osterkamp[16] and Mozumdar[17] introduced major innovations in their models, based on the measurement of 205 patients after amputations of both the upper and lower limbs. These models are widely accepted and used. Yet, we may question the accuracy of the results and ask whether the current progress in imaging methods allows for more accurate measurements. DXA, which we used for the comparison here, is one of the direct reference methods. As our results show, DXA yielded significantly lower values for lower limb weight compared to anthropometric models. This outcome is not surprising, given that most models are based on measurements of young healthy people or a limited number of cadavers. Only the Mozumdar model, which is based on measurements of patients after amputations, produced values similar to those obtained with DXA. We may therefore argue whether it is methodologically correct to use models based on the general population[14,15,16], to use a model based on patient data after amputation[17], or to make better use of the modern DXA imaging.

Our results offer a new perspective on the issue of determining the weight of the prosthesis with respect to the weight of the amputated limb. Current anthropometric models significantly overestimate the calculated limb weight compared to the DXA method. This methodology can be used in patients with acquired or congenital upper or lower limb amputation. Individual determination of the optimal weight of prosthetic equipment is very important for the proper development of the child, but also as a prevention of asymmetric loading of the musculoskeletal system in adults.

Limitations of this study

The patient group in our study was relatively small. Future studies should include a larger group of patients with balanced sex distribution for a more accurate evaluation of the results.

CONCLUSIONS

From the biomechanical point of view, the determination of the exact optimal length of the prosthesis, and especially its weight, is a neglected factor. The precise determination of the optimal weight of the prosthesis based on the previous weight of the amputated part of the limb (the amputate) is not sufficiently considered in prosthesis design. The calculation of the weight of the amputated part of the limb using indirect methods (e.g., the Zatsiorsky, Osterkamp, and Mozumdar models) over estimates the weight of the limb compared to using the direct DXA method. DXA seems to be the method of choice because it directly measures the composition of tissues and thus the weight in areas of interest for a particular patient. In our case, we measured the weight of the lower limbs in patients with TTA and in control groups. Based on our results, we recommend DXA imaging as a method of choice for the determination of amputate weight. Designing prostheses will require assessing the optimal weight of the prosthesis individually with regard to biological age and degree of physical activity.

Conflict of interest

None of the authors have a conflict of interest. The authors are all independent from funders. The sponsors had no influence on the writing of the manuscript. No other relationships or activities exist that could appear to have influenced the submitted work.

REFERENCES

1. LADLOW P, NIGHTINGALE TE, POLLY MCGUIGAN M, BENNETT AN, PHILLIP RD, BILZON JLJ. Predicting ambulatory energy expenditure in lower limb amputees using multi-sensor methods. PLoS One. 2019;14(1):1–18.
2. MCDONALD CL, KRAMER PA, MORGAN SJ, HALSNE EG, CHEEVER SM, HAFNER BJ. Energy expenditure in people with transtibial amputation walking with crossover and energy storing prosthetic feet: A randomized within-subject study. Gait Posture [Internet]. 2018;62(November 2017):349–54. Available from: https://doi.org/10.1016/j.gaitpost.2018.03.040
3. GAILEY RS, NASH MS, ATCHLEY TA, ZILMER RM, MOLINE-LITTLE GR, MORRIS-CRESSWELL N, et al. The effects of prosthesis mass on metabolic cost of ambulation in non-vascular trans-tibial amputees. Prosthet Orthot Int. 1997;21(1):9–16.
4. LIN-CHAN SJ, NIELSEN DH, YACK HJ, HSU MJ, SHURR DG. The Effects of Added Prosthetic Mass on Physiologic Responses and Stride Frequency during Multiple Speeds of Walking in Persons with Transtibial Amputation. Arch Phys Med Rehabil. 2003;84(12):1865–71.
5. ROSSI M, LYTTLE A, EL-SALLAM A, BENJANUVATRA N, BLANKSBY B. Body segment inertial parameters of elite swimmers using DXA and indirect methods. J Sport Sci Med. 2013;12(4):761–75.
6. DIANO D, PONTI F, GUERRI S, MERCATELLI D, AMADORI M, APARISI GÓMEZ MP, et al. Upper and lower limbs composition: a comparison between anthropometry and dual-energy X-ray absorptiometry in healthy people. Arch Osteoporos. 2017;12(1).
7. BAUM T, CORDES C, DIECKMEYER M, RUSCHKE S, FRANZ D, HAUNER H, et al. MR-based assessment of body fat distribution and characteristics. Eur J Radiol [Internet]. 2016;85(8):1512–8. Available from: http://dx.doi.org/10.1016/j.ejrad.2016.02.013
8. PETAK S, BARBU CG, YU EW, FIELDING R, MULLIGAN K, SABOWITZ B, et al. The Official Positions of the International Society for Clinical Densitometry: Body Composition Analysis Reporting. J Clin Densitom [Internet]. 2013 Oct 24;16(4):508–19. Available from: http://bmcpediatr.biomedcentral.com/articles/10.1186/s12887-016-0730-7
9. MARINANGELI CP, KASSIS AN. Use of dual X-ray absorptiometry to measure body mass during short- to medium-term trials of nutrition and exercise interventions. Nutr Rev. 2013;71(6):332–42.
10. ALBANESE C V., DIESSEL E, GENANT HK. Clinical Applications of Body Composition Measurements Using DXA. J Clin Densitom [Internet]. 2003 Jun;6(2):75–85. Available from: https://linkinghub.elsevier.com/retrieve/pii/S1094695006602564
11. DE LEVA P. Adjustments to Zatsiorsky-Seluyanov's segment inertia parameters. J Biomech [Internet]. 1996 Sep;29(9):1223–30. Available from: https://linkinghub.elsevier.com/retrieve/pii/0021929095001786
12. ZHENG YP, MAK AFT, LEUNG AKL. State-of-the-art methods for geometric and biomechanical assessments of residual limbs: A review. J Rehabil Res Dev. 2001;38(5):487–504.

13. KUTÁČ P, BUNC V, SIGMUND M. Whole-body dual-energy X-ray absorptiometry demonstrates better reliability than segmental body composition analysis in college-aged students. McLester CN, editor. PLoS One [Internet]. 2019 Apr 22;14(4):e0215599. Available from: http://dx.plos.org/10.1371/journal.pone.0215599
14. ZATSIORSKY V, SELUYANOV V. Mass and inertia characteristics of the main segment of the human body. In: Proceedings of the 8th International Congress of Biomechanics. Nagoya: International Series on Biomechanics; 1983. p. 1152–9.
15. ZATSIORSKY V, SELUYANOV V, CHUGUNOVA L. Methods of determining mass-inertial characteristics of human body segments. In: Chernyi G, Regirer S, editors. Contemporary Problems of Biomechanics. Massechusetts: CRC Press; 1990. p. 272–91.
16. OSTERKAMP LiK. Current Perspective on Assessment of Human Body Proportions of Relevance to Amputees. J Am Diet Assoc. 1995 Feb;95(2):215–8.
17. MOZUMDAR A, ROY SK. Method for estimating body weight in persons with lower-limb amputation and its implication for their nutritional assessment. Am J Clin Nutr. 2004;80(4):868–75.
18. COHEN J. Statistical power analysis for the behavioral sciences. 2nd ed. New Jersey: Lawrence Erlbaum Associates; 1988. 559 p.
19. DICKINSON AS, STEER JW, WOODS CJ, WORSLEY PR. Registering a methodology for imaging and analysis of residual-limb shape after transtibial amputation. J Rehabil Res Dev. 2016;53(2):207–18.
20. DE BOER-WILZING VG, BOLT A, GEERTZEN JH, EMMELOT CH, BAARS EC, DIJKSTRA PU. Variation in results of volume measurements of stumps of lower-limb amputees: A comparison of 4 methods. Arch Phys Med Rehabil [Internet]. 2011;92(6):941–6. Available from: http://dx.doi.org/10.1016/j.apmr.2011.01.007
21. PUTZ C, BLOCK J, GANTZ S, HEITZMANN DWW, DREHER T, LEHNER B, et al. Structural changes in the thigh muscles following trans-femoral amputation. Eur J Orthop Surg Traumatol. 2017;27(6):829–35.
22. TANTUA AT, GEERTZEN JHB, VAN DEN DUNGEN JJAM, BREEK JKC, DIJKSTRA PU. Reduction of residual limb volume in people with transtibial amputation. J Rehabil Res Dev. 2014;51(7):1119–26.
23. FERNIE GR, HOLLIDAY PJ. Volume fluctuations in the residual limbs of lower limb amputees. Arch Phys Med Rehabil [Internet]. 1982 Apr;63(4):162–5. Available from: http://www.ncbi.nlm.nih.gov/pubmed/7082139
24. SANDERS JE, FATONE S. Residual limb volume change: Systematic review of measurement and management. Vol. 48, Journal of Rehabilitation Research and Development. 2011. 949–986 p.
25. BOLT A, DE BOER-WILZING VG, GEERTZEN JHB, EMMELOT CH, BAARS ECT, DIJKSTRA PU. Variation in measurements of transtibial stump model volume: A comparison of five methods. Am J Phys Med Rehabil. 2010;89(5):376–84.
26. ECKARD CS, PRUZINER AL, SANCHEZ AD, ANDREWS AM. Metabolic and body composition changes in first year following traumatic amputation. J Rehabil Res Dev [Internet]. 2015;52(5):553–62. Available from: http://www.rehab.research.va.gov/jour/2015/525/pdf/JRRD-2014-02-0044.pdf
27. TUGCU I, SAFAZ I, YILMAZ B, GÖKTEPE AS, TASKAYNATAN MA, YAZICIOGLU K. Muscle strength and bone mineral density in mine victims with transtibial amputation. Prosthet Orthot Int. 2009;33(4):299–306.
28. SHERK VD, BEMBEN MG, BEMBEN DA. BMD and bone geometry in transtibial and transfemoral amputees. J Bone Miner Res. 2008;23(9):1449–57.
29. RENSTRÖM P, GRIMBY G, LARSSON E. Thigh muscle strength in below-knee amputees. Scand J Rehabil Med. 1983;15(9):163–73.
30. SHAHRIAR SH, MASUMI M, EDJTEHADI F, SOROUSH MR, SOVEID M, MOUSAVI B. Cardiovascular Risk Factors Among Males With War-Related Bilateral Lower Limb Amputation. Mil Med. 2009;174(10):1108–12.

