Pohybové ústrojí

Pokroky ve výzkumu, diagnostice a terapii

The 23rd Prague-Lublin-Sydney-
-St Petersburg Symposium

Locomotor Apparatus Adaptation II – Interdisciplinary Aspects

November 20, 2021
Medical House, Sokolská 31, Prague, Czech Republic

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.

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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).

Society For Connective Tissues CMA J.E. Purkynje & Society for Prosthetics and Orthotics CMA J.E. Purkynje & Czech Medical Association J.E. Purkynje & Medical University of Lublin & Vincent Pol University in Lublin & Children's Rehabilitation Center of Orthopaedics and Traumatology "Ogonyok", St. Petersburg

invite you for

THE 23RD PRAGUE-LUBLIN-SYDNEY--ST PETERSBURG SYMPOSIUM

Locomotor Apparatus Adaptation II – Interdisciplinary Aspects

The Symposium will be held under the auspices
of the president of the Czech Medical Association (CMA) J.E. Purkynje

Professor Štěpán Svačina, MD, DSc.

&

the honorary president of the Society for Connective Tissues CMA J.E. Purkynje

Professor Josef Hyánek, MD, DSc.

The Symposium will be held at the
Medical House, Sokolská 31, 120 26 Prague 2, Czech Republic, in November 20, 2021

This event belongs to education actions integrated into the life training system of physicians according to professional statute No. 16 of the General Medical Council.

Partneři sympozia

PROGRAMME

SATURDAY, NOVEMBER 20, 2021

8.00–9.00 REGISTRATION OF PARTICIPANTS

9.00 OPENING OF THE CONFERENCE

WELCOME SPEECHES

Professor Ivo Marik, MD, PhD
President of the Society for Connective Tissues, Czech Medical Association J.E. Purkynje

Professor Tomasz Karski, MD, PhD
Honorary member of the Society for Connective Tissues, Czech Medical Association J.E. Purkynje

Braun Martin, RNDr, PhD
Introduction of Professor Dr. Hans Zwipp (Dresden, Germany)

Braun Martin, RNDr, PhD
Introduction of Dr. Piet van Loon (Deventer, Netherlands)

9.20–12.20 | MORNING SESSIONS

9.20 | SESSION I: ADAPTATION OF LOCOMOTOR APPARATUS 1

Chairmen: Mařík Ivo, Krawczyk Petr

Toes flexions test to recognize the functional status of the foot: Examples of pathology: Knowledge since 1971
Test ohýbání prstů k rozpoznání funkčního stavu nohy: Příklady patologie: Poznatky od roku 1971

Karski Jacek[1], Karski Tomasz[2], Beata Slowinska[3], Bartosz Boryga[3] (Lublin, Poland)

[1] *Dr med. Karski Jacek, Medical University in Lublin, Poland, jkarski@vp.pl*

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Classification of Sequelae of Foot and Ankle Syndromes in Adults, Adolescents and Children
Klasifikace následků syndromů nohy a kotníku u dospělých, dospívajících a dětí

Zwipp Hans (Dresden, Germany)
Orthopedic and Traumatology Department of the University in Dresden, Germany

Influence of the weight of the transtibial prosthesis on postural stability and locomotion – final results
Vliv hmotnosti transtibiální protézy na posturální stabilitu a lokomoci – konečné výsledky

Krawczyk Petr[1], Marik Ivo[2], Zemkova Daniela[2], Uchytil Jaroslav[3], Jandacka Daniel[3], Buzga Marek[5], Sykora Ales[1] (Ostrava, Prague & Pilsen; Czech Republic)

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[5] *Faculty of Medicine, University of Ostrava*

The effect of gait school on functional mobility in patients after unilateral transfemoral lower limb amputation due to dysvascular causes
Vliv školy chůze na funkční mobilitu pacientů po jednostranné transfemorální amputací dolní končetiny z dysvaskulární příčiny

Šorfová Monika, Vitnerová Tereza (Prague; Cech Republic)
Department of Anatomy and Biomechanics, Faculty of physical education and sport, Charles University in Prague, Czech Republic

DISCUSSION AFTER EACH LECTURE

10.40–11.00 COFFEE BREAK

11.00 | SESSION I: ADAPTATION OF LOCOMOTOR APPARATUS 2

Chairmen: Kraus Josef, Krawczyk Petr

Detection of mild cognitive impairment during locomotion after stroke
Detekce mírné kognitivní poruchy během lokomoce po cévní mozkové příhodě

Hereitová Iva[1, 2], Krobot Alois[1] (Olomouc & Pilsen; Czech Republic)

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[2] *Faculty of Health Care Studies, West Bohemia University in Pilsen, Czech Republic*

Gait adaptation in children with spastic cerebral palsy
Adaptace chůze u dětí s DMO spastickou diparézou

Kraus Josef (Prague; Cech Republic)
Dept. of children neurology, University Hospital Motol, Prague, Czech Republic

Physiotherapy in geothermal water. Information on water resources in Poland. Knowledge for doctors and patients
Fyzioterapie v geotermální vodě. Informace o vodních zdrojích v Polsku. Znalosti pro lékaře a pacienty

Karski Tomasz[1], Karski Jacek[2], Domagała Marian[3], Karska Klaudia[4], Zimny Jacek[5], Struś Mieczysław[6], Szczotka Krzysztof[7] (Lublin & Łaszczów & Kraków & Wrocław, Poland)

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Evaluation of functional properties of connective tissues based on monitoring of collagen and elastin crosslinks by HPLC analysis
Hodnocení funkčních vlastností pojivových tkání na základě sledování příčných vazeb kolagenu a elastinu pomocí HPLC analýzy

Braun Martin[1], Šupová Monika[1], Suchý Tomáš[1, 2]

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TIME FOR LECTURE INCLUDING DISCUSSION IS 20 MIN.

DISCUSSION AFTER EACH LECTURE.

12.20–13.00 | LUNCH

13.00–17.00 | AFTERNOON SESSIONS

13.00 | SESSION II: RICKETS AND GENETIC RICKETS. ETIOPATHOGENESIS OF SKELETON ADAPTATION

Chairmen: Kutílek Štěpán, Šumník Zdeněk, Mařík Ivo

Not all rickets are the same
Všechny křivice nejsou stejné

Kutílek Štěpán (Klatovy, Czech Republic)
Dept. of Paediatrics; Hospital Klatovy; Klatovy, Czech Republic

Growth of Czech patients with hypophosphatemic rickets (XLH) on conventional therapy
Růst českých pacientů s hypofosfatemickou křivicí (XLH) na konvenční terapii

Zemková Daniela[1, 2], Mařík Ivo[1, 3, 4] (Prague; Czech Republic)
[1] *Centre for Defects of Locomotor Apparatus l.l.c.; Prague*
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[3] *Faculty of Health Care Studies, West Bohemia University; Pilsen, Czech Republic*

Experience with Burosumab treatment in children with XLH: a case report
Zkušenosti s léčbou Burosumabem u dětí s XLH – kasuistika

Šumník Zdeněk, Maratová Klára, Souček Ondřej (Prague; Czech Republic)
Dept. of Paediatrics; 2nd Faculty of Medicine, Charles University and Motol University Hospital, Prague, Czech Republic

Stress fracture in a 33-year-old patient with XLH treated since preschool age with vitamin D3, phosphates and surgery – a case report
Přestavbová zlomenina tibie u 33letého pacienta léčeného od předškolního věku vitaminem D3, fosfáty a chirurgicky – kasuistika

Mařík Ivo[1, 2, 4], Zemková Daniela[1, 3], Myslivec Radek[4, 1], Hudáková Olga[1, 5] (Prague, Pilsen & Pribram; Czech Republic)
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TIME FOR LECTURE INCLUDING DISCUSSION IS 20 MIN.

DISCUSSION AFTER EACH LECTURE.

14.20–14.30 | COFFEE BREAK

14.30 | SESSION III: BONE DYSPLASIAS. ETIOPATHOGENESIS OF SKELETON ADAPTATION

Chairmen: Zemková Daniela, Krulišová Veronika, Mařík Ivo

Differential diagnosis of Czech dysplasia
Diferenciální diagnostika České dysplazie

Zemková Daniela[3], Mařík Ivo[2,4], Hudáková Olga[1], Mortier Geert R[4] , Kozlowski Kazimierz (Prague, Pilsen; Czech Republic & Antwerp; Belgium & Sydney; Australia)

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Exome Sequencing as Diagnostic Tool in Locomotor Apparatus Diseases
Sekvenování exomu jako diagnostický nástroj u vrozených onemocnění pohybového aparátu

Paszeková Helena[1], Michalovská Renáta[1], Hrušková Lucie[1], Krulišová Veronika[1], Vlčková Zděnka[1], Zemková Daniela[2, 3], Mařík Ivo[2, 4] (Prague, Pilsen; Czech Republic)

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[4] *Faculty of Health Care Studies, West Bohemia University; Pilsen, Czech Republic*

Molecularly genetically verified multiple epiphyseal dysplasia, type 4 and type 5: comparison of clinical findings and radiographic features
Molekulárně geneticky ověřená mnohočetná epifyzární dysplazie, typ 4 a typ 5: srovnání klinických nálezů a radiografických znaků

Vážná Anna[1, 6], Zemková Daniela[1, 3], Mařík Ivo[1, 2], Krulišová Veronika[5], Vlčková Zděnka[5], Malíková Marcela[4], Hrušková Lucie[5], Michalovská Renáta[5]

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Register of achondroplasia 2015–2021
Registr Achondroplazie 2015–2021

Pešl Matin, Vereščáková H., Kotrylová J., Krejčí Pavel (Brno, Czech Republic/hondrolali)

Fakultní nemocnice u sv. Anny v Brně, Brno, Czech Republic

TIME FOR LECTURE INCLUDING DISCUSSION IS 20 MIN.

DISCUSSION AFTER EACH LECTURE.

15.50–16.00 | COFFEE BREAK

16.00 | SESSION IV: BIOMECHANICS & PATHOBIOMECHANICS – ADAPTATION OF LOCOMOTOR APPARATUS

Chairmen: Povýšil Ctibor, van Loon Piet, Miroslav Petrtýl

Macrodactyly of hand – etiopathogenesis and therapy: a case report
Makrodaktylie ruky – etipatogenese a léčení: kasuistické sdělení

Mařík Ivo[1, 2], Zemková Daniela[1, 3], Krawczyk Petr[4], Smrčka Václav[5], Povýšil Ctibor[6] (Prague, Pilsen & Ostrava; Czech Republic)

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Sitting as the unspoken accelerator of the "OsteoNeural Pandemic. Staggering fresh data and evidence in (Dutch) youth in the light of classic Orthopedics
Sezení jako nevyslovený urychlovač „osteoneurální pandemie". Ohromující čerstvé údaje a důkazy u (nizozemské) mládeže ve světle klasické ortopedie

van Loon Piet JM[1], Grotenhuis Andre J[2], Soeterbroek Andre M[3] (Deventer, Nijmegen & Oosterbeek, Netherlands)

[1] *Orthopedic surgeon, Posture Network Netherlands, Proktovar Hengelo*
[2] *Em. Professor, neurosurgery Radboud University Nijmegen*
[3] *Analyst, chairman Posture Network, Netherlands*

Neuroradiologic treasures on MRI in discongruent Osteoneural Growth Relations Consequent findings on MRI in case of malalignment of the skeleton (posture and deformities) and the lack of contemporary research
Neuroradiologické poklady na MRI u diskongruentních osteoneurálních růstových vztahů. Následné nálezy na MRI v případě "malalignment" skeletu (špatného držení těla a deformity) a nedostatek současného výzkumu

van Loon Piet JM[1], Grotenhuis Andre J[2], Soeterbroek Andre M[3] (Deventer, Nijmegen & Oosterbeek, Netherlands)

[1] *Orthopedic surgeon, Posture Network Netherlands, Proktovar Hengelo*

[2] *Em. Professor, neurosurgery Radboud University Nijmegen*

[3] *Analyst, chairman Posture Network Netherlands*

TIME FOR LECTURE INCLUDING DISCUSSION IS 20 MIN.

DISCUSSION AFTER EACH LECTURE.

17.00 | CLOSING OF THE SYMPOSIUM AND PLANNING
THE 24TH PRAGUE-LUBLIN-SYDNEY-ST. PETERSBURG SYMPOSIUM
Ivo Marik & Petr Krawczyk & Tomasz Karski & Piet van Loon

18.00 | DINNER

ORGANIZERS OF THE SYMPOSIUM

Professor Ivo Mařík, MD, PhD & Petr Krawczyk, MD & Martin Braun, RNDr., PhD
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Participants will receive the Programme and Certificate of Attendance
Abstracts of lectures will be published in Supplement 2 of the journal Locomotor System, vol. 28, 2021 (electronic version, ISSN 2336-4777, http://www.pojivo.cz/cz/pohybove-ustroji/)

More recent information about the Symposium will be available on the websites:
www.pojivo.cz & www.ortoprotetika.cz

Speech of welcome to

The 23rd Prague-Lublin-Sydney-St. Petersburg Symposium – Locomotor Apparatus Adaptation II – Interdisciplinary Aspects

In Lublin, October 28, 2021

Dear participants of the 23rd Prague-Lublin-Sydney-St. Petersburg Symposium, which will take place on 20th November 2021.

