CURRICULUM VITAE

Dr. Ali Al Kaissi, MD, MSc, Dsc (hon)
Krottenbach Strasse 229 Top 10
1190 Wien

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E-Mail : Ali.kaissi@oss.at
Kaissi707@gmail.com

ÖÄK – Ärztenummer: 53876 - 19

PERSONAL DATA

Date of Birth:	18.12.1951
Place of Birth:	Bagdad, Irak
Marital status:	Married to Ms. Siba Ghazi Shakir, geb. 4.12.1963 in Bagdad, Irak
Children:	Rima, geb. 4.8.1987 in London, UK, Omar, geb. 2.3.1992 in Tunis, Tunis, Hamza, geb. 22.3.1996 in Tunis, Tunis
Citizenship:	Austrian and Iraqi citizenship Possessing two nationalities in accordance with the decision of Ministers of Council in May 2014, Vienna, Austria Under the Category of High Grade Scientist [Bestätigung des Staatsinteresses durch die Bundesregierung gem. §10Abs 6 StbG]
Hobbies:	Black Belt-Dan-I-Kukinshai -Karati

QUALIFICATIONS

2017	**Doctor of Medicine and Science (Dsc-Honorary)** Turner Scientific Research Institute for Children's Orthopedic, Saint Petersburg, Russia
1988-1990	**Diploma in Pediatrics and Master Degree in (Paediatric Developmental Abnormalities)** School of Medicine, University of Warwick-UK
1987 - 1990	**School of Medicine**, University of Warwick-Paediatric Dept.UK
1987-1988	**Certificate of Pediatrics-School of Postgraduate Medicine** University of Warwick-UK
1972 – 1978	**Faculty of Medicine**, Azhar University, Kairo, Egypt,

EXPERIENCES

2005 - Present:	Consultant and Scientist Pediatric Department-Orthopedic Hospital of Speising, Vienna Austria
2005 - Present:	Consultant at Ludwig-Boltzmann Institute of Osteology-Hanusch Krankenhaus, Vienna Austria
11/1993 - 6/2005	Consultant and Clinical Researcher at the Department of Paediatric Orthopaedics, Children Hospital- Tunis, Tunesia
3/1991 - 10/1993	Establishment of the Tunisian Center of Early Recognition of Children Malformation Complex, Office Nationale de la Famille et Population, Ministry of Health Tunis,Tunisia (the clinical observations have been collected in a book 1993, Ministry of Health in Tunisia)
1/1987 - 9/1990	Clinical Attachment at the Paediatric Department at Walsgrave Hospital and Coventry & Warwickshire Hospitals, UK
1/1984 - 11/1986	Senior House Officer, Red Crescent Hospital-Bagdad, Irak
7/1983 - 12/1984	Resident at the Orthopaedic Department, Military Hospital Bagdad, Irak
5/1982 - 6/1983	Resident in the Rehabilitation Department, Military Hospital Bagdad, Irak
12/1981 - 2/1982	Resident at the Internal Medicine Department, Cardiology Department, Rheumatology Department and Endocrinology, Air Force - Hospital Bagdad, Irak
2/1980 - 12/1981	Resident at the Air Force Military Hospital Bagdad, Irak
1979 - 1980	Rotation House Officer, Hussein Univ. Hospital, Cairo, Egypt

BREAKTHROUGH IN MEDICINE

1. **Al Kaissi syndrome - ALKAS**

   Growth retardation, spine malformation, dysmorphic facies, and developmental delay

Location	**16q24.3**
Phenotype	**Al Kaissi syndrome**
Phenotype MIM number	**617694**
Inheritance	**AR**
Phenotype mapping key	**3**
Gene/Locus	**CDK10**
Gene/Locus MIM number	**603464**

2. Al Kaissi et al. **3MC syndrome-nature genetics** -2011(43: 197-203)

3. Al Kaissi A et al. A novel **syndrome resembling Desbuquois dysplasia**. Am. J. Med. Genet. 132A: 68-75, 2005

4. Al Kaissi et al. **Progressive collapse of the Thoracic Cage**. American J of Medicine, 2017

MEMBERSHIPS

1. Member of the **Royal Society of Medicine** (London) (00714261)–UK
2. Member of the **Austrian Medical Council** (53876-19)
3. Member of the **Tunisian Medical Association** (92221)
   (Conseil Regional de Tunis de L´ Ordre des Medicins)
4. Member of the **Scientific Board of the Pediatric Traumatology, Orthopaedics, and Reconstructive Surgery**, the founder of which is the Turner Scientific Research Institute for Children's Orthopedic, Saint Petersburg, Russia
5. Member of the **Science Advisory Board** (Arlington)-USA
6. Member of the **Russian ACADEMY of natural History and Medicine**

REWARDS

September 2019

- **Certificate of honor**
  Ministry of Health Republic of Dagestan-Federal Republics Of Russia

- **The Silver Dagistani Sword for Distinguished Clinicians and Researchers**
  Ministry of Health-Russia

  ..