31. HOLMES JD, ANDREWS DM, DURKIN JL, DOWLING JJ. Predicting in Vivo Soft Tissue Masses of the Lower Extremity Using Segment Anthropometric Measures and DXA. J Appl Biomech [Internet]. 2005 Nov; 21(4):371–82. Available from: https://journals.humankinetics.com/view/journals/jab/21/4/article-p371.xml
32. BELL W, COBNER DM, EVANS WD. Prediction and validation of fat-free mass in the lower limbs of young adult male Rugby Union players using dual-energy X-ray absorptiometry as the criterion measure. Ergonomics [Internet]. 2000 Oct 10;43(10):1708–17. Available from: https://www.tandfonline.com/doi/full/10.1080/001401300750004113
33. ARTHURS KL, ANDREWS DM. Upper extremity soft and rigid tissue mass prediction using segment anthropometric measures and DXA. J Biomech. 2009;42(3):389–94.
34. CARVALHO HM, COELHO-E-SILVA MJ, FRANCO S, FIGUEIREDO AJ, TAVARES ÓM, FERRY B, et al. Agreement between anthropometric and dualenergy X-ray absorptiometry assessments of lower-limb volumes and composition estimates in youth-club rugby athletes. Appl Physiol Nutr Metab. 2012;37(3):463–71.
35. WONG CK, WONG RJ. Standard and Amputation-Adjusted Body Mass Index Measures. Am J Phys Med Rehabil [Internet]. 2017 Dec;96(12):912–5. Available from: http://journals.lww.com/00002060-201712000-00011
36. FROST AP, NORMAN GIEST T, RUTA AA, SNOW TK, MILLARD-STAFFORD M. Limitations of body mass index for counseling individuals with unilateral lower extremity amputation. Prosthet Orthot Int [Internet]. 2017 Apr 19;41(2):186–93. Available from: http://journals.sagepub.com/doi/10.1177/0309364616650079

Contact address:
Petr Krawczyk, MD.
e-mail: petr.krawczyk@seznam.cz

METAFYZÁRNÍ ANADYSPLAZIE S VARÓZNÍMI FEMURY A MALÝMI ČÉŠKAMI: NOVÁ VARIANTA METAFYZÁRNÍ ANADYSPLAZIE TYPU 1 U ČESKÝCH SOUROZENCŮ

METAPHYSEAL ANADYSPLASIA WITH VARUS FEMURS AND SMALL KNEECAPS: A NEW VARIANT OF METAPHYSEAL ANADYSPLASIA TYPE 1 IN CZECH SIBLINGS

Vážná Anna[5, 1], Mařík Ivo[1, 2], Zemková Daniela[1, 4], Krulišová Veronika[3], Zděnka Vlčková[3], Hrušková Lucie[3], Paszeková Helena[3], Michalovská Renáta[3]

1 Centre for Defects of Locomotor Apparatus; Prague; Czech Republic

2 Faculty of Health Care Studies, West Bohemia University, Pilsen, Czech Republic

3 GHC Genetics, Prague, Czech Republic

4 Dept. Of Paediatrics, University Hospital Motol, Prague, Czech Republic

5 Department of Anthropology and Human Genetics, Faculty of Science, Charles University, Prague, Czech Republic

ABSTRACT

The authors present the clinical and radiological findings of two siblings in whom two previously unpublished heterozygous cis-variants (c.236T>G and c.251A>C) in the *MMP13* gene were identified by genetic testing. We propose to use the name "Metaphyseal anadysplasia with varus femurs and small kneecaps" for this newly described variant of metaphyseal anadysplasia type 1.

Key words: disproportionate short stature; short legs; metaphyseal dysplasia; metaphyseal anadysplasia type 1 (MANDP1); new variants of *MMP13*; small patella

INTRODUCTION

The authors present the clinical and radiological findings of two siblings in whom two previously unpublished heterozygous cis-variants in the *MMP13* gene were identified. The mutations in heterozygous status in the *MMP13* gene are the cause of **Metaphyseal anadysplasia type 1 (MANDP1)**. MANDP1 is heterogeneous genetic bone disease described in literature (**2, 4, 9, 12, 13**).

MANDP1 is characterized by early mild bowing of the femora and tibiae, discrete rhizomelic micromelia, distended anterior rib ends and long bone joints. Metaphyseal widening and irregularity of the long bones mimic those of rickets and are most pronounced in infancy and toddlerhood when we observed transient growth retardation.

Metaphyseal dysplasia regresses spontaneously with growth and usually disappears by the age of 6 years (**9**). The varosity of the femurs, and the rhizomelic shortening of the limbs may persist into adulthood. Catch up growth occurs together with regression of the skeletal abnormalities. The adult height may remain below target height.

MOLECULAR BASIS AND PATHOGENESIS

Metaphyseal anadysplasia can be caused by mutations either in *MMP9* or *MMP13* genes (**6, 7**). These genes are responsible for the production of matrix metalloproteinases, which play an important role in bone formation during growth.

Homozygous loss of function of *MMP9* gene is associated with the mild recessive form of metaphyseal anadysplasia (MANDP 2). Bialelic mutations in *MMP13* gene are associated with recessive metaphyseal dysplasia, Spahr type (**1, 8, 9, 10**). Heterozygous mutations in *MMP13* result in dominant form of metaphyseal anadysplasia (MANDP 1) and spondyloepimetaphyseal dysplasia, Missouri type, that tends to be more severe (**1, 3, 7, 9)**.

Matrix metalloproteinase 9 belongs to Gelatinases family and is involved in the metabolism of collagen. Matrix metalloproteinase 13 catalyses the degradation of extracellular matrix (ECM) components in the growth plate and cleave and release biologically active molecules stored in the ECM. MANDP 1 is associated with dominant-negative *MMP13* mutations that suppress activity of both *MMP9* and *MMP13*. These proteases cleave most efficiently especially collagen type II which is involved in bone formation during childhood (**2, 6, 11).**

MANDP1 (AD) belongs to the spectrum of metaphyseal dysplasias connected with *MMP13* that also comprise spondyloepimetaphyseal dysplasia, Missouri type – SEMDM (AD) and metaphyseal dysplasia, Spahr type (AR), MANDP2 (AR). Their common characteristic is regression with age. With the most common metaphyseal bone dysplasia, Schmid type (AD; gene *COL10A1)*, these dysplasias are classified as group 11 according to the Nosology and Classification of Genetic Disorders of the Skeleton, 2019 revision (**5**) with several other well-defined metaphyseal dysplasias which are not regressive.

CASE REPORTS

Two siblings with mild short stature (-1.6 and -1.9 SD) with markedly curved lower extremities, showing so-called rhizomelic shortening, were examined at our department – see **Fig. 1a–h.** In both

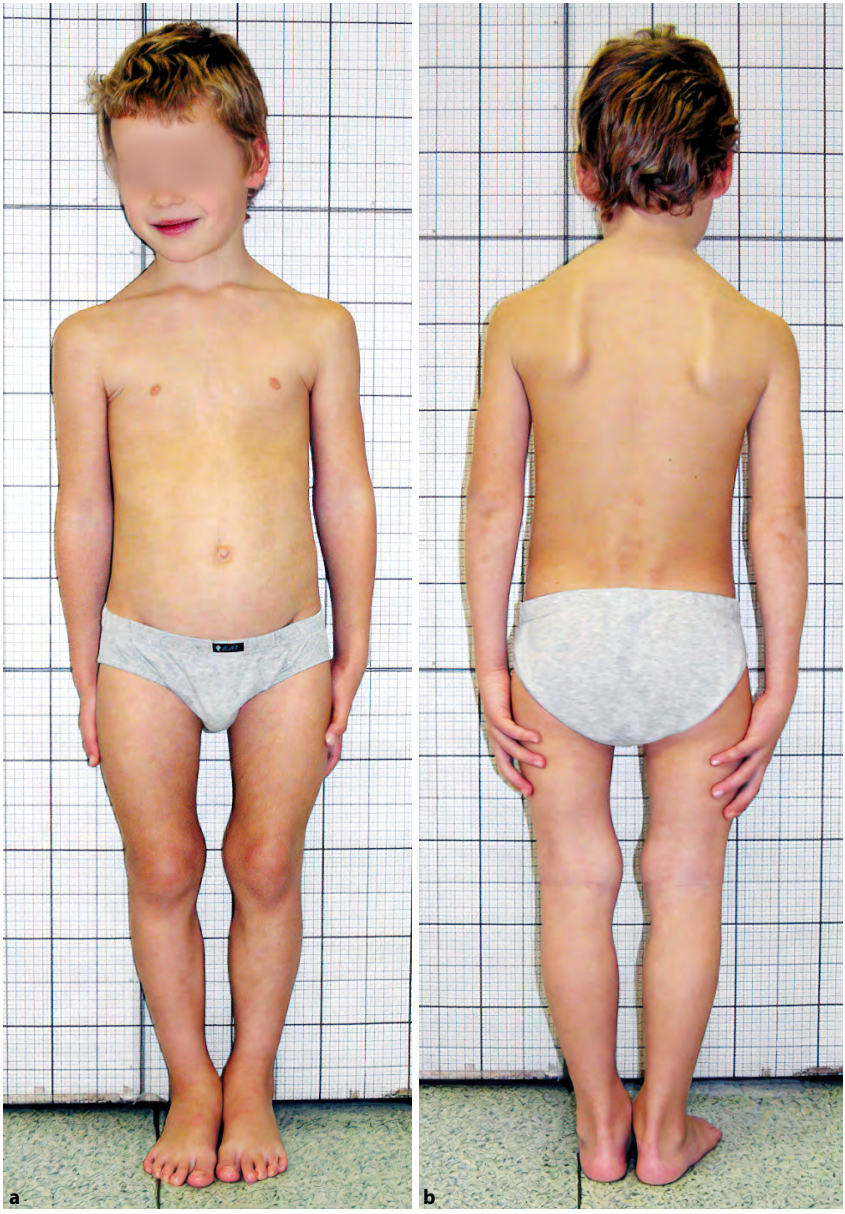


Fig. 1a–h. Phenotype of siblings: stature of a boy in 5 years and 9 months (**1a, b**) and at 9 years and 9 months (**1c, d**). Stature of a girl at 5 years (**1e, f**) and at 9 years (**1g, h**).