In spite of many difficulties – such as the Corona virus pandemic or our personal problems, we are meeting again in beautiful Prague with our Czech friends and many other colleagues from different countries.

I remember my first meeting and scientific discussion with Prof. Ivo Mařík during the "Orthopaedic Days of Prof. Jan Červenanský" in Bratislava, Slovakia. It was in 1998.

For years – since then – we have been in scientific contact and discussing scientific matters – mostly here in the Czech Republic, but also in Rhodes, St. Petersburg and in Poland – in Lublin, Kozłówka, Sarbinow, Zwierzyniec, Krasnobrod. All these meetings were nice, pleasant and – what is very important – useful for many suffering people – our patients in other countries as well. During the symposia we always learn how best and with the best methods to treat our patients.

I hope that the 23rd meeting in Prague on 20 November 2021 will be pleasant, friendly and scientifically informative for all present.

Prof. Tomasz Karski MD, PhD

Retired Head of Pediatric Orthopedic and Rehabilitation Department
of Medical University in Lublin in year 1995–2009.
Presently, Professor Lecturer in the Vincent Pol University, Lublin, Poland
E-mail: tmkarski@gmail.com
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Ladies and Gentlemen, dear colleagues!

I cordially welcome you all to the **The 23rd Prague-Lublin-Sydney-St. Petersburg Symposium**, which is held under the auspices of the President of the Czech Medical Society (ČLS JEP), Professor Štěpán Svačina, DrSc. and the Honorary President of the Society for Connective Tissues ČLS JEP Professor Josef Hyánek, DrSc.

Due to the worsening epidemiological situation, the organizers prepared both the present and online form of the Symposium.

The decision of foreign speakers to attend the Symposium in Prague in person was influenced by the progression of Covid 19 in the Czech Republic in recent days. On November 13, 2021, Professor Tomasz Karski informed me of a similar situation in Poland. He and his colleagues decided to participate in the 23rd PLSStP online symposium.

The day after, Professor Dr. Hans Zwipp wrote to me, because the Czech Republic had meanwhile been declared a high risk area for Covid19: I am not yet boostered, I have to cancel my personal participation in the symposium, which I am very sorry about. I will try to present my lecture online. However, I will prepare a publication of my topic for the journal Locomotor system.

It is a pleasure to welcome among us Dr. Piet Van Loon from Deventer, the Netherlands, an orthopaedic surgeon who is particularly interested in „movement care" based on aspects of osteovertebral and osteoneural growth relationships according to Milan Roth.

I warmly welcome all colleagues, specialists from different medical disciplines, experts in biomechanics, orthotics, physiotherapy and other participants who are interested in neuromusculoskeletal disorders from different perspectives.

My heartfelt thanks go to my close colleagues Petr Krawczyk, MD and RNDr. Martin Braun, PhD and the experienced team of the Medical House in Prague, especially Mr. Stavinoha and Mr. Ing Šubert, without whose dedication and perfect organization the symposium would not have been possible both in a face-to-face and online form.

I would also like to thank the partners of the symposium, Kyowa Kirin, represented by Ms. Mgr. Petra Lepilová, and Orling s.r.o., represented by its director Mr. Ing. Petr Dušek.

Recently, an interdisciplinary approach to congenital and acquired skeletal deformities has been adopted as the main line of thought to recognize new relationships regarding the etiology, pathogenesis, and ultimately complex and even causal therapy of genetic skeletal disorders. New discoveries are being made at the interface of disciplines. I believe that scientific lectures will expand our knowledge to benefit our affected patients. I wish you to enjoy new scientific information, and I hope you make new friendships which will help us to arrange international and interdisciplinary scientific research.

As at the beginning of every Symposium, let me briefly recall a few moments that I and a few colleagues spent in Prague during conference ADAPTATION – INTERDISCIPLINARY ASPECTS in September 12, 2020.

Take a look at the photos.

Professor Ivo Marik, MD, PhD, FABI

Chief of the Centre for Patients with Locomotor Defects, Prague, CZ
President of the Society for Connective Tissue, CMA J.E.Purkynje
Scientific secretary of the Society for prosthetics and orthotics Czech Medical Association J. E. Purkynje
Chief-Editor of the journal Locomotor System – advances in research, diagnostics and therapy
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HRLM


DIPLOMA

ABSTRACT

TOES FLEXIONS TEST TO RECOGNIZE THE FUNCTIONAL STATUS OF THE FOOT: EXAMPLES OF PATHOLOGY: KNOWLEDGE FROM 1971
TEST OHÝBÁNÍ PRSTŮ K ROZPOZNÁNÍ FUNKČNÍHO STAVU NOHY: PŘÍKLADY PATOLOGIE: POZNATKY OD ROKU 1971

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Key words: foot anatomy, foot pathology, toes flexion test in metatarsal – phalange joints, pain syndromes, physiotherapy

In children, various deformities of the feet can be found – for example: congenital, neurological, post trauma. In adults, foot deformities and pain syndromes can be a result of a changed anatomy of the foot and restricted movement of foot joints. In this lecture / article, we present the deficit of toes flexion in metatarsal – phalange joint and results of this pathology. The problem was discovered in 1971 and its many cases have been observed throughout long years of author's professional activity. In two Journals this problem was published – in 1971 in Poland in *Chirurgia Narządu Ruchu i Ortopedia Polska* (Surgery of Locomotor System and Polish Orthopedics) and in 1985 in East Germany – in *Beitrage zur Orthopädie und Traumatologie* (Contribution to Orthopedics and Traumatology).

Basic important movements of foot are following:

1. In ankle joint – the are dorsal and plantar flexion,
2. In sub – tali / talar joint it is movement of pronation and supination,
3. In metatarsal – phalange joint important is range of plantar flexion of toes.

In material, in the time 1971–2021 we have many thousand of patients – children and adults – with feet problems. The biggest group were patients with insufficiency of the foot or feet because of forefoot deformity, like hallux valgus and limitation of toes flexion.

Only full flexion of toes give / guarantee the proper and pain free gait / walking. Proper anatomy and full movement of all other joints ensure proper adaptation of the foot to the ground and proper comfortable walking.

In the therapy – the basic aim is to receive full and symmetrical movement of all joints of foot / of feet – the best are exercises in geothermal water.

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ABSTRACT

CLASSIFICATION OF SEQUELAE OF FOOT AND ANKLE SYNDROMES IN ADULTS, IN ADOLESCENTS AND IN CHILDREN
KLASIFIKACE NÁSLEDKŮ SYNDROMŮ NOHY A KOTNÍKU U DOSPĚLÝCH, DOSPÍVAJÍCÍCH A DĚTÍ

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Key words: compartment syndrome in children, postischemic syndrome, foot and ankle syndromes, classification of sequelae

The sequelae of an undiagnosed insufficiently treated or unpreventable (by crush injury) compartment or postischemic syndrome, most often after lower leg fracture or popliteal artery injury, are caused by necrosis and contracture of the extrinsic foot muscles. Therefore claw toes, pes equinus or other forms, such as a severe pes equino varus related to the compartment involved will decide the kind of foot deformity. In cases of a combined compartment syndrome of the lower leg and foot, not only the extrinsic but also the intrinsic muscles especially the short flexors are involved, leading to extensive claw toeing of the hallux and the lesser toes as well. In the case of an isolated compartment syndrome of the foot one will see contracted hammer toes most often after open or third degree closed calcaneal fractures. A new classification of all the different deformities of the foot and ankle as sequelae of a compartment and/or postischemic syndrome, is introduced distinguishing **5 degrees of deformity**:

Type 1 Lesser toes contraction (a,b)
Type 2 Big toe contraction (a,b)
Type 3 Lesser toes + big toe contraction (a,b)
Type 4 Pes equinus (a-c)
Type 5 Pes equinovarus (a-c)

Patients Between 1994 and 2006, a total of 66 patients with sequelae of a compartment and/or postischemic syndrome were treated operatively at the Department of Trauma and Reconstructive Surgery of the University Hospital"Carl Gustav Carus" of the Technical University of Dresden. Patients with contract hammer toes after calcaneal fractures were seen most often (n = 26). Another large group of 24 patients suffered from the sequelae of a compartment and/or postischemic syndrome of the extrinsic muscles of the superficial and deeper compartment of the flexor tendons, produc-

ing a severe pes equino varus. Less common (n = 16) were the deformities caused by an isolated compartment syndrome, such as necrosis of the anterior tibialis, long extensor muscles, peroneal muscles or a combined compartment syndrome of the lower leg and foot. Examples of all 5 types of deformity are shown which are seen even in children and adults and how the problems can be solved surgically.

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ABSTRACT

INFLUENCE OF THE WEIGHT OF THE TRANSTIBIAL PROSTHESIS ON POSTURAL STABILITY AND LOCOMOTION – FINAL RESULTS
VLIV HMOTNOSTI TRANSTIBIÁLNÍ PROTÉZY NA POSTURÁLNÍ STABILITU A LOKOMOCI – KONEČNÉ VÝSLEDKY

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Keywords: weight prosthesis, amputation, kinematic analysis, anthropometry, densitometry, oxygen consumption

Introduction

Decision on the type of prosthesis indication is based on the empirical experience of the attending doctors, physiotherapists and prosthetic technicians based on a local assessment of the extent of disability, type of performance on a limb amputation, stump shape, condition muscles, skin cover, any possible pain or tenderness of preserved limb. During the specification of the appropriate type of prosthesis must indicate age, physical fitness, mental level of the patient and the coincidence of other diseases that can negatively influence the prostheses. From a biomechanical aspect the

neglected factor is determining the exact – optimum length of the prosthesis and in particular its weight. For patients with lower extremity amputations is optimal biomechanics of walking critical for the length of the prosthesis. The authors present the results of kinematic measurements in patients with transtibial amputation who were considered to have a prosthesis in the original weight of the amputee segment of the limb.

Patients and methodology

The authors examined two groups of people. The first group consisted of 14 patients with transtibial, amputation, the second control group consisted of 14 healthy people with identical anthropometric parameters. Structure of the patient group with transtibial amputation. The mean age of patients in the group is 53.8 years (aged 28 to 70). In 11 cases amputation was due to trauma, in 2 cases amputation was caused by a tumor. In one patient the cause of amputation complications was in diabetes mellitus. The average lifetime of the patient's prosthesis was 16.8 years.

Determination of the weight of the amputee part of the limb

Patients underwent detailed anthropometric measurements, based on which mass calculations of the amputee segment of the limb were performed. A total of 3 calculations were performed using Zaciorski, Osterkamp and Mozumdar methods. The results of the calculations were compared with Hologic's full-body densitometry, which was critical for determining the weight of the amputated part of the limb.

Assessing the subjective perception of the heavier prosthesis by the patient.
The subjective perception of the patient's load when using the „heavier prosthesis" was evaluated using the Borg's RPE (Rating of Perceived Exercise) scale.

Kinematic and kinetic analysis

Kinetic and kinematic analysis was performed in a biomechanical laboratory with 3 Kistler power platforms and Qualisys Oqus, Sweden with 9 cameras at Diagnostic Centre of Human Movement PdF, Ostrava University, Czech Republic. The group of patiens included patients with transtibial amputation with approximately the same length of the stump. The condition was also the absence of other comorbidities, deformity of the skeleton, and limb movement in the joints of the lower limb. All patiens had the same type of socet liner. Each of the patients studied had a prosthesis – replacement of the prosthetic foot so that all patiens had exactly the same equipment with identical biomechanical parameters 3 weeks before the examination in the biomechanical laboratory. The structure of the prosthesis - bench, static, dynamic alignment was checked, depending on the used footwear when measuring all the probands.

Indirect kolorimetry – oxygen consumption

Laboratory tests determining the values of resting metabolism. Based on the measurement of inhaled oxygen consumption and exhaled carbon dioxide, the method enables to objectively and currently determine the energy output of the client in idle mode and under load. The patient is examined for the tredmilus with a prosthesis and a prosthesis where the weight is added to the weight of the limb segment.

Results

Comparison of methods for measuring the weight of amputated limb parts in patients with TTA. The lowest weights of the hypothetical distal part of the amputated limb were determined using the DXA method, namely 3.11 ± 1.13 kg. Compared to the DXA method, anthropometric models showed significantly higher values of the weight of the amputated part of the limb by calculation by regression equation. The highest weight was given by the equation according to Osterocamp 4.85 ± 0.77 kg, the lowest value was given by the anthropometric model according to Mosumdar. Significant differences between the values measured by the DXA method and the results calculated by anthropometric models were not found only in the Mosumdar model (AMM). In other cases, the differences were statistically significant ($p < 0.001$) and materially significant, the value of Cohenovad was greater than 0.8, which can be considered a significant change.

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.

Evaluation of spatiotemporal parameters of walking

The evaluation of spatiotemporal parameters related to both lower limbs, which relate to walking speed, double support time, stride width and step cycle duration, shows that when comparing walking with prosthesis without weights, walking with prosthesis with weight and walking of a healthy individual there are no significant statistically significant differences. When comparing walking with a prosthesis without weights and walking with a prosthesis with weights, there are no significant statistically significant differences in the duration of the walking cycle. Compared with the control group, there is a statistically significant increase in cycle time in patients with prostheses without weights. When comparing walking with prosthesis with weight and walking of the control group, we do not observe a statistically significant difference, the length of the cycle is close to walking of healthy people.