December 2017

- **Honorary Doctorate in Medicine and science Dsc**
  Pediatric Orthopedic Institute n.a. H. Turner, Department of Foot and Ankle Surgery, Neuroorthopaedics and Systemic Disorders, Parkovaya str., 64-68, Pushkin, Saint-Petersburg, Russia

- **Russian Golden Medal For Distinguished Clinicians**
  Turner Orthopedic Research Institute-Saint Petersburg-Russia

INTERNATIONAL COLLABORATIONS

2011 – present

Research Partnership with Pediatric Orthopedic Institute n.a. H. Turner, Department of Foot and Ankle Surgery, Neuroorthopaedics and Systemic Disorders, Parkovaya str., 64-68, Pushkin, Saint-Petersburg, Russia

1998 – present

Research collaboration with Prof Michael Baraitser- Department of Genetics-Great Ormond Road-London-we wrote together 22 papers we still working together in studying children and families with Dysmorphic features in connection with orthopaedic abnormalities. all@lokibooks.viane.co.uk

At present

University of Saskatchewan-CanadaDept. Of Environmental Medicine

Project Title

Bioaccumulation of Novel Flame Retardants and Exposure-related Effects in Wild Terrestrial Animals from Two Trophic Levels in the Canadian Arctic.

First Published paper

"Chronic arsenicosis and cadmium exposure in wild snowshoe hares(Lepus americanus) breeding near Yellowknife, Northwest Territories(Canada), part 2: Manifestation of bone abnormalities and osteoporosis

Authors Amuno S, Al Kaissi A. Jamwal A. , Niyogi S., Quenneville
Science of the Total Environment Journal; 2019; 612 ; 1559–1567

Second paper published, Dec.2020

Amuno S, Al Kaissi A. *A Radiographic densitometry study* . Ecotoxicology and Environmental Safety Journal

Skeletal pathology and bone mineral density changes in wild muskrats (Ondatra zibethicus) and red squirrels (Tamiasciurus hudsonicus) inhabiting arsenic polluted areas of Yellowknife, Northwest Territories (Canada**)**

February 2020 - February 2024

Pediatric Kidney clinic Machatshkala Dagistan Republic-Russia (batdalova.kidney) Bone disorders- www.Kidney.clinic.ru

Location: Makhachkala, Russia

Position: Visiting Professor in collaboration with Prof. Zulfiya Batdalova (zul.bat@mail.ru)

Description: Examining children and adults with various forms of developmental deficits ranging from skeletal dysplasia to syndromic association , neurological deficits and so forth. All types of musculo-skeletal disorders. The objective is to approach dignosis, which is the baseline tool for proper mangament.

November 2015 - present

Project Title: Spine Malformation Complex

Location: Russia

Department: Iliazarov Institute-Kurgan; Russia (Prof Sergey R)

Position: Professor Clincial and radiological documentation for children with various spine malformation complex to approach for diagnostic entities and management Research Partnership with Prof. Sergey Ryabykh

January 2010 - December 2024

Department: Pediatrics: Clincial and radiological documentation for children with various forms of congenital malformations to approach for diagnostic entities and management Saint Petersburg, Russia

Location: Pediatric Orthopedic Institute n.a. H. Turner, Department of Foot and Ankle Surgery, Neuroorthopaedics and Systemic Disorders, Parkovaya str., 64-68, Pushkin, Saint-Petersburg, Russia

Position: Professor

2004 - 2005

Research partnership and collaboration with Dr Peter Turnpenny- Department of Genetics- Exeter-. Royal Devon and Exeter NHS Foundation trust-UK

2002 - 2007

Research collaboration with Prof. Kazimierz Kozlowski-Department of Imaging-Children Hospital- Sydney-Australia: we wrote and published together 8 papers (Till 2004) and for 2005 we submitted a remarkable number of studies to high classical journals-our project was documented in October 2004. kazimiek@chw.edu.at

1999 - 2001

Research collaboration with Prof Pierre Maroteaux-Necker Enfants Malades-Paris.

1993 - 1995

Research collaboration with Prof. Eberhard Passarge-University of Essen-department of human genetics- investigating Tunisian families with Bloom syndrome.