Fig. 1a, b. boy 5 y. 9 m.

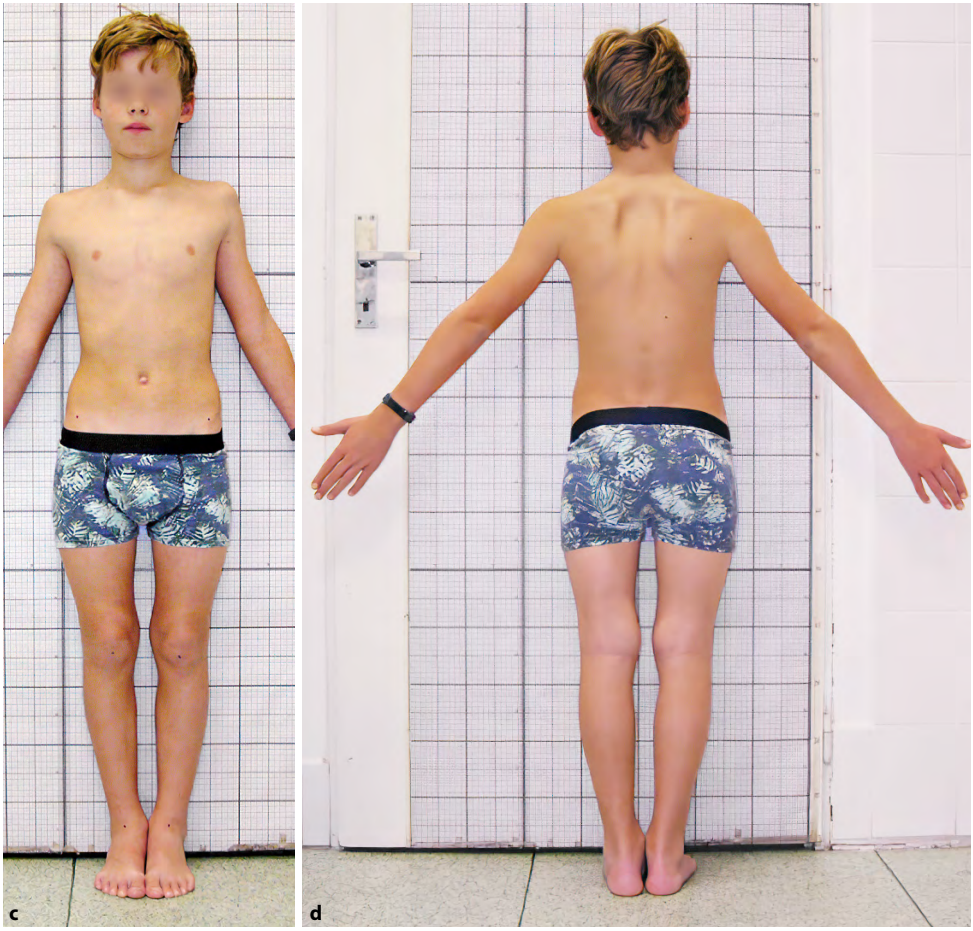


Fig. 1c, d. boy 9 y. 9 m.

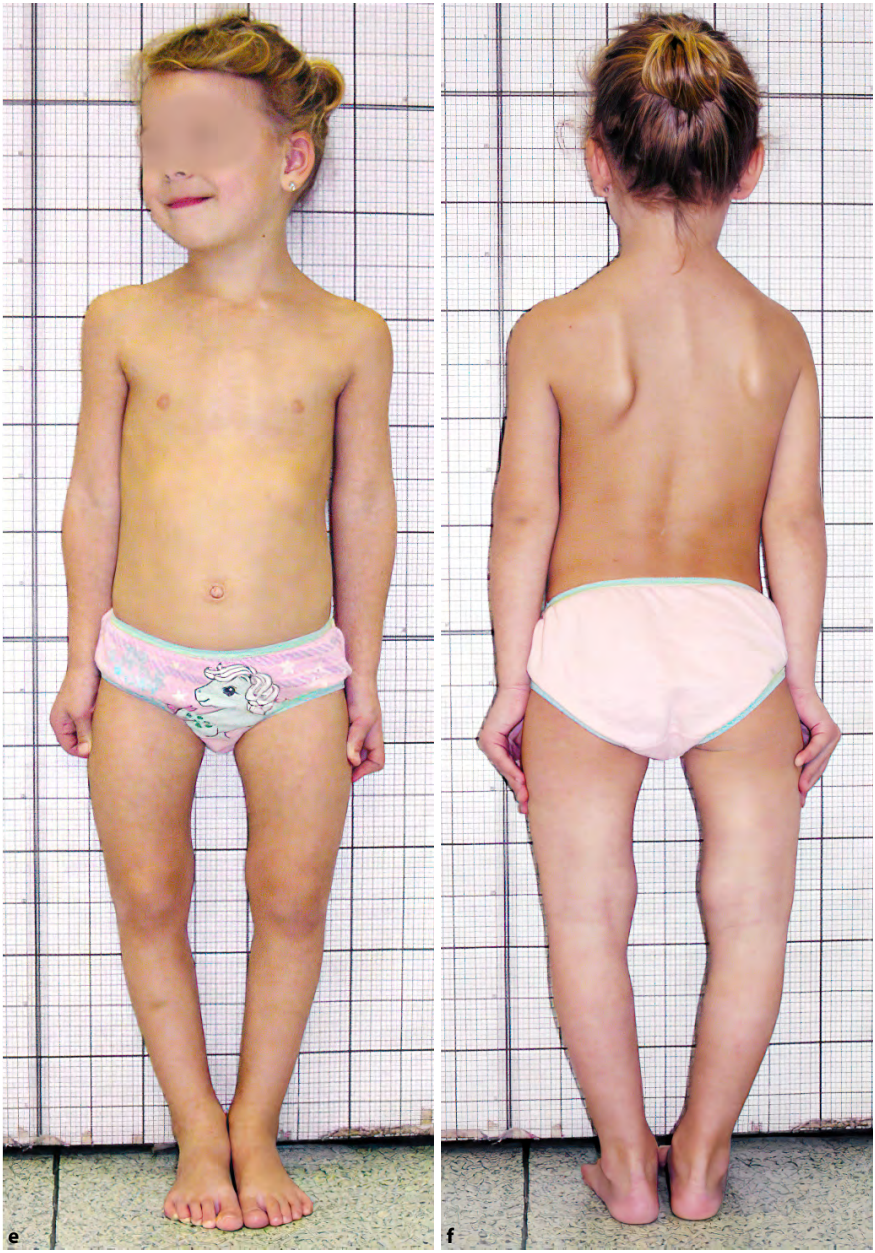


Fig. 1e, f. girl 5 y.

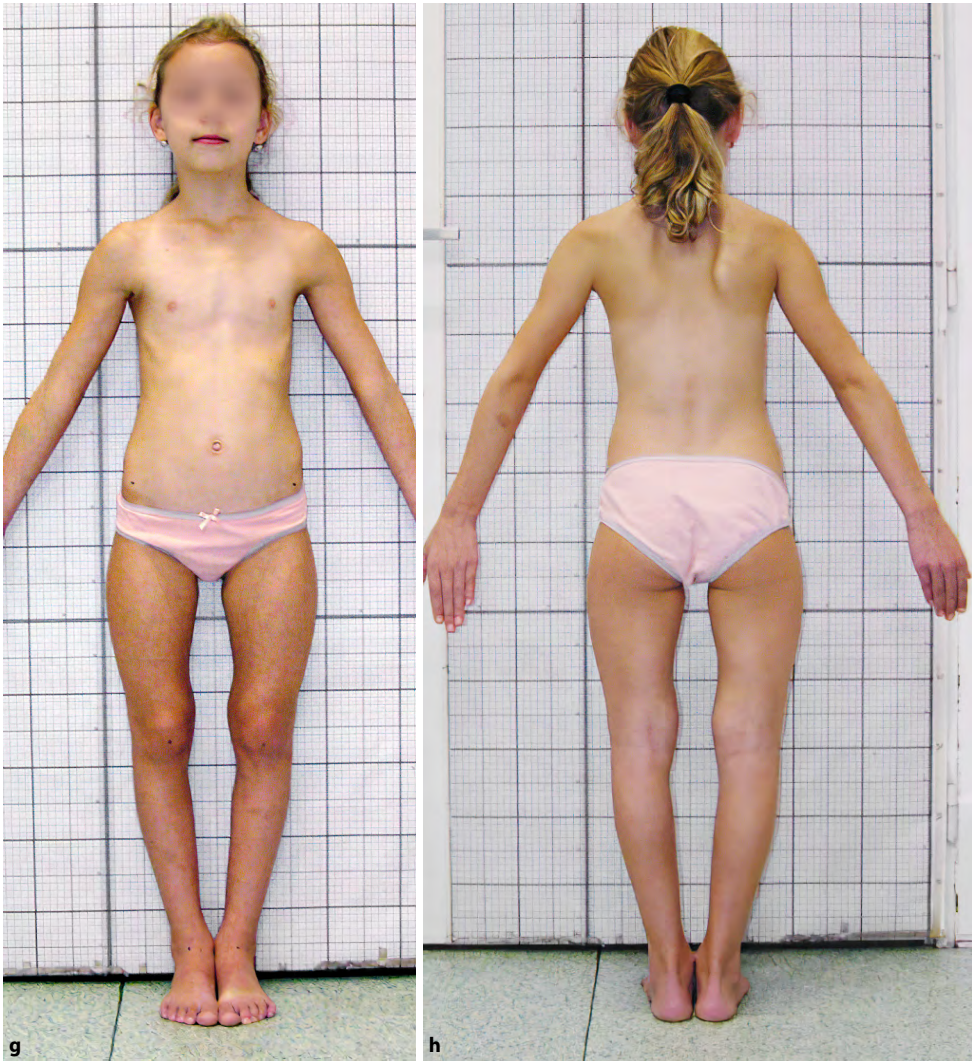


Fig. 1g, h. girl 9 y.