Spatio-temporal parameters of the amputated limb
When using a prosthesis with a weight, the standing and swing phase time was extended. The duration of the standing phase of a prosthesis with a weight was prolonged statistically significantly compared to walking on a prosthesis without weights.

The swing phase time of the amputated limb when using weights is not statistically significantly different from walking without using weights. When comparing walking on a prosthesis with a weight and a control group of healthy individuals, the duration of the standing and swinging phases is close to the values of the control group and there is no statistically significant difference between these values.

When comparing walking with prosthesis with weight and without weight, there is no statistically significant difference in the duration of the step cycle. Compared to the control group, there is a statistically significant increase in cycle time in patients with prostheses without weights. On the contrary, when comparing walking with prosthesis with weight and walking of the control group, we do not observe a statistically significant difference, the length of the cycle is close to walking of healthy people.

Spatio-temporal parameters of the unaffected limb
Comparison of spatiotemporal parameters of walking in an unaffected limb does not show statistically significant changes in stride length or stride time when walking on a prosthesis with a weight compared to walking on a prosthesis without weights and against walking of healthy persons.

When walking on a prosthesis with a weight, the standing and swinging phases of the unaffected limb were prolonged, but this difference is not statistically significant compared to walking without weights. However, the time of both of these phases is closer to the walking of the control group, when using weights these parameters do not differ statistically significantly from the control group, while without weights the difference is statistically significant compared to the control group of healthy individuals. Thus, it can be stated that there is no significant statistical difference in the group of amputated people compared to the control group, and the duration of the standing and swinging phase of a healthy limb step approaches the walking of healthy people.

The duration of the gait cycle in a healthy limb is not statistically significant when comparing gait amputees without weights and with weights on the prosthesis. When comparing the duration of the step cycle time, there is a statistically significant difference between patients with prostheses without weights and the control group. When comparing walking with prosthesis with weight and walking of the control group, we do not observe a statistically significant difference in the duration of the cycle.

Evaluation of angular parameters
Angular parameters of the hip joint

When walking on a prosthesis using weights compared to walking on a prosthesis without weights, there is an increase in the observed maximum angular values of extension in the standing phase, flexion in the swing phase and adduction in the standing phase on the hip joint of the amputated limb and healthy limb. However, these changes are not statistically significant.

When comparing the angular parameters of the hip joint of a group of healthy people with a group of amputated with and without weights, a statistically significant difference is evident, with healthy individuals having greater flexion in the hip joint at the heel, maximum flexion in the swing phase and abduction in the swing phase.

Angular parameters of the knee joint

The maximum values of knee flexion in the swing phase on the amputated and unaffected limb are close to the parameters of healthy individuals.

The maximum flexion and internal rotation of the knee joint of the amputated limb in the standing phase is statistically significantly smaller in amputated individuals with prostheses with and without weights than in the healthy group.

The observed values of the angle in the knee joint of a healthy limb when walking on a prosthesis with and without weights differ statistically significantly only in the value of the maximum internal rotation in the standing phase, when this weight is higher in the group of healthy individuals.

Evaluation of kinetic analysis

The calculated values of the moments of forces in the knee joints indicate an increased abduction moment in the knee joint of the unaffected limb in the group of amputated weights, which may mean relieving the medial part of the knee joint of the unaffected limb. The values are close to the values of a group of healthy people.

Subjective assessment by the patient
Questionnaire evaluation, Borg RPE scale: 70% of patients rated walking with a heavier prosthesis as a medium load, 30% as a lightweight load. All patients perceived walking with heavier prosthesis as more stable.

Evaluation of energy expenditure
Patients were also tested for oxygen consumption at a constant walking speed with the original lighter prosthesis and the prosthesis loaded to the original limb weight. Oxygen consumption is slightly higher when walking with a prosthesis with a weight. However, when compared to the value of energy expenditure of a group of healthy individuals, the energy expenditure is comparable without a statistically significant difference.

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.

Our findings in the biomechanical experiment show that there is no significant increase in energy consumption when walking with a heavier type of prosthesis. Analysis of the spatiotemporal parameters confirms that walking with a prosthesis with a weight leads to a symmetrical loading of both lower limbs.

The results show that walking with weights is more symmetrical for most spatio-temporal parameters, outside the time of the swing phase. Symmetry index values are up to 4% when walking with weights, out of step length symmetry, which means that walking with weights is more natural in terms of symmetry than when walking without weights and symmetry index values are closer to the control group of healthy individuals.

ABSTRACT

EFFECT OF GAIT SCHOOL ON FUNCTIONAL MOBILITY OF PATIENTS AFTER UNILATERAL TRANSFEMORAL AMPUTATION DUE TO VASCULAR ETIOLOGY
VLIV ŠKOLY CHŮZE NA FUNKČNÍ MOBILITU PACIENTŮ PO JEDNOSTRANNÉ TRANSFEMORÁLNÍ AMPUTACÍ DOLNÍ KONČETINY Z DYSVASKULÁRNÍ PŘÍČINY

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Keywords: Amputation, gait school, prosthesis, risk of falling, physiotherapy, Berg balance scale, Time up and go test, Activities-specific balance confidence scale

Objectives

The aim of this work is to present the effect of gait school in patients after unilateral transfemoral amputation due to vascular etiology. The study evaluate the effect of this program on the ability of functional mobility, risk of falling and subjective balance of confidence in individuals who are included into this program.

Methods

This study is stylized as a quantitative research. Design of this study is quasi-experiment, which means that there is no random division of probands into experimental and control group. The theoretical part summarizes the findings and approaches of expert knowledge of transfemoral amputation. The practical part evaluate the effect of gait school on patients with transfemoral amputation, due to vascular etiology. All probands were hospitalized at the Malvazinky Rehabilitation Clinic in a program of the gait school. The effect of this 3 weeks program under the guidance of professional physiotherapists was conducted. Rehabilitation lasted for 60 minutes every weekday. The effect of this program was evaluated using the Berg balance scale, the Time up and go test and the Activities-specific balance confidence scale.

Results

Statistical data analysis was performed using a non-parametric Mann-Whitney U test and a paired T test. The correlation of selected tests were evaluated by Person's correlation coefficient. The aim of the gait school for patients after transfemoral amputation was successfully achieved, the functional mobility was improved and the risk of falling was reduced. Furthermore, there was no statistically significant effect on subjective confidence in balance of gait school program.

ABSTRACT

DETECTION OF MILD COGNITIVE IMPAIRMENT DURING LOCOMOTION AFTER STROKE
DETEKCE MÍRNÉ KOGNITIVNÍ PORUCHY BĚHEM LOKOMOCE PO CÉVNÍ MOZKOVÉ PŘÍHODĚ

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Key words: stroke, mild cognitive impairment, dual task, gait.

Cognitive impairment after stroke is common and can cause disability with major impacts on quality of life and independence. There are also indirect effects of cognitive impairment on functional recovery after stroke through reduced participation in rehabilitation and poor adherence to treatment guidelines. Mild cognitive impairment entails a high risk of developing dementia. There is growing evidence that cognitive decline results in the deterioration of gait patterns. Though it is commonly associated with the later stages of dementia, a decline in gait performance may also be

detected much earlier. A dual task paradigm might improve the observation of this phenomenon. In this article, we attempt to establish the following: whether there is a distinct profile of cognitive impairment after stroke; whether the type of cognitive deficit can be associated with the features of stroke-related damage; and whether interventions can improve poststroke cognitive performance. There is not a consistent profile of cognitive deficits in stroke, though slowed information processing and executive dysfunction tend to predominate.

ABSTRACT

GAIT ADAPTATION IN CHILDREN WITH SPASTIC CEREBRAL PALSY
ADAPTACE CHŮZE U DĚTÍ S DMO SPASTICKOU DIPARÉZOU

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Key words: spastic cerebral palsy, DMO, acquired deformities of locomotor apparatus, abnormal gait, treatment

Manifestations of neurologic insult develop sequentially: a) primary manifestations – loss of selective motor control, changes in muscular balance, and muscle tone abnormalities; b) secondary manifestations – abnormal growth and development of musculoskeletal system often resulting in abnormal gait; c) manifestations – compensations children adopt to overcome secondary manifestations. The gait abnormalities in children with cerebral palsy are the consequence of contractures across joints, muscle spasticity, and phasically inappropriate muscle action. Though abnormalities involving one of the major joints of the lower extremity will usually have consequences on the function of the other joints, it is possible to recognize certain primary disorders at each joint. Four primary gait abnormalities of the knee have been identified by Sutherland: jump knee, crouch knee, stiff knee, and recurvatum knee. Other types of gait abnormalities are: scissoring, tip-toe, stooping, Trendelenburg, steppage and vaulting gait. In addition in-toeing, internal rotation, adduction, and ankel equinus deformity which most common lower extremity deformity can be seen. Hip disorders and scoliosis are most common in CP. The development of balance control shows a clear progression of the emergence of organized muscle response patterns, with tonic background muscle activity decreasing and phasic bursts of activity. Older children with spastic diplegia have muscle activation patterns typically seen in normal children who are at the pull-to-stand stage of development. Differences in balance control in CP children are due to both CNS deficits and biomechanical changes in postural alignment. There are tissue changes too, with theory of as a possible mechanism for stiffness in patients with central nervous system lesions. The adaptation of spastic muscles may include formation of higher proportion of abnormal crossbridge attachments binding cross bridges, resulting in abnormal stiffness in muscle tissue, which impairs movement. Gait assessment tools include Edinburgh Visual Gait Scale, Dynamic Gait Index, Functional Mobility Assessment, Observational Gait Scale (OGS). Anticipatory postural adjustments (APAs) play an important role

in the performance of many activities. Children with diplegia are unable to generate APAs of the same magnitude as children with typical development and hemiplegia and have higher baseline muscle activity prior to movement. In children with diplegia, COP (center of pressure) is posteriorly displaced and peak acceleration is smaller during bilateral extension compared to children with hemiplegia. Specific treatments to improve gross motor function, ambulation, and mobility may include physical therapy interventions such as gait training, neurodevelopmental therapy (Vojta, Bobath), strength training, and fitness/exercise training, occupational therapy interventions such as task-oriented or context-focused therapy, and constraint-induced movement therapy (CIMT), surgical interventions such as multilevel surgery (including tendon or muscle lengthening, tendon transfers, tendon release), surgery for positional deformities or joint abnormalities, orthopedic interventions such as casting, bracing, and orthotics, medications such as botulinum toxin A (BoNT-A) for lower limb or upper limb treatment, baclofen, and diazepam, other potentially useful treatments such as electrical stimulation, vibration therapy, hippotherapy, or music therapy. Movements of the lower limbs during gait have been analyzed extensively whereas data on upper body movements are scarce. Children with cerebral palsy compensated more for their gait deviations and were less stable. This was expressed by their greater variability in arm movements and increased movements at the thorax. The thorax showed an increased forward tilt with greater range of motion over the gait cycle. The shoulders were more abducted with increased elbow flexion. Gait analysis with the full-body marker set has offered prospects for a better understanding of compensatory mechanisms for the pathological gait pattern in children with diplegic cerebral palsy.

ABSTRACT

PHYSIOTHERAPY IN GEOTHERMAL WATER. INFORMATION ON WATER RESOURCES IN POLAND. KNOWLEDGE FOR DOCTORS AND PATIENTS
FYZIOTERAPIE V GEOTERMÁLNÍ VODĚ. INFORMACE O VODNÍCH ZDROJÍCH V POLSKU. ZNALOSTI PRO LÉKAŘE A PACIENTY

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Key words: deformations, illnesses, orthopaedics, neurology, physiotherapy, therapy in geothermal water.

The period of human life can be divided into three phases. If there is not any primary pathology, a rapid development, proper shaping and growing is observed in new-borns, infants, children and adolescents. Anabolic processes prevail.

The early adult life – from 20 to 50 years of age – is a phase of full activity, good health, with predominance of anabolic processes.

From around the age of 50 / 55, a period of slow domination of catabolic, or regressive, processes begins, leading to dysfunctions and diseases. Hence the suffering of the elderly caused by the diseases of the spine, hips, knees, feet and shoulders. In patients mostly we see: feet problems – valgus or plano – valgus deformity with gait insufficiency, pain syndromes of tarsus (back part of foot), knee instability and pain syndromes in situation of varus of shank and valgus of knee, arthrosis of hips, spondyloarthrosis – mostly in hiperlordosis of lumbar spine or in lumbar left convex scoliosis, entezopathy of shoulders.

These processes can be slowed down or even inhibited to a large extent by the appropriate rehabilitation. The best conditions for therapeutic rehabilitation are in geothermal waters. In the lecture we present the methods of physiotherapy, especially by using of geothermal water.

The beneficial effects of geothermal waters are based on three essential elements:

1. warm water with a temperature of 36 or 38 degrees is an excellent analgesic in pain syndromes, and at the same time an essential anesthetic,
2. water provides – according to Archimedes' law – less weight of body, offering the perfect possibility of kinesiotherapy.
3. minerals in geothermal waters are an important element for tissue reconstruction, for all anabolic, nutritional and regenerative as well as energy processes important in our every day activity, in physical work and in sport.