REVIEWER IN THE FOLLOWINGS SCIENTIFIC BODIES (refer to Publon.com)

1. **Reviewed more than 300 papers for the international peer journals worldwide**.
2. Official Reviewer of Spine Journal-USA
3. Editorial Board Member of BMJ case report journal
4. Scientific Reviewer of the Ministry of Science and Technology-Copenhagen, Denmark
   Anna Thoren Head of section Danish Council for Strategic Research Direct Phone: + 45 339 552 63 E-mail: anth@fi.dk Danish Agency for Science, Technology and Innovation
   Bredgade 40 DK-1260 Copenhagen K
   Phone: +45 3544 6200
   Fax: +45 3544 6201
   E-mail: dasti@dasti.dk
   www.dasti.dk

CONFERENCES AND SCIENTIFIC MEETINGS

2009

Early senile ankylosing vertebral hyperostosis in Paediatric patients; a novel spine malformation
Ali Al Kaissi, Goerg Kalchhauser , Goerg Haller , Franz Grill and Klaus Klaushofer ECTS. Wien- Österreich

2009

Novel Mutation in the Carboxyl-terminal Propeptide of the Procollagen α1(I) Chain in a girl with prenatal cortical hyperostosis and multiple fractures
Katharina M Roetzer , Franco Laccone , Alexander Krebs, Franz Grill , Simon Robins, Franz Varga , Klaus Klaushofer , Ali Al Kaissi Cambridge-UK

2008

Evidence of reduced bone turnover and disturbed mineralization process in a boy with Stickler syndrome.Al Kaissi, Paul Roschger, F Grill, K Klaushofer Barcelona Spain ECTS

2007

Differential diagnosis of spine maldevelopment and osteoporosis A Al Kaissi, E Zwettler, F Grill and K Klaushofer Hofburg Palace. Vienna, Austria. Osteologie

2006

Craniocervical malformation complex and syndromic associations
A Al Kaissi, F Varga, F Grill, Klaushofer Hofburg palace. The international Bone research conference in Vienna, Austria.

2004

Congenital dysplastic hips in three generation Tunisian family with Beighton dysplasia like Al Kaissi et alBritish Orthopaedic association- British Hip Society- Sheffield-Marriott Hotel- UK.

2003

The syndromic complex association of congenital hip dislocation in Tunisian families Al Kaissi et al. The French- Tunisian Radiology Socieities- Tunis

2002

Limb inequality and the syndromic complex association
Al Kaissi et al., Les Lundis d orthopaedie (seminar) Tunis.

2001

Marfanoid Habitus in Orthopaedic practice; the precise diagnostic identification for proper management
El Mechtel Hotel- Tunis Al Kaissi et al., Les Lundis d orthopaedie (seminar)

2001

Patellar instability and dislocations in children with serious heritable disorders
Al Kaissi et al The Annual scientific meeting of the Sotcot (The Tunisian Society of Surgery and Orhtopaedics) Tunis.

2000

The management of children with Achondroplasia
Tunis. Al Kaissi et al. Les Lundis d orthopaedie (seminar) , Ali Al Kaissi and Professor Maher B . Ghachem

2000

The annaul scientific meeting of the Tunisian Medical Scietific Society
El Mechtel- Hotel Tunis. Al Kaissi et al., Developmental abnormalities and bone disorders in syndromic association- Lecture

1999

ELLIS Van Crevealed (Clinical and radiological features)-Poster
Etiological diagnosis in three families with congenital scoliosis, kyphosis and kyphoscoliosis-Poster
Al Kaissi et al.,journee medicale de la CNSS , Biezierte , Tunis

1999

Differential diagnosis in dwarfism secondary to osteochondrodysplasia, 6 september-Tunis
Al Kaissi et al.,Les Lundis d orthopaedie (seminar) , Ali Al Kaissi & Professor Maher B . Ghachem

1998

1.Osteogenesis imparfecta (different skeletal abnormalities over 3-generations)
Al Kaissi et al. journee medicale de le CNSS 19 et 20 mai , Soussa-Tunis , Three presentations

(3 M syndrome) associated with Scheurmann's disease (presentation of one family)

Ectodermal dysplasia (orthopaedic abnormalities in 2 families)

1997

Particularites de deux observations de syndrome de Noonan rentrant dans le cadre d un grossesse gemellaire (presentation)
Tunis. Al Kaissi et al. XVIIIeme congres Maghrebin de pediatrie

1997

Ligamentous hyperlaxity and the syndromic complex association (differential diagnosis on clinical and radiological grounds) presentation of 12 Tunisien families-Tunis
Al Kaissi et al.Les Lundis d orthopedie , service du Prof . Maher ben Ghachem