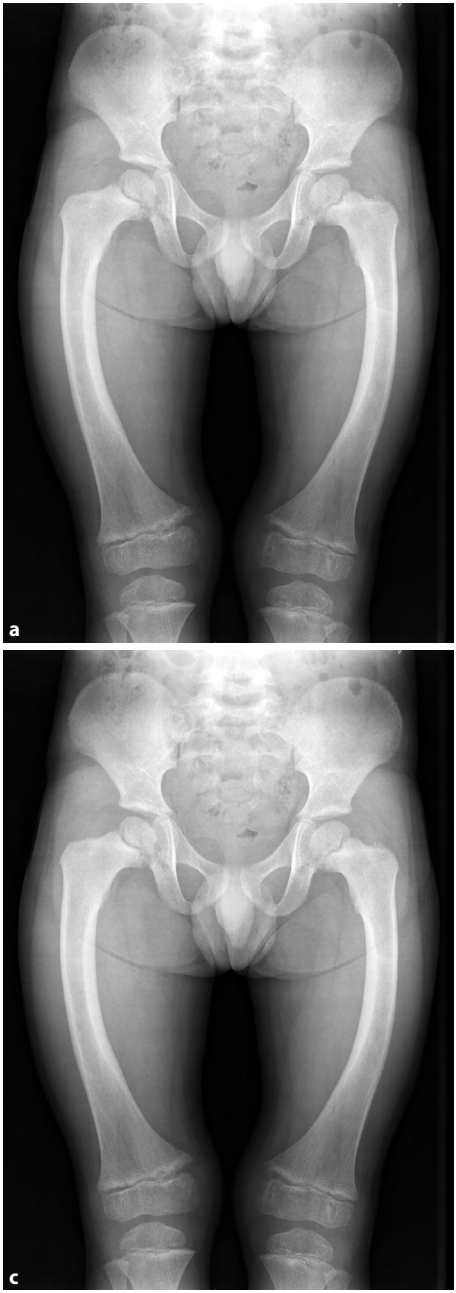


Fig. 2a–c. X-rays of lower limbs of a boy at of 5.5, 5.75 and 7 years of age showing cupped metaphyseal margins of the proximal femurs, enlargement of the growth plates around the knee joints and enlargement of the metaphyses.
a: boy 5 y. 6 m.
b: boy 5 y. 9 m.
c: boy 7 y.

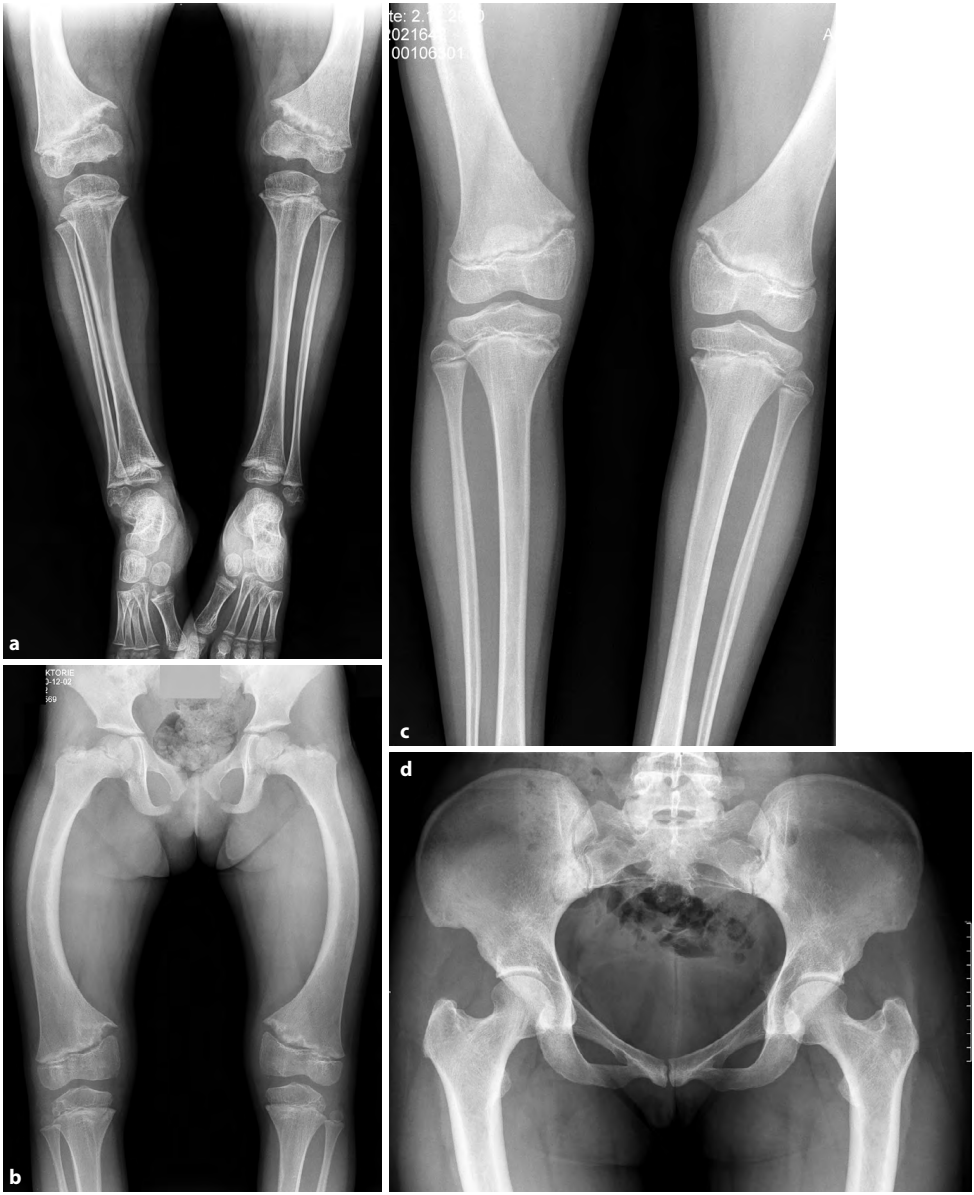


Fig. 3a-d. X-rays of lower limbs of a girl aged 3.5, 5.5 and 9 years show enlargement of the growth plates around the knee joints and a striking fraying and irregularity of the distal margins of the femoral metaphyses (**a**), which improves with age (**b, c**). Mother's pelvis (**d**). X-ray examination of the pelvis and femurs of the children and pelvis and hips of their mother showed wide pelvic blades and varus curvature of the proximal third of the diaphysis of both femurs (**2a, c, 3 b, d**).