Information about Rehabilitations Geothermal Centers. It is giving in the lectures & publication the information where there are Geothermal Rehabilitation Centers in Poland and where geothermal waters are potentially easy to obtain. The use of these waters is a collective joint task of Polish doctors, physiotherapists, the Polish Geothermal Association and of Polish people. The therapy in Geothermal Centers in Poland can be profitable not only for Polish patients but also for the suffering persons from surroundings countries.

ABSTRACT

EVALUATION OF FUNCTIONAL PROPERTIES OF CONNECTIVE TISSUES BASED ON MONITORING OF COLLAGEN AND ELASTIN CROSSLINKS BY HPLC ANALYSIS. HODNOCENÍ FUNKČNÍCH VLASTNOSTÍ POJIVOVÝCH TKÁNÍ NA ZÁKLADĚ SLEDOVÁNÍ PŘÍČNÝCH VAZEB KOLAGENU A ELASTINU POMOCÍ HPLC ANALÝZY

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Keywords: connective tissues, collagen, elastin, crosslinks, HPLC

The structural proteins collagen and elastin play a key role in the proper function of organs and the musculoskeletal system in the body of vertebrates and humans. Depending on the type and stress of a particular connective tissue, these proteins are present in different proportions and their content and rate of their degradation varies in many pathological conditions and during the natural aging process. Knowledge of significant changes in connective tissues at the molecular level and detailed characterization of biochemical processes leading to changes in the biomechanical properties of connective tissues and causing failure of their functionality are among the most current tasks and challenges. It also makes it possible to model more precisely the behaviour of specific connective tissue under load or aging, and use these data within the development of advanced types of composite tissue replacements using tissue engineering technologies.

Under physiological conditions, collagen is an extracellular, water-insoluble scleroprotein that makes up 25-30% of all proteins in the mammalian body and approximately 10% of the total human weight. However, during the aging process, collagen is degraded and deficient, and in addition to the typical loss of bone strength, it also affects other organs such as heart, blood vessels, muscles and skin.

Another important fibrous protein that does not dissolve in water and polar substances is elastin. It has significant elastic properties, occurs in the body e.g. in tendons, ligaments, blood vessels and skin, and gives them characteristic elasticity. However, unlike collagen, elastin production ends with maturity and later there is an inevitable gradual loss of elasticity of the tissues in which this protein is located and often therefore leads to their defects. The half-life of elastin is approximately between 50 and 70 years, and in addition to the natural aging process, its accelerated degradation can also be supported by an inappropriate lifestyle.

In this context, a special role plays collagen and elastin crosslinks, i.e. molecules located between the fibrils of these proteins and naturally occurring due to aging and ongoing enzymatic or non-enzymatic reactions in the organism in selected pathological conditions. It is mainly caused by the

presence of oxidative and carbonyl stress, inflammation, glycation and other pathobiochemical reactions. During the formation and accumulation of these crosslinks in connective tissues due to post-translational modifications of proteins, a number of undesirable structural changes adversely affect the biomechanical, biological and functional properties of the affected tissues.

Excessive cross-linking of proteins reduces their elasticity, increases tissue stiffness and fragility, reduces protein and proteoglycan synthesis, and leads to easier degradation by proteases and a higher risk of mechanical damage. Senescence and specific pathological states (e.g. diabetes, cardiovascular disease, renal failure, inflammatory autoimmune diseases, etc.) lead to degradation processes in connective tissues associated with the breakdown of collagen and elastin-based structures and the release of selected crosslinks as their degradation products.

Typical collagen crosslinking molecules present so called advanced glycation end-products (AGEs), represented by the chemically well-defined pentosidine, a non-enzymatic crosslink formed by Maillard reaction, which leads to higher tissue stiffness during its increased accumulation in tissues. Analogously, in elastin there are cross-linking molecules desmosine and isodesmosine which are formed in the presence of lysyl oxidase. Desmosine and isodesmosine crosslink are elastin specific, as they are not found in other proteins at all.

This work focuses mainly on the analytical aspects of detection and quantification of these enzymatically and non-enzymatically formed collagen and elastin crosslinks in connective tissues. It enables the evaluation of the tissue quality and degree of its degradation. For analysis of the above mentioned crosslinks, methods based on reversed-phase high performance liquid chromatography (RP HPLC) were developed and optimized. By combining HPLC system with fluorescence and UV detectors, sufficient sensitivity for quantitative analysis of the monitored crosslinking molecules in real biological samples was achieved.

This contribution describes both the demanding procedure of sample tissue processing and the HPLC methodology for the determination of these important markers of degradation processes in human connective tissues, and presents selected examples of chromatographic separation of these crosslinks.

We expect that quantitative evaluation of the content of relevant collagen and elastin crosslinks present in the analysed tissues can contribute to a better understanding of changes in their mechanical and functional properties and to clarify the mechanism of defect formation in pathologically affected connective tissues.

Acknowledgements

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ABSTRACT

NOT ALL THE RICKETS ARE THE SAME
VŠECHNY KŘIVICE NEJSOU STEJNÉ

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Key words: rickets, vitamin D deficiency rickets, calcium deficiency rickets, X-linked hypohosphatemic rickets (XLH), recombinant human monoclonal antibody to FGF23

Rickets is the clinical consequence of defective bone mineralization in children. Clinical features of rickets include bone pain, and deformities, muscle weakness, profound sweating. Findings include biochemical and radiographic abnormalities (metaphyseal cupping). The diagnosis of rickets is based on personal history, physical examination, and biochemical testing, and is confirmed by radiographs. First evidence of rickets dates back to ancient Egypt around 1500 BC, rachitic changes were also apparent in skeletons originating from ancient Greece and Rome. In 1645–1650 Francis Glisson and Daniel Whistler provided exact description of rickets and hypothesized about its relationship to poor nutrition. Further research, mostly in the early 20th century , revealed that rickets is caused by vitamin D and calcium deficiency. In 1937 Fuller Albright noticed that not all children with rickets responded favourably by healing to high doses of vitamin D, and that excessive hyperphosphaturia was present in these patients.. He postulated a theory about rare heritable form of (vitamin D-resistant) rickets, caused by a circulating phosphaturic factor. In the meantime several subtypes of rickets were discovered, with X-linked hypohosphatemic rickets (XLH) being the most frequently encountered one. Since 1972 the XLH has been treated by orally administered phosphate and calcitriol. Fuller Albright´s theory was proven true sixty years later after its first presentation, during nineties, with the discovery of phosphatonins, in particular fibroblast growth factor 23 (FGF23). Since 2017 biological therapy of XLH is available with burosumab, which is a recombinant human monoclonal antibody to FGF23. Several clinical trials with burosumab have shown its effectivity in the XLH treatment by improving gait, alleviating pain and overal improving quality of life. In conclusion, not all rickets are result of vitamin D-deficinecy and we should be aware of the heritable/ inborn types of rickets as well.

ABSTRACT

GROWTH OF CZECH PATIENTS WITH HYPOPHOSPHATEMIC RICKETS (XLH) ON CONVENTIONAL THERAPY
RŮST ČESKÝCH PACIENTŮ S HYPOFOSFATEMICKOU KŘIVICÍ (XLH) NA KONVENČNÍ TERAPII

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Key words: hypophosphatemic rickets, X-linked hypophosphatemia, human monoclonal antibody IgG1, growth retardation, anthropometric parameters of Czech group

Hypophosphatemic rickets is a rare, genetically determined disease classified in the 26th group of Genetic skeletal disorders. In contrast to rickets caused by vitamin D deficiency, it is characterized by hypophosphatemia and normal serum levels of calcium.

Characteristic clinical features include slow growth, bone pain and bone deformities.

This group of diseases is caused by mutations in various genes involved in regulating renal phosphate reabsorption (***PHEX***, *FGF23*, *DMP1*, *ENPP1, CICN5, SLC34A3*). The X-linked form is most common. Hypophosphatemic rickets is one of the bone dysplasias in which research and a deeper understanding of etiopathogenesis have led to the discovery of more effective treatments.

The aim

The aim of our study was to evaluate the growth of body height and individual body segments of Czech patients with hypophosphatemic rickets treated by conventional methods (oral administration of phosphate and calcitriol).

Methods

We evaluated available anthropometric parameters in a group of Czech patients born 1940 – 2007 and examined during 1988 – 2018 in Ambulant centre for defects of locomotor apparatus or in University Hospital Motol.

29 patients (20 women, 9 men) with clinical-radiological diagnosis hypophosphatemic rickets. In 21 patients the final height is known, 15 patients were longitudinally followed up and treated (phosphates, vitamin D3) during the growth period. 14 patients underwent surgery.

Body height, weight, BMI, sitting height and subischial leg length were compared to norm by means of SD score.

Results

Final height of Czech patients differs significantly from the norm. Males achieved on average **155.7** +/– 10 cm and females **146.4** +/– 10 cm, i.e. -3.5 SD and -3.1 SD, resp.

The difference between males and females was not statisticaly significant.

Final height of patients born before 1980 was **-3.68** +/- 1.8 SD, after 1980 **-2.71** +/- 1.35 SD (n.s.) The substantial growth retardation occurred before 5 years of age. Further worsening of growth dynamics was observed in puberty, however the variability during puberty was considerable. Body proportions were significantly disturbed. In most patients worsening occurred during puberty. Most patients underwent surgical treatment. Most adult patients were obese or overweight.

Conclusion

Patients treated conventionally by phosphate and calcitriol significantly differ from healthy population. **Existing therapy does not prevent growth retardation, disproportional habitus and bone deformities and brings undesirable side-effects (e.g. osteosclerotic changes, hyperparathyroidism).**

ABSTRACT

EXPERIENCE WITH BUROSUMAB TREATMENTIN CHILDREN WITH XLH – A CASE REPORT
ZKUŠENOSTI S LÉČBOU BUROSUMABEM U DĚTÍ S XLH – KASUISTIKA

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Key words: X-linked hypohosphatemic rickets (XLH), recombinant human monoclonal antibody to FGF23, case report

XLH, a disorder of renal phosphate wasting and the most common heritable form of rickets, is caused by loss-of-function mutations in the *PHEX* gene encoding phosphate-regulating endopeptidase homolog X-linked, which results in excess circulating fibroblast growth factor (FGF-23). FGF-23 is the primary regulator of phosphate homeostasis and acts by controlling phosphate reabsorption in the kidney. Excess FGF-23 impairs renal phosphate reabsorption,which leads to hypophosphatemia, and decreases the synthesis of calcitriol. Chronic hypophosphatemia leads to rickets and osteomalacia, which often result in stunted growth, lower-limb deformity, pain, and physical dysfunction that can limit daily activities.

Conventional therapy involves lifelong administration of phosphate solution and active vitamin D. However, this treatment is not effective for important part of patients and can cause significant side effects. An alternative to this treatment is anti-FGF-23 monoclonal antibody with the generic name burosumab which was approved for use in XLH in 2018 – this case report illustrates our experience with this novel therapy.

ABSTRACT

STRESS FRACTURE IN A 33-YEAR-OLD PATIENT WITH XLH TREATED SINCE PRESCHOOL AGE WITH VITAMIN D3, PHOSPHATES AND SURGERY – A CASE REPORT
PŘESTAVBOVÁ ZLOMENINA TIBIE U 33LETÉHO PACIENTA S XLH LÉČENÉHO OD PŘEDŠKOLNÍHO VĚKU VITAMINEM D3, FOSFÁTY A CHIRURGICKY – KASUISTIKA

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Keywords: stress/fatigue fracture of tibia, hypophosphatemic rickets, X-linked hypophosphatemia, pathobiomechanics, conventional therapy, human monoclonal antibody IgG1/Burosumab-twza against FGF23

The aim is to summarize the latest findings on hypophosphatemic rickets in terms of diagnosis, etiopathogenesis and conventional treatment and to highlight the promising treatment with human IgG1 monoclonal antibody (Burosumab-twza) against FGF23 in children with XLH.

Introduction

In 1937, Fuller Albright et al. distinguished hypophosphatemic rickets (XLH) from vitamin D deficiency rickets in infants. The *incidence* of XLH is 1:20,000–25,000 live births. XLH is a serious disease that affects approximately 3,000 children and 12,000 adults in the United States.

Etiology and pathogenesis: The PHEX gene mutations are involved in regulation of phosphate reabsorption. It was localized to Xp22.2-p22.1. Approximately 180 mutations have been identified in PHEX. PHEX is predominantly expressed in bone and tooth cells. Dysfunction of PHEX protein caused by mutation leads to increased fibroblast growth factor 23 (FGF 23) produced by osteocytes by an unknown mechanism (Liu et al. 2006). FGF23 plays an important regulatory function in cell signalling. FGF23 mutations inhibit renal phosphate transport (phosphate reabsorption) and cause renal phosphate depletion and low blood phosphorus levels.

This leads to impaired bone growth, impaired bone mineralisation, altered bone structure, reduced skeletal strength and the development of characteristic skeletal deformities during the growth period and problems with bone mineralisation throughout the patient's life.

Main clinical findings: most children with XLH have growth disturbances, lower limb deformities, bone pain and severe tooth pain. Patients do not suffer from muscle weakness, tetany, and cramps,

unlike those with vitamin D deficiency rickets. Some adults with XLH have permanent problems or complications such as joint pain, impaired mobility, tooth abscesses and hearing loss. Intellectual development is not impaired; adult height ranges from 130-160 cm. The heart-shaped pelvis may be an obstacle to spontaneous childbirth.

Laboratory findings: Elevated serum fibroblast growth factor 23 (FGF23) concentrations, hypophosphatemia (decreased tubular resorption of inorganic phosphate) and hyperphosphaturia, elevated serum alkaline phosphatase (ALP) levels, osteocalcin, bone ALP (BALP), parathyroid hormone (PTH) – tertiary hyperparathyroidism, elevated urinary pyridinoline and deoxy-pyridinoline levels (in recent years the serum marker CTX has been used).

Main radiographic features: mild to moderate rachitic changes:
- wide growth epiphysis, enlarged metaphyses, tubular shape of long bones
- sparse trabecular bone structure and thinner fibrous cortical bone
- varus deformities of the lower limbs and spine (scoliosis/kyphoscoliosis),
- mild chest deformity, heart-shaped pelvis,
- premature closure of growth plates and craniosynostosis of the sagittal suture.
- low bone density in childhood as opposed to generalized osteosclerosis in adults.
- early osteoarthritis and spondylarthritis
- Looser's zones and fractures of long bones in adulthood

Histology: extension of unossified osteoid sutures in trabeculae (osteoid volume more than 5%).

Case report

In the case of a 33-year-old man with hypophosphatemic rickets, the authors demonstrate an example of a stress fracture (pseudofracture or fatigue fracture) of the ventral tibial cortex without reparative reaction after 1.5 years. The man was conventionally treated from 6 years of age (vitamin D3 and phosphate supplementation until adulthood, later only vitamin D3). Corrective osteotomies of both lower limbs were performed in childhood – from 7 to 15 years of age. At the age of 31, he began to complain of pain in the right tibia after prolonged walking and standing.

The radiographs present an atypical fracture of the right tibia in the anterior cortex at 31.5 years and 33 years. No healing was demonstrated after 1.5 years. Pain relief of the affected limb is inadequate with a right lower extremity weight-bearing regimen. Therefore, corrective osteotomy and provision of an intramedullary nail is planned.

In any case, causal treatment with burosumab, i.e. human IgG1 monoclonal antibody (Burosumab-twza) against FGF23 should be indicated in this patient.

Conclusion

Calcium and phosphate metabolism is impaired throughout life in patients with XLH.

Conventional treatment is not sufficient to prevent growth retardation, disproportionality and progression of skeletal deformities during growth period. Deterioration of bone structure and reduction of skeletal strength persist throughout life due to metabolic bone disease.

Long-term supplementation with calcitriol and inorganic phosphate is often accompanied by secondary/tertiary hyperparathyroidism, hypercalcemia, hypercalciuria and nephrocalcinosis. Adult patients suffer from generalized osteosclerosis and early osteoarthritis and spondylarthritis.

The U.S. Food and Drug Administration (FDA) approved Crysvita (burosumab-twza), the first drug approved to treat adults and children ages 1 year of age and older with x-linked hypophosphatemia (XLH), a rare, inherited form of rickets.

"FDA NEWS RELEASE. FDA has approved the first treatment for a rare inherited form of rickets, x-linked hypophosphatemia. For Immediate Release: April 17, 2018".

Several children are currently being treated with burosumab in the Czech Republic. We hope that this promising pharmacological treatment will be available for all children with proven XLH from preschool age and also for adults with XLH in the treatment of fractures, corrective osteotomies, for the treatment of stress fractures and hyperostosis and other bone injuries resulting from metabolic bone disease. Treatment with burosumab will undoubtedly prevent the adverse effects of conventional treatment with vitamin D3 and phosphates, namely nephrocalcinosis and hyperparathyroidism.

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ABSTRACT

DIFFERENTIAL DIAGNOSIS OF CZECH DYSPLASIA
DIFERENCIÁLNÍ DIAGNOSTIKA ČESKÉ DYSPLAZIE

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Key words: Spondyloepiphyseal dysplasia with metatarsal shortening, SED-MS, collagenopathy type II, R275C mutation (p. Arg275Cys), Spondyloperipheral dysplasia

Spondyloepiphyseal dysplasia with metatarsal shortening (SED-MS), formerly Czech dysplasia, is a rare autosomal dominant type II collagenopathy characterized by early onset progressive spondyloarthropathy, osteoarthritis of the hip and knee joints (with or without synovial osteochondromatosis), and normal growth with hypoplasia/dysplasia of the third and fourth metatarsals, in the absence of ophthalmic disorder and cleft palate. Some affected individuals have progressive hearing loss. This dysplasia is probably caused exclusively by the R275C mutation (p. Arg275Cys), which affects the integrity and stability of collagen fibrils. The main aim of the authors is to present their experience and recent literature findings on SED-MS 17 years after the first description of the disease with a focus on differential diagnosis.

The principal differential diagnosis includes Albright's hereditary osteodystrophy, Brachydactyly E, Idiopathic juvenile osteoarthritis, Progressive pseudorheumatoid dysplasia, and other type II collagenopathies, specifically Osteoarthritis with mild chondrodysplasia (OMIM 604864), Spondyloperipheral dysplasia (OMIM 271700) and Stanescu type of Spondyloepiphyseal dysplasia (OMIM 616538).

In two patients originally described as Czech dysplasia, the R275C mutation was not found, but an additional mutation in exons 53 and 54 was detected. Mutations in these exons encoding C-propeptide usually cause Torrence platyspondylic lethal dysplasia (PLSD-T) and Spondyloperipheral dysplasia (SPD). Clinical research has shown that both SPD and PLSD-T represent a phenotypic continuum, as the same missense mutation in C-propeptide can cause either disorder. The phenotypic

spectrum is very broad, ranging from lethal to relatively mild, with moderate to moderately disproportionate small stature. Predicting phenotype and disease course based on genotype can be very misleading, as shown in our cases. **Spondyloperipheral dysplasia** is a rare AD disease with very variable phenotype, typically characterized by platyspondyly, brachydactyly type E changes (short metacarpals and metatarsals), bilaterally short ulna, and usually disproportionately short stature. Our cases provide evidence that there is phenotypic overlap between Czech dysplasia and mild forms of Spondyloperipheral dysplasia. Metatarsal shortening is a part of the clinical picture of both Spondyloperipheral and Czech dysplasia. In contrast, joint and spinal involvement in Spondyloperipheral dysplasia is not progressive in the same degree.

ABSTRACT

EXOME SEQUENCING AS DIAGNOSTIC TOOL IN LOCOMOTOR APPARATUS DISEASES
EXOMOVÉ SEKVENOVÁNÍ JAKO DIAGNOSTICKÝ NÁSTROJ U VROZENÝCH ONEMOCNĚNÍ POHYBOVÉHO APARÁTU

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Key words: clinical exome sequencing, molecular diagnosis, genetic skeletal disorders, rare pathogenic variants

Clinical exome sequencing is a method for comprehensive diagnosis and interpretation of undiagnosed genetic and rare diseases including those of locomotor apparatus, where all the clinically validated protein-coding regions of genes (known as the exome) are screened using Next-generation sequencing technology (NGS).

We performed massive parallel sequencing on NextSeq™ 550 (Illumina) using Clinical Exome Solution (CES v2) kit by Sophia™, followed by verification via Sanger sequencing. The CES panel covers the coding regions (± 5bp of intronic regions) of more than 4,490 genes with known mendelian/inherited disease-causing mutations and spans 12 Mb of target region. SOPHiA DDM™ platform analyses complex genomic NGS data by detecting, annotating and pre-classifying genomic variants such as SNVs, indels and CNVs.

Dedicated features in SOPHiA DDM™ reduce the complexity of determining the clinical significance of genomic variants. Virtual panels and cascading filters restrict the interpretation to subgroups of genes of interest for suspected disease (e.g. Multiple epiphyseal dysplasia, Neuromuscular disorders, Short stature panel, Skeletal dysplasia) or according to patient's clinical features (e.g. according

to HPO known disease-gene associations). Familial variant analysis enables us to quickly identify causative variants by selecting different inheritance modes (de novo, autosomal dominant, autosomal recessive, X-linked) and to shorten the candidate variant list accordingly.

During the last two years we have sequenced and analyzed patients with various clinical features and suspect diagnoses affecting the musculoskeletal system e.g. osteogenesis imperfecta, osteopoikilosis, Desbuquois syndrome, Ehlers-Danlos syndrome, metaphyseal dysplasia Schmid type, metaphyseal anadysplasia type 1 and multiple synostoses syndrome 1. Among these patients, that have been previously considered undiagnosable by common technologies, we have reported several rare pathogenic variants or VUS with relation to rare locomotor apparatus diseases or syndromes applicable to the clinical background.

These results demonstrate the efficiency of clinical exome sequencing approach in performing molecular diagnosis of undiagnosed inherited diseases and syndromes affecting the locomotor apparatus.

ABSTRACT

MOLECULARLY GENETICALLY VERIFIED MULTIPLE EPIPHYSEAL DYSPLASIA, TYPE 4 AND TYPE 5: COMPARISON OF CLINICAL FINDINGS AND RADIOGRAPHIC FEATURES
MOLEKULÁRNĚ GENETICKY OVĚŘENÁ MNOHOČETNÁ EPIFYZÁRNÍ DYSPLAZIE, TYP 4 A TYP 5: SROVNÁNÍ KLINICKÝCH NÁLEZŮ A RADIOGRAFICKÝCH ZNAKŮ

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Keywords: Multiple epiphyseal dysplasia, MED/EDM type 4 and 5, variant of gene SLC26A2 and MTN3

Multiple epiphyseal dysplasia (MED) is generalized **skeletal dysplasia** associated with significant morbidity. The main clinical symptoms are joint pain, joint deterioration, waddling gait, contractures, lumbar hyperlordosis and short stature. Radiological features comprise delayed, irregular mineralization of the epiphyseal ossification centres of whole skeleton and of the carpal and tarsal bones. Epiphyses are usually flattened.

In the past, the division into a more severe Fairbank and a milder Ribbing type was widely used. Currently, MEDs are divided according to **genetic cause**.

Case reports

The authors present two families with MED type 4 and type 5. The aim of the paper is to compare radio-clinical findings.

Case 1

The first family is represented by 6 years girl, daughter of healthy parents, who was referred to our centre due to hip pain. Our clinical-radiological examination showed a short stature of antenatal origin (-1.5 SD), short trunk, varous wide femoral necks, flattened epiphyses, and habitual luxation of patella. Her 21 months old brother has a growth disorder (-3 SD) with short limbs. Radiological examination showed wide acetabulum. Proximal epiphyses of the femurs were not ossified yet. Based on clinical and radiological findings, MED was diagnosed in both children. Their 4years old sister is healthy. Genetic testing has demonstrated in both children homozygous pathogenic variant c.1957T>A p(Cys653Ser, rs104893924) of gen SLC26A2 which is associated with recessive MED type 4. Parent carry one copy of the pathogenic variant. The *SLC26A2* gene provides instructions for making a protein that transports ions, particularly sulfate ions, across cell membranes. It is essential for the normal development of cartilage.

Case 2

The second case is a 6 years boy followed-up for growth failure of prenatal origin (-3.1 SD). The clinical picture was inconspicuous. His mother had total hip replacement at the age of 37 years and her father at 40 and 60 years. We suspected a dominant transmission of cartilage extracellular matrix disorder. Genetic examination revealed probably pathogenic variant of gene MTN3 c.437T>C p(Leu146Pro) in heterozygous state. Pathogenic variants in this gene are associated with MED5 (AD) and osteoarthritis susceptibility 2 (AD) and spondyloepimetaphyseal dysplasia AR. In the meantime, growth hormone treatment (SGA/IUGR) has begun. The boy developed the features characteristic for MED: flexion hip contractures, lumbar hyperlordosis and genua valga. X-ray examination at the age of 7,5 years showed small flat epiphyses of femoral heads, small ossification centres of great trochanters, accentuated lumbosacral lordosis, platyspondyly in the thoracic and lumbar region, sharped edges of metaphyses of femur and tibia at the knee region, small, flattened epiphyses of distal femur, proximal tibia, and fibula bilaterally. Ossification of carpal bones was delayed and irregular. Epiphyses of 2-5th metacarpals were flat, irregular and partially sclerotic. Pseudoepiphyses of the first and second metacarpal bones were present.

Conclusion

Both these genes, MTN3 and SLC26A2, encode proteins which, in different ways, influence structure and integrity of the cartilage extracellular matrix.

The clinical and radiological findings were similar. In AR MED4, other features reminiscent of diastrophic dysplasia are described in the literature: club foot, cleft palate, clinodactyly, double layer patella or ear swelling. In our case with MED 5 spondyloepimetaphyseal changes were outlined.

The majority of individuals with dominant MED have mutation in COMP gene (typ 1). Other cause of MED are changes in genes encoding collagen IX: COL9A1 (type 6), COL9A2 (type 2) and COL9A3 (type 3). The authors discuss the relationships between MED and collagenopathies II and other disorders of cartilage extracellular matrix.