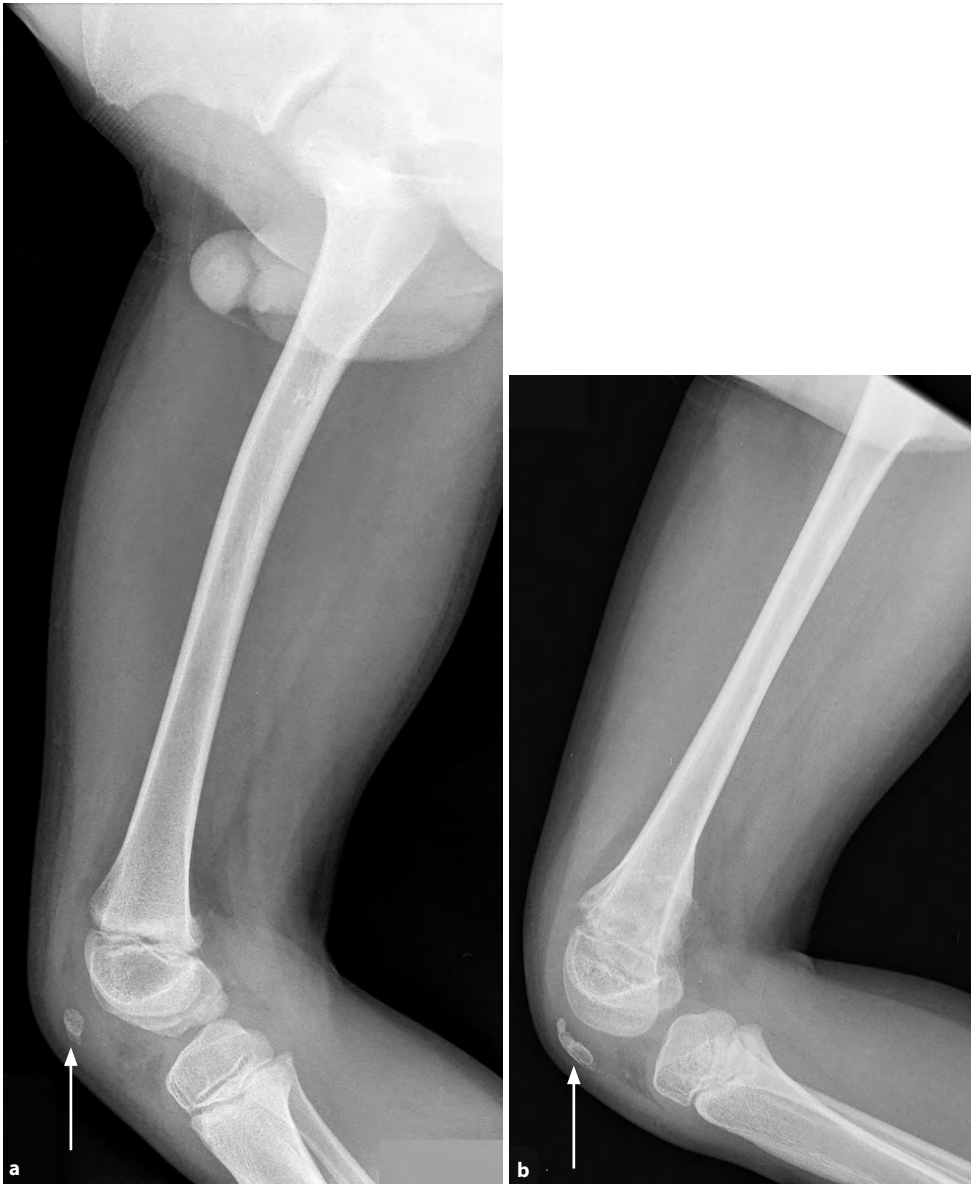


Fig. 4a, b. Lateral films of the knee joints verified very small ossification centres of the patellae in a 7-year-old boy (with fragmentation) and a 5.5-year-old girl.

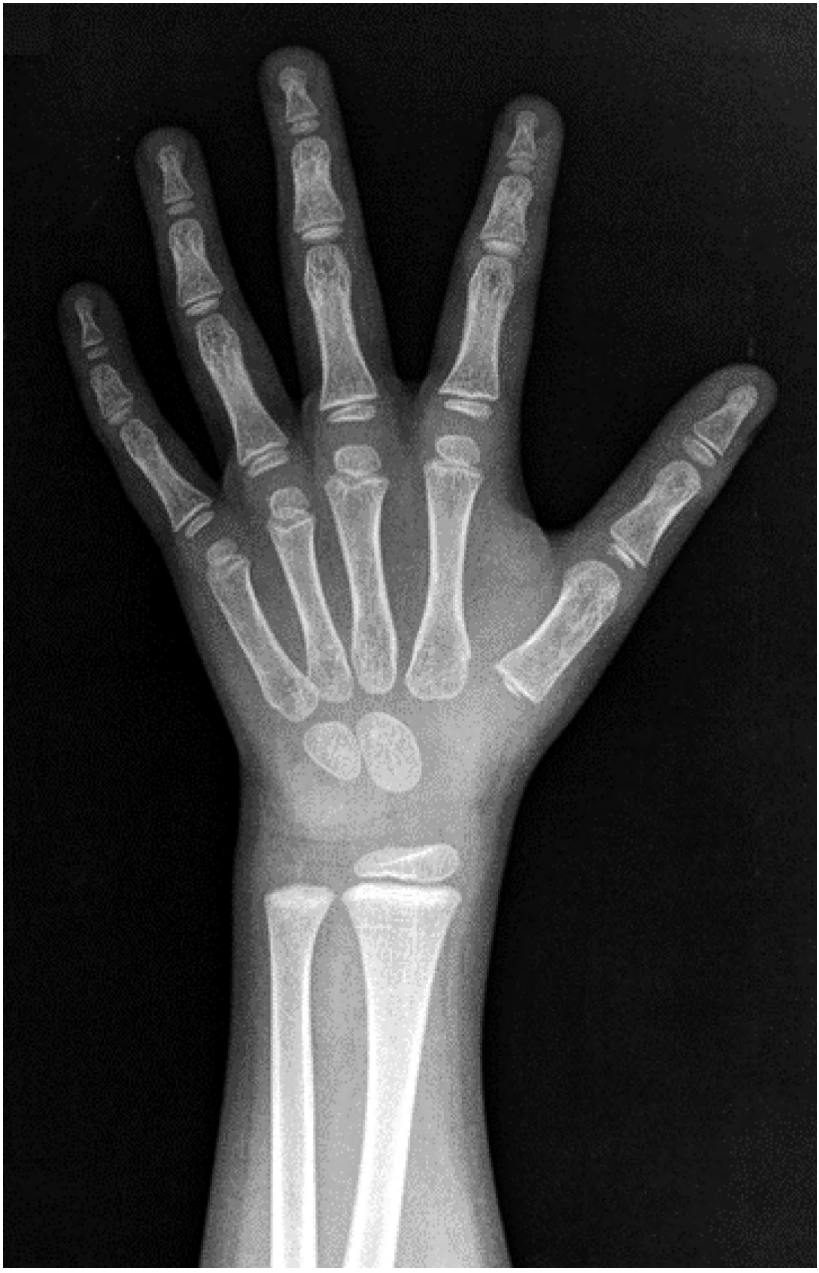


Fig. 5. X-ray of the left hand of a boy aged 5.5 years: Bone age determined by TW3 method: RUS 6.9 years, Carp 2.0 years

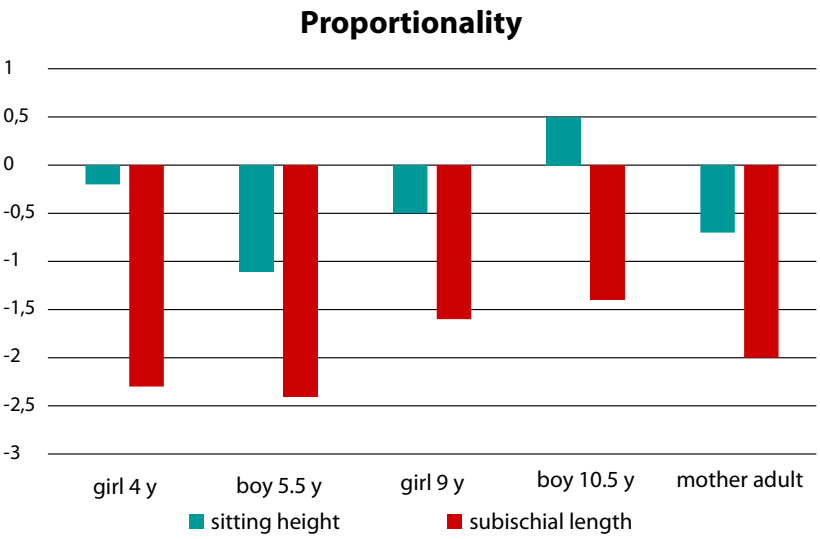


Fig. 6. Proportionality plot showing regression of disproportionality

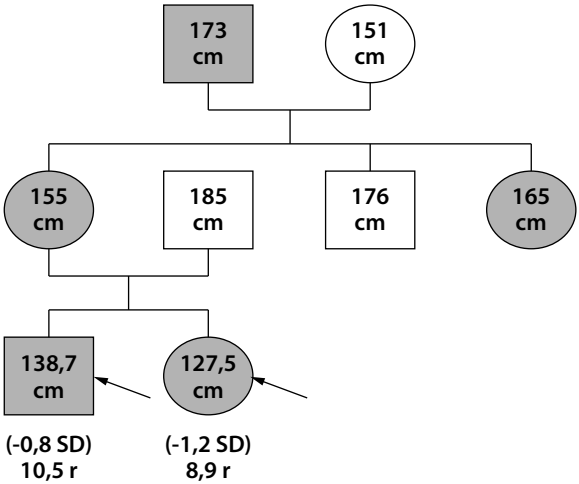


Fig. 7. Family tree

children, rickets was excluded biochemically and the most common metaphyseal dysplasia, Schmid type, was excluded by clinical, anthropological and radiological examination. At the first visit of the boy at 5.5 years and of the girl at 3.5 years, and during visits later, the X-ray examination showed cup enlargement of the metaphyses, varus curvature of the proximal third of the diaphysis of both femurs. (**Fig. 2a-c** and **Fig. 3a-c**). In the girl, there was a striking fraying of the distal metaphysis of the femur (**3a**). Small ossification centres of the patellae (in the boy with fragmentation) was proved at lateral films of the knee joints – see **Fig. 4a, b.**

In both siblings we found short statures near the lower limit of normal with significantly shortened lower limbs and normal hands and feet length. Bone age assessment by TW3 revealed delayed ossification of the carpal bones compared with ossification of radius and ulna and short bones of the hand in the children – **Fig. 5**.

In both siblings, there was regression of the disproportionality, but it remains evident – see **Fig. 6**. Growth dynamics improved. Both children still have waddling gait, cupping of the metaphyses, femoral varus, and rhizomelic limb shortening that persists into adulthood. The girl has grade 1 scoliosis by Cobb (improved from grade 2 by corset therapy) and internal tibial torsion which was partially corrected by derotation braces.

Their mother (154.9 cm) showed similar characteristics. The shortening of the lower extremities is borderline. Both mother and her sister (165 cm) had curvature of the lower extremities in childhood, which regressed with growth. X-ray examination of both children and their mother showed wide pelvic blades and varous femoral diaphysis – see **Fig. 2a,c and 3b,d**. Genealogy shows evidence of AD transmission – **Fig. 7**.

CONCLUSION

In the differential diagnosis, we considered the heterozygous form of metaphyseal anadysplasia, where the height of the body ranges from -1 SD to -2 SD, and metaphyseal dysplasia, Schmid type, where the adult height is lower than -3 SD (between 135–160 cm). In both probands, clinical examination verified short stature within the limits of the wider standard with rhizomelic shortening of lower limbs. Radiological examination showed regressive metaphyseal dysplasia with age in both siblings consistent with MANDP1 cases published in the literature, varosity in the proximal third of both femurs, which was also shown on maternal radiographs, and small ossification centres of the patellae.

Two previously unpublished heterozygous variants in the *MMP13* gene, namely heterozygous cis-variants (i.e. both variants on the same allele) c.236T > G and c.251A > C, were described in the family by genetic testing.

Detected variants c.236T > G and c.251A > C in *MMP13* gene are missense variants leading to amino acid substitution (p.Val79Gly, p.Asp84Ala respectively). As these variants were not reported in litera-

ture, prediction programs were used for ascertaining pathogenicity. Most algorithms developed to predict the effect of missense changes on protein structure and function suggest that these variants are likely pathogenic resp. pathogenic and thus having disruptive effect. Furthermore, we can postulate that both cis-variants represent complex allele.

To confirm the pathogenicity, we performed segregation analysis in probands' family. Both variants in *MMP13* gene was found not only in their mother, but also in their maternal aunt and maternal grandfather with skeletal dysplasia – **Fig. 7**. Both variants were excluded in healthy relatives.

Based on above mentioned facts we consider cis-variants c.236T>G and c.251A>C in *MMP13* gene as causal for MANDP1 in probands.

For AD hereditary newly described variant of metaphyseal anadysplasia, type 1 we propose to use the name "**Metaphyseal anadysplasia with varus femurs and small kneecaps**" because of the persistence of varus femurs into adulthood and previously undescribed small kneecaps in both siblings.

Acknowledgements

We thank Prof. Kazimierz Kozlowski, MD, for many years of selfless consultation and all-round support.

REFERENCES

1. BONAFÉ L, LIANG J, GORNA MW, ZHANG Q, HA-VINH R, CAMPOS-XAVIER AB, UNGER S, BECKMANN JS, LE BÉCHEC A, STEVENSON B, GIEDION A, LIU X, SUPERTI-FURGA G, WANG W, SPAHR A, SUPERTI-FURGA A., 2014, MMP13 mutations are the cause of recessivemetaphysealdysplasia, Spahr type. Am J Med Genet A. 2014 May;164A(5):1175-9. doi: 10.1002/ajmg.a.36431. Epub 2014 Mar 19. PMID: 24648384.
2. LAUSCH, E., KEPPLER, R., HILBERT, K., CORMIER-DAIRE, V., NIKKEL, S., NISHIMURA, G., UNGER, S., SPRANGER, J., SUPERTI-FURGA, A., ZABEL, B., 2009, Mutations in MMP9 and MMP13 determine the mode of inheritance and the clinical spectrum of metaphyseal anadysplasia. Am. J. Hum. Genet. 85: 168-178, 2009. Erratum: Am. J. Hum. Genet. 85: 420 only. [PubMed: 19615667]
3. LI, D., WEBER, D. R., DEARDORFF, M. A., HAKONARSON, H., & LEVINE, M. A. 2014, Exome sequencing reveals a nonsense mutation in MMP13 as a new cause of autosomal recessive metaphyseal anadysplasia. European Journal of Human Genetics, 23(2), 264–266. doi:10.1038/ejhg.2014.76
4. MAROTEAUX, P., VERLOES, A., STANESCU, V., STANESCU, R., 1991, Metaphyseal anadysplasia: a metaphyseal dysplasia of early onset with radiological regression and benign course. Am. J. Med. Genet. 39: 4–10. [PubMed: 1867263]
5. MORTIER GR, COHN DH, CORMIERDAIRE V, ET AL. 2019, Nosology and classification of geneticskeletaldisorders: 2019 revision. Am J Med Genet Part A.;1–27. https://doi.org/10.1002/ajmg.a.61366

6. SHARONY, R., BOROCHOWITZ, Z., COHEN, L., SHTORCH-ASOR, A., ROSENFELD, R., MODAI, S., &REINSTEIN, E. 2017, Prenatal course of metaphyseal anadysplasia associated with homozygous mutation in MMP9 identified by exome sequencing. Clinical Genetics, 92(6), 645-648. Doi:10.1111/cge.13020
7. SONG, C., LI, N., HU, X.-Y., ET AL. 2018, A de novo variant in MMP13 identified in a patient with dominant metaphyseal anadysplasia. European Journal of Medical Genetics. doi:10.1016/j.ejmg.2018.11.009
8. SPAHR A, SPAHR-HARTMANN I., 1961, Familial metaphysial dysostosis. Study of 4 cases in siblings. HelvPaediatr Acta. Dec;16:836-49. French. PMID: 13915518.
9. SPRANGER JW, BRILL PW, HALL CH, NISHIMURA G, SUPERTI-FUNGA A, UNGER S., 2018, Metaphyseal chondrodyslasia, Schmid type; Metaphyseal chondrodyslasia, Spahr type; Metaphyseal anadysplasia. In: Bone Dysplasias – An Atlas of GeneticDisorders of Skeletal Development. 4th ed. Oxford University Press: 33–36; 41–43; 44–49.
10. TADROS, S., SCOTT, R.H., CALDER, A.D., HURST, J.A., 2017, Metaphyseal dysplasia, Spahr type; missense MMP13 mutations in two Iraqi siblings. Clin. Dysmorphol. 26, 13–17.
11. TARDIF, GINETTE, PASCAL REBOUL, JEAN-PIERRE PELLETIER A JOHANNE MARTEL-PELLETIER, 2004, Ten years in the life of an enzyme: the story of the human MMP-13 (collagenase-3). Modern Rheumatology [online]. **14**(3), 197–204. ISSN 1439-7595, 1439-7609. doi:10.3109/s10165-004-0292-7
12. WIEDEMANN, H.R., SPRANGER, J., 1970, Chondrodysplasia metaphysaria Dysostosis metaphysaria) – einneuer-Typ?. Z. Kinder-Heilk. 108, 171–186. https://doi.org/10.1007/BF00439884
13. WIEDEMANN, H.-R., 1992, Metaphyseal anadysplasia: observation of a patient from infancy to the fifth decade of life. Dysmorph. Clin. Genet. 6: 123–127. https://pascal-francis.inist.fr/vibad/index.php?action=getRecordDetail&idt=4167524

Corresponding author:
Professor Ivo Marik, MD, PhD
Centre forDefects of LocomotorApparatus, l.l.c.
Olšanská 7, 130 00 Praha 3
Czech Republic
e-mail: ambul_centrum@volny.cz

INFORMACE O SPOLEČNOSTI PRO POJIVOVÉ TKÁNĚ ČLS J. E. PURKYNĚ (SPT)

Vážená paní kolegyně, vážený pane kolego,

dovolujeme si Vás informovat o možnosti stát se členem **Společnosti pro pojivové tkáně** (SPT), která v roce 2004 navázala na plodnou desetiletou činnost Společnosti pro výzkum a využití pojivových tkání vedenou panem prof. MUDr. M. Adamem, DrSc. Posláním SPT je podpora rozvoje výzkumu pojivových tkání, šíření nových poznatků týkajících se všestranných analýz tkání z obecného pohledu, moderních klinických přístupů k diagnostice a léčbě. Dalším posláním SPT je usnadnění styků mezi jednotlivými odborníky navázáním spolupráce s různými vědeckými, odbornými, výrobními a farmaceutickými společnostmi.

Vědecké poznání a aplikace nejnovějších poznatků v klinické praxi nabyly v posledních letech nebývalého zrychlení, a to nejenom v zahraničí, ale i u nás. Tato skutečnost bezprostředně souvisí s kvalitativním rozvojem poznání i v nebiologických vědách a v moderních inženýrských přístupech. Stále více se prokazuje, že vše se vším souvisí – není náhodou, že nové poznatky a objevy vznikají na rozhraní oborů a různých vědních disciplín. Lidská společnost v posledních desetiletích dosáhla nové civilizační kvality – ve vědě a v jejich aplikacích zcela jistě, avšak v morálce a etice ne tak příliš. Biomedicína je v současné době rozsáhlou interdisciplinární vědou, která bez kooperace s jinými vědními obory by byla odsouzena ke stagnaci. Proto cílem SPT je nejenom integrovat odborníky v biomedicíně, ale i v technických sférách.

Prioritní snahou SPT je presentovat odborné veřejnosti a specialistům v klinické praxi nejnovější poznatky v oblasti pojivových tkání. SPT je i společenskou organizací klinických pracovníků, vědců, pedagogů, která si klade za cíl společensky sblížit nejenom pracovníky v aktivní službě, ale i kolegyně a kolegy v důchodovém věku a v neposlední řadě i studenty a mladé doktorandy z vysokých škol, universit a akademických ústavů.

SPT organizuje během každého roku alespoň dvě odborná a společenská setkání, kde vedle odborných přínosů je kladen důraz také na společenské – přátelské diskuse všech vás, kteří nechtějí stagnovat a kteří nechtějí přemýšlet o nových poznatcích izolovaně a osamoceně.