ABSTRACT

REGISTRY OF ACHONDROPLASIA 2015–2021
REGISTR ACHONDROPLAZIE 2015–2021

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Keywords: achondroplasia, bone dysplasia, registry, average growth rate, paternal age effect, achondroplasia complications, c natriuretic peptide

Introduction

Achondroplasia (ACH) is the most prevalent genetic form of nanism caused by an activating mutation in the FGFR3 receptor tyrosine kinase. More than 20 years of research have yielded much insight into ACH's molecular mechanisms. The turning point is the year 2021, the first long-term effective pharmacological therapy is approved, C-natriuretic peptide (Vosoritide/VOXZOGO, Biomarine, San Rafael, CA, USA). However, the opportunity for ACH patients may be limited by a fragmented system of professional monitoring.

Methods and File

The Achondroplasia Registry (ReACH) is the first Central European registry, enrolling patients with skeletal dysplasia since 2015. The register was approved by the Ethics Committee of the Masaryk University and at St. Anna's Hospital. Patient data with ACH was collected through paediatric and

adolescent practitioners, orthopaedics, endocrinologists and other specialists. After informed consent was given, the data was entered in the online TrialDB system and stored in the Oracle 9i database of the Masaryk University. The average age of patients at ReACH is 8.5 +/- 4.8 years. But ReACH also includes data from several adults. The frequency of selected neurological, orthopaedic, or ORL diagnoses was recorded, taking into account the age of reported complication

Results

The pilot group included 51 children, 22 of them girls. A total of 89 measurements were obtained, mainly heights, weights, but also other parameters. The average growth rate (AGV) was calculated, with values without exception in the lower decile for appropriate age. The pilot group partially experienced a paternal age effect (fathers were two years older than the general group), with a total of 43.5% of fathers at ReACH. Only one patient under 11 years of age has undergone orthopaedic limb extension and one patient has received growth hormone treatment. Potential contraindications to approved treatments, such as co-morbidities low blood pressure and renal impairment, were not noted in any patient. ReACH continues to collect data with annual frequency, allowing comparisons of growth in the observational period and after eventual initiation of therapy in treated patients.

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ABSTRACT

MACRODACTYLY OF HAND - ETIOPATHOGENESIS AND THERAPY: A CASE REPORT
MAKRODAKTYLIE RUKY – ETIPATOGENESE A LÉČENÍ: KASUISTICKÉ SDĚLENÍ

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Keywords: Macrodactyly; overgrowth; hand dysplasia; lipofibromatous hamartoma; median nerve; PI3K-AKT; Somatic mosaicism; epiphysiodesis; debulking

Introduction

Isolatated macrodactyly of the hand is a rare severe congenital disease that is usually not inherited. The fingers are abnormally large due to overgrowth of the finger skeleton and soft tissues. Enlargement of one or multiple digits, was described in the literature nearly 200 years ago. Macrodactyly in hand has a preference for the median nerve territory, mainly involving index, thumb and middle finger (**Wu JH et al. 2008**). Most patients suffer from progressive overgrowth. It may present at birth and combine with syndactyly, digital deviation, thenar eminence hypertrophy, palm and forearm hyperplasia (**Cerrato F et al. 2013**).

Etiology and Pathogenesis

Somatic mosaicism PIK3CA and oncogenic AKT1 variants can be found in cases with isolated macrodactyly (**Tian W et al. 2020**).

Clinical findings and picture with apparent territorial soft tissue overgrowth and bone overgrowth are pathognomonic of lipomatosis of nerves (LN) which involves benign fibro-fatty infiltration. Nerve territory-oriented macrodactyly is more common than lipomatous type (**Cerrato et al. 2013**).

Macrodactyly caused by lipofibromatous hamartoma (LFH) of the nervus medianus occurs in approximately one-third of cases (Tahiri Y et al. 2013). The main symptom is enlargement over the volar forearm, wrist or hand, with or without digital hypertrophy, followed by paresthesia. Biopsy reveals abundant mature fat cells and fibrous connective tissue infiltrating between the neural fascicles and the space between the epineurium and perineurium. Magnetic resonance imaging is pathognomonic for the diagnosis of LFH.

Treatment

There are a number of surgical techniques that make it possible to primarily reconstruct the affected fingers rather than amputate.

The median nerve LFH treatment should be focused separately on nerve compression symptoms and macrodactyly. Carpal tunnel release is the mainstay of treatment for neuropathy, and ray or digital amputation, wedge osteotomy, middle phalangectomy with arthroplasty, soft tissue debulking and epiphysiodesis are suggested options in the management of macrodactyly **(Cerrato et al.** 2013, **Tahiri Y et al. 2013).**

The **aim** of this presentation is to show the preliminary result of surgical correction of overgrown phalanges due to LFH by means of drilling epiphysiodesis. The timing of this surgery was based on the anthropometric prediction of overgrowth of the affected hand rays.

Case report

The boy was first examined at the Centre for Defects of Locomotor Apparatus l.l.c. at the age of 11 years and 4 months.

The current disease

Larger 3rd finger was observed from birth, 4th finger from 1 year.

Lymphoscintigraphy at 7 years showed normal arrangement of lymphatic system of both HK from wrists and to subclavian region.

Genetic examnation at 8 years - conclusion: suspection on non genetic cause of isolated limb defect. Normal male karyotype 46, XY was found,

Color Doppler of upper extremity vessels at 8.5 years - conclusion: no evidence of vascular malformation or flow changes in the right upper extremity.

Neurological examination at 9 years - conclusion: post-ADHD (Attention Deficit Hyperactivity Disorder), social immaturity.

Rheumatological examination at 9 years – conclusion: painless swelling of II, III and IV fingers, susp. lymphedema. Short-term lymphatic drainage without effect.

Clinical, radiological and anthropological examination at 11 years and 4 months
Body height is 153.6 cm (0.7 SD), target height is 175 +/-8.5 cm according to parents' height, bone age is accelerated by 1–1.5 years in accordance with the growth curve. Body weight 63 kg (2.3 SD).

Conclusion: obesity, macrodactyly of 3rd and 4th fingers, partial aplasia of large pectoral muscle on the left (Poland's syndrome), mild orofacial stigmatization (epicanths, high palate, malocclusion, larger incisors), Attention Deficit Hyperactivity Disorder (ADHD).

Prediction of the growth of the right hand: the hand will grow 3–3.5 cm by adulthood, with an increase of 1.5 cm in the fingers. Early epiphysiodesis of the 3rd finger phalanges could lead to partial alignment. The growth of the phalanges from the apophyses and into the width cannot be influenced.

Treatment

1st surgery at 11 years 6 months: verification and neurolysis of lipofibromatous hamartoma (LFH) of the median nerve and digital nerves of the 3rd and 4th fingers, carpal tunnel release, reduction of the

lipofibromatous tissue of the palm, 3rd and 4th fingers, drilling epiphysiodesis of the 3rd finger and distal phalanx of the 4th finger.

The surgical wound on the 3rd finger took 5 months to heal. A short skin syndactyly of the 3rd and 4th fingers developed. A 20 degrees contracture of the distal interphalangeal joint was successfully corrected by overnight splinting (Barinka's splint). Flexion of the fingers into the palm of the hand is unrestricted; reading of the 3rd and 4th finger is reduced.

Anthropometric examination at 12 years 9 months was carried out to evaluate further growth of the right hand – conclusion: the length of the right hand is already stagnant, the length of the 3rd finger is almost equal, it cannot be excluded that the left hand will outgrow the right hand by 5–14 mm.

2nd surgery at 12 years 10 months: deepening of interdigital space between 3rd and 4th fingers, excise hypertrophic scars. Primary healing.

The operations and results are demonstrated in photographs and X-rays.

Conclusion

During 1st surgery we proved lipofibromatous hamartoma (LFH) of the nervus medianus which explained pathogenesis of inherited macrodactylia of the right hand.

Magnetic resonance imaging (MRI) should be indicated to confirm the diagnosis of LFH.

We would like to emphasize that the treatment of hand macrodactyly and other congenital limb defects must be solved in close cooperation of specialists in the fields of orthopaedic surgery, hand surgery, orthopaedic prosthetics and rehabilitation, and last but not least, clinical anthropology, radiology and genetics.

The treatment of hand macrodactyly can be a challenging task that should be left exclusively to those trained in congenital hand deformities.

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Scoliosis, COmorbidities) study group. Phenotypic and genetic spectrum of isolated macrodactyly: somatic mosaicism of PIK3CA and AKT1 oncogenic variants. Orphanet J Rare Dis. 2020 Oct 14;15(1):288. doi: 10.1186/s13023-020-01572-9. PMID: 33054853; PMCID: PMC7556951.

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ABSTRACT

SITTING AS THE UNSPOKEN ACCELERATOR OF THE "OSTEONEURAL PANDEMIC". STAGGERING FRESH DATA AND EVIDENCE IN (DUTCH) YOUTH IN THE LIGHT OF CLASSIC ORTHOPEDICS

SEZENÍ JAKO NEVYSLOVENÝ URYCHLOVAČ „OSTEONEURÁLNÍ PANDEMIE". OHROMUJÍCÍ ČERSTVÉ ÚDAJE A DŮKAZY U (NIZOZEMSKÉ) MLÁDEŽE VE SVĚTLE KLASICKÉ ORTOPEDIE

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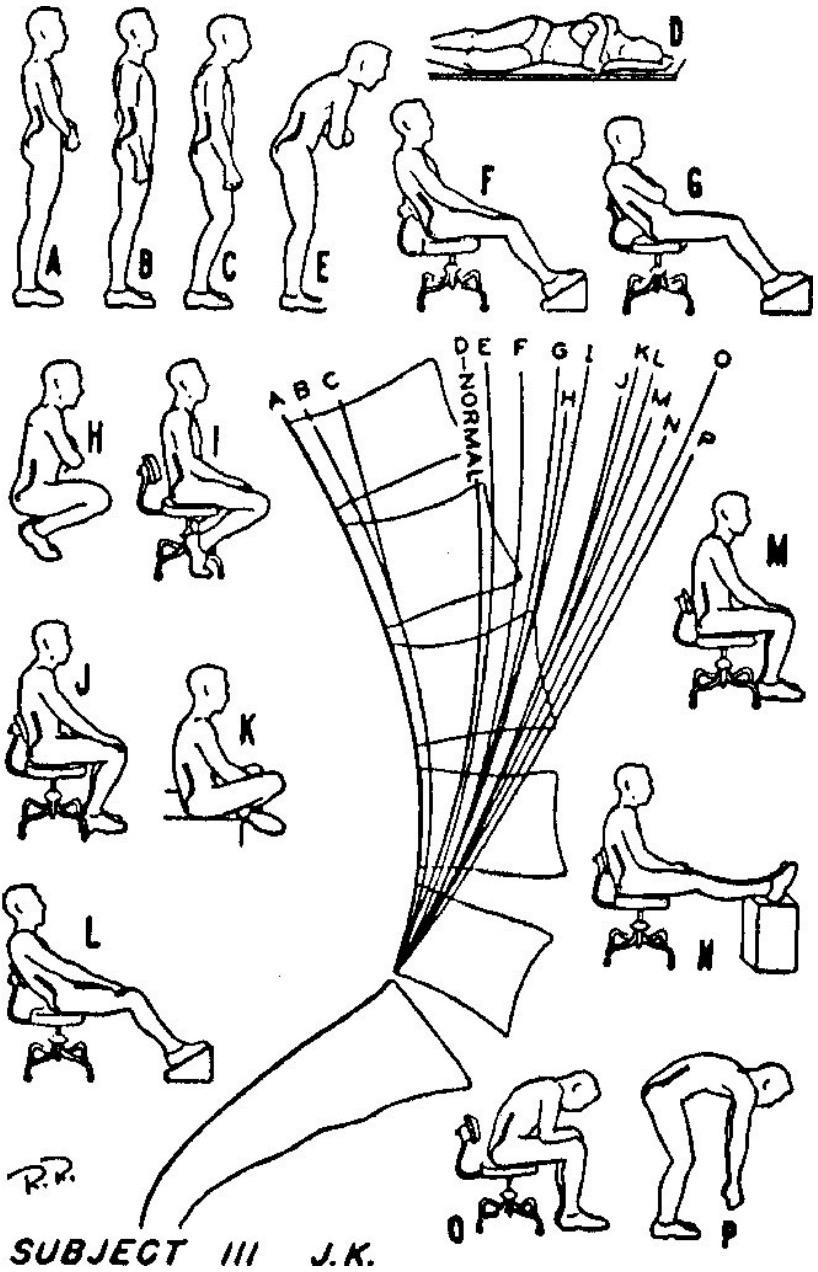
Keywords: sitting of children, main cause of postural detoriation, scoliosis and hyperkyphosis, discongruent osteoneural growth

Introduction of a visualized literature-survey

In the Global Burden of Diseases published in the Lancet (2018) in most "Western" countries the number one and two of greatest socio-economic burdens are Low Back Pain and Arthrosis. Out of the very first book „Orthopaedia" by the French theologist, physician and professor in Paris, Nicolas Andry de Bois-Regard in 1741, sitting of children on chairs was depicted as the main cause of postural detoriation leading to spinal deformities as scoliosis and hyperkyphosis. The iconic logo of orthopedics, the crooked tree must be explained as an allegory on the goal of prevention. He already concepted effective prevention by sitting with a greater angle between the torso and the femur $>>90^0$. Biomechanics and medical knowledge on the relation sitting and spinal deformation in childhood culminated in (German) Orthopedic mastery in prevention as in A. Baginsky's "Handbuch der Schulhygiëne" (1883) in posture protecting school furniture and strong recommendations to provide all children on school with orthopedic based school gymnastics in which "stretching' is main part to reach natural alignment. Malalignment is a very strong factor in etiology of low back pain and arthrosis by inducing overload and shearforces on cartilage and discs.