Pro uhrazení nejzákladnějších nákladů na korespondenci se členy společnosti, jejich informovanost a pořádání odborných kolokvií, symposií a společenských odborných setkání byl stanoven **roční členský příspěvek pro aktivní kolegyně a kolegy 200 Kč** a **pro studenty a důchodce 100 Kč.**
SPT vydává časopis Pohybové ústrojí – pokroky ve výzkumu, diagnostice a terapii, do kterého se i vy můžete aktivně zapojit odbornými články a vašimi zkušenostmi. **Pro současné odběratele časopisu PU a další zájemce doporučujeme přihlásit se na http://www.pojivo.cz/en/newsletter/, zadat jméno a e-mailovou adresu, na kterou bude časopis posílán. Na webové doméně SPT ČLS JEP http://www.pojivo.cz/cz/pohybove-ustroji/ naleznete ve formátu PDF všechna jednotlivá čísla a dvojčísla časopisu (včetně Suplement) vydaná od roku 1997 (bezplatný přístup).**
Milí kolegové, nestůjte opodál a připojte se k české inteligenci – v oblasti pojivových tkání, ke které i Vy zcela jistě patříte. V naší krásné české zemi je třeba, aby prameny poznání byly stále živé a permanentně udržované. Poslání každého z nás není náhodné. Jsme velice zavázáni našim předkům, kteří rozvíjeli kvalitu odbornosti v naší zemi. Nepřipusťme útlum vědy u nás. Nenechme se zmanipulovat programovanou lhostejností, vyrůstající z neodbornosti, závisti a z patologického prosazování ekonomicko-mocenských zájmů.

Těšíme se na Vás a na Vaše zkušenosti – přijďte mezi nás!

Za výbor společnosti:

Prof. MUDr. Ivo Mařík, CSc. – předseda
Prof. MUDr. Josef Hyánek, DrSc. – čestný předseda
Prof. Ing. Miroslav Petrtýl, DrSc. – místopředseda
RNDr. Martin Braun, PhD – vědecký sekretář
Ing. Jana Zelenková – pokladník

Přihlášku do Společnosti pro pojivové tkáně ČLS JEP, z.s. najdete na adrese:
http://www.pojivo.cz/cz/wp-content/uploads/2020/02/PrihlaskaCLS_JEP_SPT_form.pdf

Přihlášku do Ortopedicko-protetické společnosti ČLS JEP, z.s. najdete na adrese:
http://www.pojivo.cz/cz/wp-content/uploads/2020/02/PrihlaskaCLS_JEP_OPS_form.pdf

INFORMATION ABOUT SOCIETY FOR CONNECTIVE TISSUES CMA J. E. PURKYNĚ (SCT)

Dear Sir/Madam, dear Colleagues,

We have great pleasure to inform you about the possibility of joining the **Society for Connective Tissues** (SCT) that was established in 2004 in order to continue the ten-year fruitful activities of the Society for Research and Use of Connective Tissue headed by Professor M. Adam, MD, DSc. The activities of the SCT are aimed at supporting the research development in the field of connective tissues, the dissemination of knowledge related to the all-purpose analyses of the tissues in general, and the application of the up-to-date approaches to the diagnostics and clinical practice. Further, the SCT is determined to facilitate contacts between the respective specialists by means of collaboration with various research, professional, production and pharmaceutical companies.

In the last few years, the scientific knowledge and the application of the latest findings in the clinical practice have accelerated on an unprecedented scale, not only abroad, but also in this country. This fact is closely connected with the qualitative development of the knowledge in the non-biological sciences and in the up-to-date engineering approaches. The fact that all things are mutually connected is becoming more and more evident. It is fairly obvious that the new knowledge and discoveries arise on the dividing line between the different fields and disciplines of science. In the last few decades, the human society has reached the new qualities of civilization. This applies, in particular, for the disciplines of science and their applications; however, this statement can hardly be used with reference to the moral and ethical aspects of the human lives. At present, the biomedical science is a wide-ranging interdisciplinary science which, in case of lack of cooperation with other scientific disciplines, would be condemned to stagnation. That is the reason why the SCT is aimed at integrating the specialists both within the biomedical science and within the engineering fields.

The priority objective of the SCT is to present the professional public and specialists involved in the clinical practice with the latest knowledge in the field of connective tissues. The SCT is also a civic society whose aim is to bring people close together by joining members of the clinical staff, researchers and teachers including the retired ex-colleagues and, last but not least, the undergraduates and PhD students from universities and academic establishments.

The SCT is planning to organize at least two professional and social meetings each year. Beside the professional contribution of these meetings, emphasis will be laid on social activities – informal

discussions of all those who do not want to stagnate and who do not want to acquire the new knowledge in solitary confinement.

The annual membership fee is 200 Czech crowns for full workers, and 100 Czech crowns for students and pensioners. This membership fee shall be used to cover the basic costs on correspondence with the members of the Society in order to inform them about organizing colloquiums, symposiums and social meetings.

The SCT is also engaged in publishing of the interdisciplinary journal entitled **Locomotor System – Advances in Research, Diagnostics and Therapy**. You are invited to contribute to the journal writing professional articles, exchanging experience or, simply sharing your opinions. You can find the volumes of Locomotor System journal at http://www.pojivo.cz/cz/pohybove-ustroji/ since 1997 (free of charge). Since 2013 only electronic edition of the journal is available. That is why we recommend to all subscribers and those interested apply at http://www.pojivo.cz/en/newsletter, enter personal data, titles and e-mail address where the journal will be mailed.

Dear Colleagues, do not stand aside (suffering from terrible lack of time) and join the professional people in the field of connective tissues to whom you undoubtedly belong. In this beautiful country, the sources of knowledge should be kept alive and maintained permanently. Our role in this process is not accidental. We are much obliged to our ancestors who had developed the qualities of proficiency in this country. Do not allow the decline of science. Do not let the programmed indifference arising from lack of professionalism, enviousness, and pathological promotion of economic and power interests manipulate us.

We are looking forward to meeting you. We will be pleased if you join us and share your experience with us.

On behalf of the committee of the Society for connective tissues:

Professor Ivo Marik, MD, PhD – chairman
Professor Josef Hyánek, MD, DSc – honorary chairman
Professor Miroslav Petrtýl, MSc, DSc – vice-chairman
Braun Martin, Dr, PhD – research secretary
Zelenková Jana, Eng – treasurer

Membership application form of the Society for Connective Tissues, Czech Medical Association J.E. Purkynje, Prague you can find on the following link:
http://www.pojivo.cz/cz/wp-content/uploads/2020/02/PrihlaskaCLS_JEP_SPT_form.pdf

Membership application form of the Orthopaedic-Prosthetic Society, Czech Medical Association J.E. Purkynje, Prague you can find on the following link:
http://www.pojivo.cz/cz/wp-content/uploads/2020/02/PrihlaskaCLS_JEP_OPS_form.pdf

TÉMATIKA PŘÍSPĚVKŮ

K uveřejnění v časopise Pohybové ústrojí se přijímají rukopisy prací z oblasti pohybového ústrojí člověka, které se týkají především funkce, fyziologického i patologického stavu kosterního a svalového systému na všech úrovních poznání, diagnostických metod, ortopedických a traumatologických problémů, příslušné rehabilitace a léčebné i preventivní péče. Předmětem zájmu jsou týmové práce z oboru dětské ortopedie a osteologie, dále problémy z oboru biomechaniky, patobiomechaniky a bioreologie, biochemie a genetiky. Redakce časopisu má zájem publikovat články kvalitní, vysoké odborné úrovně, které přinášejí nové poznatky, jsou zajímavé z hlediska aplikací a nebyly doposud nikde uveřejněny s výjimkou publikace ve zkrácené formě.

Redakce přijímá původní práce a kazuistiky, souborné články, které informují o současném stavu v příslušných oblastech souvisejících s pohybovým ústrojím a abstrakty příspěvků z národních a mezinárodních konferencí, věnovaných hlavně pohybovému ústrojí. Původní práce a kazuistiky doporučuje publikovat v anglickém jazyce. Rukopisy jsou posuzovány 2–3 oponenty redakční rady. Redakční rada si vyhrazuje právo provádět recenze a drobné úpravy, případně zkrácení rukopisu. Je velmi žádoucí, aby autor reagoval na případné připomínky.

Nevyžádané rukopisy ani přílohy se nevracejí. Redakce si před uveřejněním prací vyhrazuje rovněž právo na určení pořadí umístění v časopise i jazykovou korekturu.

Příspěvky, uveřejňované v časopise, jsou excerpovány v periodických přehledech EMBASE/Excerpta Medica, vydávaných nakladatelstvím Elsevier a Bibliographia medica Čechoslovaca. Při výběru příspěvků k uveřejnění dáváme přednost rukopisům, zpracovaným podle jednotných požadavků pro rukopisy, zasílané do biomechanických časopisů – Uniform Requirements Submitted to Biomedical Journals (Vancouver Declaration, Br. Med. J., 1988, 296, pp. 401–405).

ÚPRAVA RUKOPISŮ

Rukopis se píše v textovém editoru Word ve formátech doc, docx nebo rtf. Na titulní straně uveďte název článku, pod ním jméno autora, případně autorů, úřední název jejich pracoviště a konečně adresu prvního autora. U českých rukopisů uvádějte název článku a pracoviště také v angličtině.

Na další straně uveďte stručný souhrn (do 250 slov), který má informovat o cílech, metodách, výsledcích a závěrech práce, doplněný překladem do angličtiny. Za ním připojte nejvýše šest klíčových slov v češtině resp. angličtině.

Vlastní text je u původních prací obvykle rozdělen na úvod, materiál a metodiku, výsledky, diskusi, závěr a případné poděkování. Souborné referáty, diskuse, zprávy z konferencí apod. jsou bez sou-

hrnu a jejich členění je dáno charakterem sdělení. Před začátky jednotlivých odstavců nevkládejte žádné mezery ani tabelátory, odstavce by měly mít alespoň čtyři řádky.

TABULKY A OBRÁZKY

Tabulky a obrázky doplněné legendou vkládejte do dokumentu na zvláštním listě s příslušným označením nahoře, příp. jako samostatný soubor. Vyobrazení se číslují v pořadí, v jakém jdou za sebou v textu. V dokumentu označte jejich předpokládané umístění v textu. U českých rukopisů uvádějte texty k obrázkům i v angličtině. Obrázky by měly mít rozlišení 150 dpi u perokreseb (schémata a grafy 600 dpi) a uložené jako typ TIFF File (*.tif) nebo JPEG Bitmap File (*.jpg) tabulky a grafy uložené ve formátech Microsoft Excel (*.xls) nebo jako vektorové obrázky ve formátech (*.eps, *.pdf).