Keegan; Change of lumbar curve in diferent positions.

Biomechanics of sitting

Leonardo da Vinci was the first to give the biomechanical definition of a natural human posture in standing and sitting: a vertical line between ears, shoulder and hip. In sitting slumped or sloughed all loading forces and tensile forces are in disarray. In 1904 Wüllstein showed in young dogs that prolonged flexion leads to massive damage to discs and cartilage in their spines. In 1974 the studies of the orthopedic surgeon A. Nachemson and B.Andersson were published in which they showed with intradiscal pressure measurements the compressing forces in slumped sitting were higher as even in heavy load lifting! Keegan researched spinal curves in different degrees of sitting.

There is consensus about the negative role of prolonged sitting on the development and maintenance of the optimal form and function of the musculoskeletal system. Harrison et al (1999) in their article "Sitting biomechanics part 1" provide all the elements gathered from the accumulated scientific knowledge on this complex postural set-up. We presume that the basic knowledge, that balancing an upright animal body (humans, penguins, birds, kangaroo's) is based on identical mechanisms and that the only difference in men is their relatively big and unsurpassed weight of their heads, is axiomatic and not under any discussion. Sitting on chairs is quite new in evolution of mankind and especially in children.

Schoberth studied 1035 schoolchildren in sitting positions. In a relaxed posture none could maintain the physiologic lumbar lordosis. At the command to properly sit up, he found that only 30.5% could only keep the lumbar lordosis by muscular contractions. Recent studies

The risk of sitting: anatomical backgrounds

The resilient element or that part of the total elasticity, which is formed by the optimal curvatures in a healthy, or young spine in the standing position is diminished and loses its protective effect when sitting. The intervertebral discs thus lose protection against overpressure in this suspension system, which also determines the normal movement patterns (ROM) of, by a suboptimal or unphysiologically distribution (in apex and magnitude) of lordosis and kyphosis. Keegan shows in his study in 1953 that the least burdensome position for muscles, discs and vertebrae is the posture in which the angle between spine and thighs is 135 degrees.

The fact is that very much children nowadays have no intrinsic "tools" to hold the "protecting lordosis" position for a long time because of their sitting habits. Their propriocepsis and balancing skills were not adjusted and trained and true maintenance is missing.

Discongruent Osteoneural Growth (acc. to Roth) in modern youth

Milan Roth could not foresee that the intensified sedentary lifestyle of children could become the main source of discongruency in the Osteoneural Growth. It is clear, that a lack of exercise with fre-

quent and maximal stretching of all muscles and nerves, as part of a sedentary lifestyle will hinder the stretch growth of nerve cells and by them innervated fascia's, leading to structural tightness of the Central Nervous System and neuromuscular structures.

Preventing kyphosis in the thoracolumbar joint, or maintaining a lumbar lordosis and maintenance of length of neuromuscular structures is therefore obvious of great importance in prevention of many spinal deformities and other musculoskeletal and neurologic disorders.

In the Netherlands there is starting awareness in society, that the youngest generations are heading for a myriad of health problems with their locomotor apparatus, but an orthopedic explanation including the loss of postural guiding is still missing.

Recently the major Dutch sport associations (football, skating, hockey, tennis) warned about the serious declined motor skills of the youngest generations. The most impressive medical evidence came from one of the worlds biggest longitudinal survey in newborns: Generation-R where on MRI in 550 9-year-old children amongst many other findings, in 73% the existence of a "bulging disc" on at least one motion segment was found. Presumed relationship with obesity proofed to be absent.

In preparing preventive solutions the design of sitting devices for children should be altered and the importance of early exercises and guidance to healthy postures restored in education.

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ABSTRACT

NEURORADIOLOGIC TREASURES ON MRI IN DISCONGRUENT OSTEONEURAL GROWTH RELATIONS CONSEQUENT FINDINGS ON MRI IN CASE OF MALALIGNMENT (BAD POSTURE AND DEFORMITIES) OF THE SKELETON AND THE LACK OF CONTEMPORARY RESEARCH

NEURORADIOLOGICKÉ POKLADY NA MRI U DISKONGRUENTNÍCH OSTEONEURÁLNÍCH RŮSTOVÝCH VZTAHŮ. NÁSLEDNÉ NÁLEZY NA MRI V PŘÍPADĚ "MALALIGNMENT" SKELETU (ŠPATNÉHO DRŽENÍ TĚLA A DEFORMITY) A NEDOSTATEK SOUČASNÉHO VÝZKUMU

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Keywords: spinal deformities, MRI, osteoneural relationships, discongruent osteoneural growth relations

Introduction

In spinal deformities the X-ray is still the gold-standard in assessment of the bony structures.

MRI has taken over this position in assessing the non-bony parts of the spine and its neurogenic contents (central cord-root complex). On the spatial relationship of the cord-root complex with the bone and the visualization of present tension or tightness (caliber) in this complex MRI is very informative.

Nevertheless, there is almost no recent study to confirm the problematic anatomic Osteoneural relationships that were found already in the sixties of the last century on the pneumoencephalographies and pneumomyelographies on which the late prof. Milan Roth, neurologist at Purkynje University Brno concepted his Osteoneural Growth Relations. Out of biology and anatomy the Holzer's Neuroprotective Mechanism depicts that any animal in capable during growth to arrange morphogenesis in such a way complete freedom of neural structures from neural structures will be present in adulthood. The human species is the only that can change these physical surroundings and its lifestyle (sedentary) resulting in failure of this mechanism. Constant contact and rubbing and friction during movements between neural tissue and bony structures is a well-known source of pain, loss of isolation and neuropathy and myelopathy and gross patho-anatomy like in Arnold Chiari Malformation and syringomyelia.

In the light of rapid increasing incidence of spinal deformities and certainly spinal pain syndromes (Lancet 2018) and more knowledge on the role of the central cord-root system during the period of

growth a relationship between abnormal morphology on the outside of the body must represent abnormal relationships in the bony canal and v.v.

Method and material

In a series of cases with a spinal pain syndrome or deformation a comparison will be shown of all the diagnostic features available to visualize the relationship between external and internal features in spinal deformations like "bad posture". Besides clinical photographs in standing (and sitting) position and of clinical tests to show the functional signs in postural problems based on neuromuscular tightness, surface topography derived models (Diers GmbH, Schlangenbad, Germany), standing radiographs and MRI are put alongside to explain the relationships between the main systems involved in deformations during growth: the CNS and the musculoskeletal system.

Results

In all cases there is direct relationship between the external form of the spine (posture) and the internal features of the central cord/roots complex. Especially the position and the caliber of the central cord and roots in relation to the osseous boundaries of the canal will be shown and explained. The ever present and in fact pathological contact zones between the cord and roots complex at the apices of pathological curves and at the cervicothoracic and the lumbosacral areas present at early ages will be highlighted and related to physiological and pathophysiological signs and symptoms that can occur during adulthood.

Discussion

No epidemiologic or cohort study is necessary for understanding pathogenesis if biomechanical and neurodynamic knowledge on formation of bone and the peculiar way the nervous tissue grows by stretch is the basic principle. But this visualization of an understandable relationship between external and internal features (almost never described in literature) of the involved tissues can be used by other institutions with a great number of cases and resources for research on a bigger scale to provide also statistical evidence of the relationships. Concomitant features of a "short cord" like Arnold Chiari malformation and syringomyelia can be explained out of the neuro-osseous growth discongruency.

Conclusion

The skeleton and the CNS are strongly related in patho-anatomical and patho-physiological features and have very intense relationship on cellular level, important to develop a well-formed and well-functioning body. An attempt is made to visualize with available diagnostic tools that the anatomic

neuro-osseous growth relations that will end-up in spinal deformities if discongruency between the two types of growth occur. The outside always reflects what is going on inside the body. Early and regular assessment of children's posture like in school screening with Adam's bending test and Finger Floor test will give the opportunity to prevent or correct by nonsurgical ways (exercise and bracing) spinal deformities and its consequences for the function of the neural structures. There is a need for further extensive research to compare these modalities in various patient groups. If clinical examination proves to be a good predictor of what an MRI will show, the need for expensive diagnostics can be lowered. Proper clinical examination (also digitalized) can provide a powerful tool to arrange a system for better screening and prevention on spinal pain syndromes in adulthood and early possibilities to adjust the morphogenesis and /or correct early deformities.

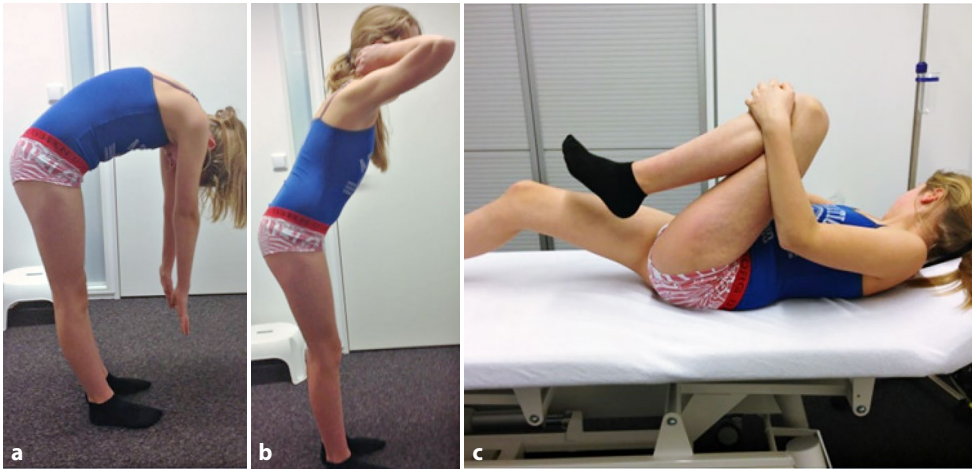
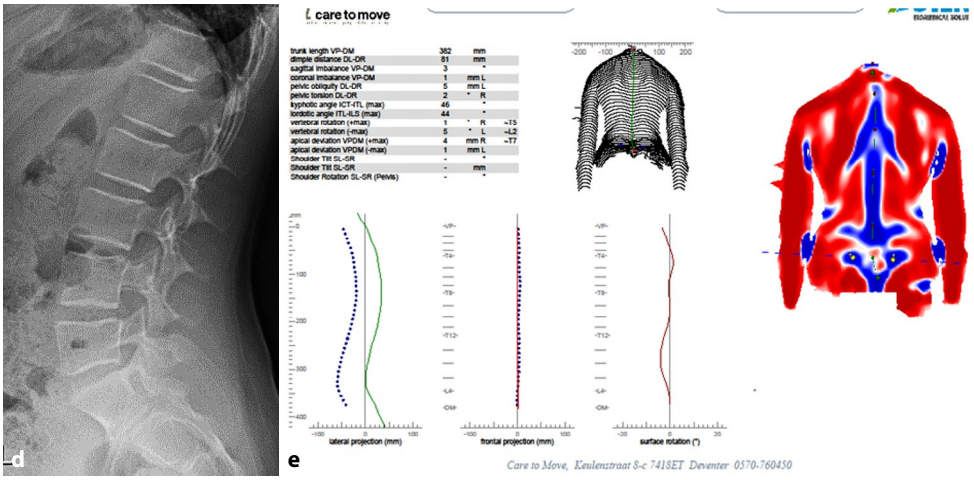


Figure: Example of the youngest case. Girl ,11 years old with chronic pain hip-groin-buttock after horse-riding. Clinical examination shows shortage of flexibility (Finger Floor Distance) (a), no lordosis at the 'scoop test' (b) and hip flexion contractures (c).



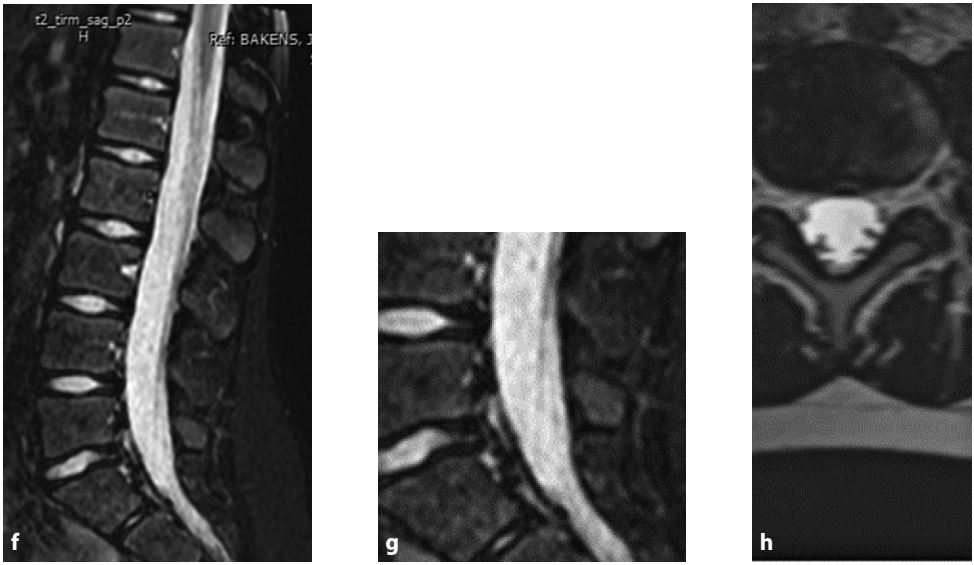


Figure: Example of the youngest case. Girl ,11 years old with chronic pain hip-groin-buttock after horse-riding.
On X-ray there is only some "lordosis" in the distal discs with an apparently pars intra-articularis S1 at risk to "break" into a spondylolysis (d). On the Formetric scan at the left an elongated kyphosis and no compensating lordosis that passes the central bodyline of gravity (e).
The MRI shows shallow discs, only slight lordosis and on the detail a lifted ligamantum posterior with bulging or disrupted annulus L5–S1 (f). The cauda and roots are displaced (by intrinsic tension) and tight to the posterior wall (g), on the transversal plane to be seen as "crowding" (h). Postural, stretching and mobility exercises were sufficient. Hopefully she understands that maintenance is the key for the durability of health.

PROF. DR. MED. HANS ZWIPP

Scientific curriculum vitae

Born 1st March 1949 in Neustadt (near Coburg)

Professor Zwipp, Chairman Emeritus of the Dresden Surgical Clinic, is an internationally renowned traumatologist, scientist, teacher, writer and above all an excellent physician respected by his patients and colleagues.

To this day, he is an invited speaker at numerous international symposia. Since 2017, he has been actively involved in the international symposium Prague-Lublin-Sydney-St. Petersburg, which we organize annually with colleagues from Lublin and St. Petersburg. We highly appreciate his publications in the journal Locomotor System – Advances in Research, Diagnosis and Therapy.

He has devoted his professional life to, surgery, orthopaedics and traumatology, as evidenced by his scientific curriculum vitae, which is attached. He received numerous honors and awards for his outstanding work during his lifetime.

- 1969 – A-levels at Martin-Butzer High School in Dierdorf in Westerwald
- 1969–1975 – Study of veterinary medicine, theology and humam medicine in Vienna, Berlin, Bochum and Essen
- 1975 – graduared aculty in Essen
- 1975–1977 – medical assitent in St. Vincenz hospital/ Bethesda in Essen
- 1978–1993 – surgery practise at Teaching Hospital in Hannover (MHH) in Lower Saxony (headed by Prof. Dr. med. H. Tscherne)
- 1982 – specialist in surgery
- 1984 – specialist in orthopaedics and traumatology
- 1984 – superintendent doctor at Trauma and Surgery Clinic in Hannover (MHH)
- 1985 – expert in trauma surgery problematics
- 1986 – specialist in sport medicine
- 1993 – specialist in urgent medicine
- 1992–1993 – the head of Trauma and Surgery Clinic in Hannover (MHH)
- Since 1994 – Department of Trauma and Reconstruction Surgery of University Hospital „Carl Gustav Carus" in Dresden
- 1994 – professor of surgery and recostruction surgery
- 1995 – specialist in hand surgery
- 2007 – the head of Surgery Clinic at University Hospital „Carl Gustav Carus" in Dresden

Research and publication activity :

- Research projects (36)
- Publications (409)
- Lectures (394)
- Leadership of dissertations

Honours and awards:

- 1987 – Hermann – Kümmel´s Price from NWD - Surgeons
- 1988 – Hans – Liniger´s Price of German Association for Trauma Surgery (DGU)
- 1994 – founding member and the president of ESFAS (European Foot and Ankle Society), Dresden
- 1994–2006 – chairman of the board AOI for surgery of foot and ankle
- 1998–1999 – president of Surgery Association in Saxony

- 1999 – 2001 – member of the bord of German Association for Trauma Surgery
- 2000 – 2004 – member of consulting board od German AO
- 2002–2008 – head of Working Association for Foot of DGU
- 2003 – Honorary membership of AO Alumni Association, Chile
- 2005–2007 – member of the board of DGU
- 2006 – Commemorative Medal of 3rd Medical Faculty, Charles University, Prague
- 2011 – Honorary membership of Czech Society for Trauma Surgery
- 2017 – Honorary membership of the Society for Connective Tissues, Czech Medical Association J. E. Purkynje

On the occasion of The 19th Prague-Lublin-Sydney-St Petersburg Symposium, which took place at the Medical House in Prague on 13–16 September 2017, Prof. Zwipp was awarded Honorary Membership in the Society for Connective Tissues of the Czech Medical Association J.E. Purkyne (CMA JEP).

The Committee of the Society for Connective Tissues of the CMA JEP, z.s. at the meeting on 21.10.2021 approved the nomination of Prof. Hans Zwipp for the award of the Honorary Medal of the CMA JEP.

This award was presented to him by Prof. Štěpán Svačina, MD, DSc., President of the CMA JEP, on the occasion of the 23rd Prague-Lublin-Sydney-St. Petersburg Symposium (Medical House in Prague, 20.11.2021).

Professor Zwipp shared his experience with the Symposium participants with a well-documented lecture „Classification of Sequelae of Foot and Ankle Syndromes in Adults, Adolescents and Children", which he is preparing for publication in the journal Movement System – Advances in Research, Diagnosis and Therapy.

Thank you Hans and good Health!

On behalf of the Committee of the Society for Connective Tissues CMA JEP

Professor Ivo Mařík, M.D., Ph.D.
president
E-mail: ambul_centrum@volny.cz

Prague, November 20, 2021

From the left Professor Tomastz Karski, Professor Hans Zwipp, Professor Ivo Mařík, Professor Ctibor Povýšil
The 19th Prague-Lublin-Sydney-St. Petersburg Symposium, September, 13–16, 2017

DR. PIET VAN LOON (Petrus Johannes Maria)

Scientific curriculum vitae

Born May 17, 1954 in Haarlem, The Netherlands

Dr. Piet Van Loon from Deventer is an orthopaedic surgeon fighting for "care to move" in the Netherlands based on aspects of Osteovertebral and Osteoneural growth relations by Milan Roth.

To this day, he is an invited speaker at numerous international symposia.

Since 2017, he has been actively involved in the Annual International Symposium Prague-Lublin-Sydney-St. Petersburg, where he presented interesting and very important two lectures titled *"Sedentary lifestyle and discongruent neuro-osseous growth relations (M. Roth) as external and internal etiologic factors of spinal deformity and skeletal malalignement. Modern youth and their short cord"* and *„Thoracolumbar lordotic intervention in spinal deformities. Effective mechanical growth modulation by muscular forces induced by bracing"*.

In 2019, Dr. Piet van Loon contributed in frame of the 20th annual PRAGUE-LUBLIN-SYDNEY-ST. PETERSBURG SYMPOSIUM, held in Kroměříž, to the section

„Spine Disorders I: Pathogenesis, Diagnosis and Treatment: Lectures dedicated to the memory of Milan Roth" with two lectures: *"A short cord can cause scoliosis: Osteoneural growth relations by Milan Roth (1923–2006) – a concise concept in morphogenesis and a useful scientific base for Orthopedics and Neuroscience" and „Growth of the human skeleton, a lifestyle dependant, tension based biomechanical and neurodynamic process"*.

Note: Assoc. Professor MUDr. Milan Roth, DSc. was a scientist, radiologist, who in the 60-90s of the 20th century experimentally proved the pathogenesis of skeletal deformities (spine and limbs) on the basis of disorders of macroneurotrophic growth of the spinal cord, its roots and peripheral nerves. During his lifetime, the importance of his work was unfortunately not appreciated behind the Iron Curtain. The results of his work on idiopathic scoliosis have been developed in clinical practice by only a few enlightened colleagues in this country, and abroad by Professor Dudin in St. Petersburg and Dr. Piet van Loon in Holland. His work is now also of interest to the Scoliosis Research Society in the USA.

Dr. Piet van Loon was accepted into Editorial Board of the journal Locomotor System – advances in research, diagnosis and therapy where he published a review of the life work of Assoc. Professor Milan Roth, DSc. – see Locomotor System journal, 2018, No. 1. (see http://www.pojivo.cz/pu/PU_1_2018.pdf)

Positions

Consultant Orthopedics	2018 – present	Proktovar/ Care to Move Hengelo
Consultant Orthopedics	2013 – present	Care to Move Orthopedic Clinic Deventer
Founder / boardmember	2016 – present	Houding Netwerk Nederland
Consultant Orthopedic surgery	2011–2013	Gelre Hospital Apeldoorn
Adviser	2011	Mediferia Clinic, Amersfoort.
Consultant Orthopedic surgery	2007–2010	Slingeland Ziekenhuis Doetinchem
Consultant Orthopedic/Spinal surgery	1991–2007	Rijnstate Hospital Arnhem

Training and consultant	1984–1991	Radboud University Nijmegen. (Prof. van Renst, Prof. Slooff)
Residency Orthopedics	1984–1985	Gemeente Ziekenhuis Arnhem
Residency Gen.Surgery	1982–1984	Mariastichting Haarlem
Residency Orthopedics	1980–1982	Military Hospital Utrecht
Residency Orthopedics	1979–1980	Ziekenhuis Zonnestraal Hilversum (Prof. Koekenberg)
Medical study	1971–1979	Free University Amsterdam

Between 2008 and 2018 a conflict with the Dutch Inspectorate of Healthcare, started on false allegations on having misconceptions in Medical science and by that performing "experimental surgery", ended in a letter full of excuses by the Inspectorate on behalf of the Minister of Health.

Relevant functions and memberships

1986–1989	President Vereniging Orthopedisch Chirurgisch assistenten (VOCA)
1993–1998	Secretary Spine Study Group of the Dutch Orthopedic Society (NOV)
1998–2001	President Spine Study Group of the Dutch Orthopedic Society (NOV)
2001–	Cofounder and member of the Dutch Spine Society
1995–2008	Member advisery board Scoliosevereniging (Dutch Scoliosis Association)
1992–	Member European Spinal Deformity Society, now European Spine Society
2004–2009	International Fellow Scoliosis Research Society
2012–	Member Society on Scoliosis Orthopedic and Rehabilitation Treatment SOSORT and Dutch Medical Societies : KNMG/ FMS, DSS, NOV, NVOT.

Inventor/ patentkeeper Brace for Spinal Deformities called TLI brace EPO: 04 808 807.4-2310 / 2008

Inventor /patent keeper Sit Active device with posture optimalisation, called Zami.

2020	Honorary membership of the Society for Connective Tissues (SCT), Czech Medical Association J.E. Purkynje (CMA JEP).

Piet van Loon, MD published about 40 scientific papers in international medical journals.

A few of his outstanding publications were influenced by the lifetime work of Associate Professor Milan Roth DSc.

We introduce two of his most appreciated papers:

VAN LOON P, VAN ERVE R. The Development of TLI (Thoracolumbar Lordotic Intervention) as an Effective Bracing Concept for the Postural Spinal Problems –A Review. J Spine, Vol 4, 2015, No. 3, 7 p. DOI: 10.4172/2165-7939.1000226.

VAN LOON PJ, GROTHENIUS JA. Legacy of Milan Roth: Osteoneural growth relations, the biomechanic and neurodynamic processes of physical body growth in vertebrates with tension as its tool to overcome gravity. Clinical implications of discongruent osteoneural growth. Locomotor System journal vol. 25, 2018, No. 1, p. 25–67.

The Committee of the Society for Connective Tissues of the CMA JEP at the meeting on 21.10.2021 approved the nomination of Dr. Piet van Loon for the Medal of Honour of the CMA JEP at its meeting on 21 October 2021.

This award was presented to him by Prof. Štěpán Svačina, MD, DSc., President of the CMA JEP, on the occasion of the 23rd Prague-Lublin-Sydney-St. Petersburg Symposium (Medical House in Prague, 20.11.2021).

Dr. van Loon shared his experience with the participants of the symposium through two lectures:

„Sitting as the unspoken accelerator of the "OsteoNeural Pandemic. Staggering fresh data and evidence in (Dutch) youth in the light of classic Orthopedics"
and
„Neuroradiologic treasures on MRI in discongruent Osteoneural Growth Relations Consequent findings on MRI in case of malalignment of the skeleton (posture and deformities) and the lack of contemporary research"

Dear Piet, thank you very much for your fruitful cooperation and good Health!

On behalf of the Committee of the Society for Connective Tissues CMA JEP

Professor Ivo Mařík, M.D., Ph.D.
president
E-mail: ambul_centrum@volny.cz

&

Petr Krawczyk, M.D.
President of the Society for Prosthetics and Orthotics CMA J.E. Purkynje
E-mail: krawczyk@proteorcz.cz

Professor Mařík (left) and Dr. Piet van Loon. The 21st Prague-Lublin-Sydney-St. Petersburg Symposium, 3rd–5th September 2019, Humpolec

Systém výživy kloubů dle výzkumu prof. MUDr. Milana ADAMA, DrSc.

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