Pojmenování souborů

Název souboru by neměl obsahovat znaky s diakritikou a znaky: „.“ „,“ „:“ „;“ „!“ „?“. Pro lepší následnou orientaci v záplavě souborů je vhodné v názvu souboru uvádět **verzi, jméno autora** (bez diakritiky) a **název článku** (bez diakritiky).

LITERATURA

Seznam odkazů na literaturu se připojí v abecedním pořadí na konci textu. Odvolání na literaturu uvádějte ve vlastním textu příslušnými čísly v kulatých závorkách.

V seznamu citované literatury uvádějte údaje o knihách v pořadí: příjmení a iniciály prvních tří autorů s případným dodatkem „et al.“, název knihy, pořadí vydání, místo vydání, nakladatel, rok vydání, počet stran: Frost HM. The Laws of Bone Structure. 4 ed. Springfield: C.C.Thomas, 1964, 167 s.

Citace z časopisů uvádějte tímto způsobem: příjmení a iniciály prvních tří autorů (u více autorů vložte za jménem třetího autora et al., za jmény autorů nemusí být tečky), název článku, název časopisu nebo jeho uznávaná zkratka, ročník, rok vydání, číslo, strany: Sobotka Z, Mařík I. Remodelation and Regeneration of Bone Tissue at some Bone Dysplasias. Pohybové ústrojí, 2, 1995, č. 1:15–24.

Příspěvky ve sbornících (v knize) se uvádí v pořadí: příjmení a iniciály prvních tří autorů, název článku, editor, název sborníku, díl, místo, nakladatelství a rok vydání, strany ve sborníku (knize): Mařík I, Kuklík M, Průžek J. Evaluation of growth and development in bone dysplasias. In: Hajniš K. ed. Growth and Ontogenetic Development in Man. Prague: Charles University, 1986, s. 391–403.

KOREKTURY

Redakce považuje dodaný rukopis za konečné znění práce. Větší změny při korekturách nejsou přípustné. Prosíme, aby autoři pečlivě zkontrolovali text, tabulky a legendy k obrázkům. Pro zkrácení publikační lhůty je možno připojit prohlášení, že autor netrvá na autorské korektuře. V rámci časových možností je snahou redakce všechny příspěvky zaslat autorům zpět k odsouhlasení konečné úpravy prací. Korektury prosím provádějte s využitím korekturních nástrojů v programu Acrobat Reader. Prosíme o co nejrychlejší zpětnou vazbu redakci časopisu.

ADRESA PRO ZASÍLÁNÍ PŘÍSPĚVKŮ

Rukopisy zasílejte na adresu:

Prof. MUDr. Ivo Mařík, CSc.
Ambulantní centrum pro vady pohybového aparátu s.r.o., Olšanská 7, 130 00 Praha 3
Tel.: (+420) 222 582 214, e-mail: ambul_centrum@volny.cz

SUBJECT MATTER OF CONTRIBUTIONS

The journal Locomotor System will publish the papers from the field of locomotor apparatus of man which are above all concerned with the function, physiological and pathological state of the skeletal and muscular system on all levels of knowledge, diagnostic methods, orthopaedic and traumatologic problems, rehabilitation as well as the medical treatment and preventive care of skeletal diseases. The objects of interest are interdisciplinary papers on paediatric orthopaedics and osteology, further object of interest are problems of biomechanics, pathobiomechanics and biorheology, biochemistry and genetics. The journal will accept the original papers of high professional level which were not published elsewhere with exception of those which appeared in an abbreviated form.

The editorial board will also accept the review articles, case reports and abstracts of contributions presented at national and international meetings devoted largely to locomotor system. The papers published in the journal are excerpted in EMBASE / Excerpta Medica and Bibliographia medica Čechoslovaca.

MANUSCRIPT REQUIREMENTS

Manuscripts should be submitted in text editor Microsoft Word in format *.doc, docx or *.rtf. While no maximum length of contributions is prescribed, the authors are encouraged to write concisely. The first page of paper should be headed by the title followed by the name(s) of author(s) and his/her (their) affiliations. Furthermore, the address of the corresponding author should be indicated to receive correspondence and proofs for correction. Papers are reviewed by two (and/or three) reviewers.

The second page should contain a short Abstract(up to 250 words) followed by the key words (no more than 6). The proper text of original paper is laid out into introduction, material and methods, results, discussion and if need be acknowledgement. The reviews, discussions and news from conferences are without summaries and their lay-out depends on the character of communication. The paragraphs should not begin with any spaces from the left margin nor tabs and should contain at least four rows.

ILLUSTRATIONS AND TABLES

Authors should supply illustrations and tables on separate sheets in the document. They should be numbered in the same order as is their desired location in the text. The figures should include the relevant details. Pictures should have resolution min. 150 dpi, drawings and graphs in bitmap resolution 600 dpi. They should be saved as tif or jpg format, tables and graphs in Microsoft Excel or as vector graphics in formats *.eps or *.pdf. Figure legends should be provided for all illustra-

tions on a separate page and grouped in numerical order of appearance. On the back of figures, their number and name of the author should be indicated.

REFERENCES

References must be presented in a numerical style. They should be quoted in the text in parentheses, i.e. (1), (2), (3, 4), etc. and grouped at the end of the paper in alphabetical order.

The references of books should contain the names and initials of the first three authors, with eventual supplement „et al.", title of book, number of edition, place of publishing, name of publisher, year of appearance and number of pages, for instance: Frost HM. The Laws of Bone Structure. 4. ed. Springfield: C.C.Thomas, 1964, 167 p.

The references of papers published in journals should be arranged as follows: the names and initials of the first three authors (eventually after the name of the third author introduce et al.), title of the paper, journal name or its abbreviation, year, volume, number and page numbers, for instance: Sobotka Z, Mařík I. Remodelation and Regeneration of Bone Tissue at Some Bone Dysplasias. Locomotor System 1995: 2, No.1:15–24.

The references of papers published in special volumes (in a book) should be arranged in the following order: names and initials (there do not have to be dots after initials) of the first three authors, title of paper, editor(s), title of special volume (a book), place of publication, publisher, year of publication, first and last page numbers, for instance: Mařík I, Kuklík M, Brůžek J. Evaluation of growth and development in bone dysplasias. In: Hajniš K. ed. Growth and Ontogenetic Development in Man. Prague: Charles University, 1986:391–403.

Manuscripts and contributions should be sent to the Editor-in-chief:

Professor Ivo Mařík, M.D., Ph.D.
Ambulant Centre for Defects of Locomotor Apparatus
Olšanská 7
130 00 Prague 3
Czech Republic
Phone: (+420) 222 582 214
e-mail: ambul_centrum@volny.cz

OBSAH ROČNÍKU 2020

KASUISTIKY

HISTORIE/PALEOPATOLOGIE

ZPRÁVY

CONTENTS OF VOLUME 2020

CASE REPORTS

HISTORY/PALEOPATHOLOGY

NEWS

OBSAH ROČNÍKU 2021

KAZUISTIKY

ZPRÁVY

CONTENTS OF VOLUME 2021

CASE REPORTS

NEWS

Právní služby poskytovatelům zdravotních služeb:

- obchodní právo - založení společnosti, transformace soukromé ordinace na společnost, registrace poskytovatele zdravotních služeb,
- konzultace v oblasti medicínského práva – školení personálu ve věcech vedení a nakládání se zdravotnickou dokumentací, informovaný souhlas pacienta,
- smluvní agenda – nájemní smlouvy, kupní a úvěrové smlouvy, smlouvy o službách,
- smlouvy se zdravotními pojišťovnami – úprava smluvních dokumentů, korekce plateb,
- otázky náhrady škody na zdraví a z titulu zásahu do osobnostních práv – konzultace vznesených nároků, jednání s pacienty, zastupování v soudním řízení,
- a všechny další otázky, s nimiž se poskytovatelé zdravotních služeb v praxi setkávají

V případě zájmu o nezávaznou konzultaci a poskytnutí bližších informací nás neváhejte kontaktovat.

SVOBODA & KUČERA | ADVOKÁTI

AK Kučera s.r.o. | Stavitelská 1099/6, 160 00 Praha 6 | T +420 222 965 861 | M +420 731 510 675 | E kucera@sk-advokati.cz | W www.sk-advokati.cz

PROTEOR CZ s. r. o. – nestátní zdravotnické zařízení
Ostrava | U Parku 2/2720 | 702 00 Ostrava | tel.: 596 139 259, 596 139 295
Provozovna Olomouc | Mošnerová 7/1184 | 779 00 Olomouc | tel.: 585 414 776, 585 414 823
Provozovna Brno | Milady Horákové 50 | 602 00 Brno | tel. 733 184 083
E | ostrava@proteorcz.cz | olomouc@proteorcz.cz | brno@proteorcz.cz | **www.proteorcz.cz**

lékařská péče v oborech ortopedie a ortopedická protetika ● zdravotní péče v ortotice a protetice ● konsilia pro zdravotnická zařízení ● výjezdová pracoviště v kraji ● zakázková činnost pro zdravotnická zařízení ● skoliotická poradna pro léčbu skolióz páteře mladistvých ● aplikace a výroba individuálních ortopedických vložek pro sport ● výroba individuálních zdravotnických prostředků – protéz končetin, ortéz, ortopedických vložek ● podologická poradna pro pacienty s problémy nohou (syndrom diabetické nohy, bolesti nohou) ● specializované centrum pro aplikaci a výrobu myoelektrických protéz horních končetin