1997

Lecture in the Embryological development and the congenital malformations, clinical & radiological approach to the diagnosis of 10 Tunisien families with different orthopaedic abnormalities
Tunis. Al Kaissi et al. societie Tunisien de chirurgie (Tunisian Society of Surgery, Orthopaedics and Traumatology)

1996

Hyperlaxitie ligamentaire ; 20 may (Presentation)
Tunis. Al Kaissi et al. Journée médicale CNSS

1995

Aetiological diagnosis of 14 Tunisien families presented with different syndromic associations Al Kaissi et al., Oral presentation ; Societe Tunisiene de chirurigie , orthopaedic et tramatologie , sotcot , El Mehdia- Tunis

Oligodactyly and odontoid hypoplasia in a family with Cornelia de Lange syndrome

a. 3-families with different orthopaedic abnormalities secondary to Osteochondrodysplasia (Achondroplasia, Chondrodysplasia punctata, Ellis van crevealed syndrome)
b. A family of variable skeletal malformations- three generations-of Marfan syndrome
c. Congenital scoliosis in a family with Ehlers Danlos syndrome type I.
d. Multiple Exostosis in two families
e. Facial features as a key factor in two families with Larsen syndrome.
f. Congenital kyphoscoliosis as the presenting clinical abnormality in Osteo genesis imperfecta (2 families)
g. Congenital scoliosis and sudden infant death syndrome in a family with Noonan syndrome.

1995

The early recognition of families with Adenocarcinoma through proper identification of the paraneoplastic syndromes, presentation of 16 Tunisian families with adenocarcinoma and different syndromic association
Al Kaissi et al, The French Tunisian conference- Institute Salih Ezaiz (Hospital of Oncology) Tunis.

1994

Clinical diagnosis of 3 generational Tunisien family with Ectodermal dysplasia (Oral presentation)
Al Kaissi et al. journée Genetique; el Mechtel; organised by Professor H. Chaabouni,

1994

Different clinical presentations in a Tunisien family with the clinical diagnosis of E. E. C. syndrome
Al Kaissi et al.the 66th. Oral presentation- Annual meeting of the BRITISH Peadiatric association: Universirty of Warwick-UK

1993

Congree arabe pediatrie; El Mechtel – Tunis; Ali Al Kaissi et al.

a. Case report of Noonans Syndrome (oral presentation)
b. Orthopaedic abnormalities in a family with ectodermal dysplasia (poster)
c. Klippel – Fiel anomaly in two generations (poster)
d. The etiological diagnosis of arthrogryposis multiplex congenit (poster)

1993

Omnipratique et pathologie infantile , Syndrome de Papillon –lefevre
a propos d un cas – France (presentation) Buslama & Al Kaissi XXVemes journees , de la societe francaise de pedodontie Reims 14-15 – 16 mai

1993

Invisible Syndromes in normal looking family subjects, though their main presentation is Learning difficulties, these were encountered in
(1. A family with Noonan syndrome
(2. A family with Aarskog syndome.
(3. syndrome of Hunter –mild type of Mucopolysacharoidosis).
Al Kaissi et al., Oral presentation 1993- Saturday 8 May ; the third regional scientific meeting of the Ministry of health, City of Ariana-TUNIS : Three presentations

PUBLICATIONS 1987 - 2020

Refer to Research Gate
https://www.researchgate.net/scientific-contributions/39876458_Ali_Al_Kaissi

2020

2020
A Constellation of Orthopaedic Deformities in Connection with Cartilage Oligomeric Matrix Protein Mutation
Background: Trendelenburg's gait can be observed in Legg-Calvé-Perthes disease, antalgic gait observed in osteoarthropathy and waddling gait is usually seen in genu varum and circumduction gait in patients with genu valgum.

2020
The articular and the craniocervical abnormalities are of confusing age of onset in patients with Maroteaux-Lamy disease (MPS VI)
Background: Maroteaux-Lamy disease (MPS Type VI) is an autosomal recessive lysosomal storage disorder. Skeletal abnormalities are vast. Early recognition may facilitate timely diagnosis and intervention, leading to improved patient outcomes.

2020
Progressive Deformity of the Lower Limbs in a Patient with KID (Keratitis-Ichthyosis-Deafness) Syndrome
Purpose: Progressive deformity of the lower limbs can be encountered in a long list of syndromic associations.

2020
British Hip Society
Beighton and Kozlowski, in 1980, first defined this disorder, in Afrikaners, the syndrome , evident at birth, is constantly manifested by dwarfism, ligamentous hyperlaxity, congenital scoliosis, and multiple dislocations(hip dislocation, radial head dislocation, scoliosis, spatulate thumbs, and generalized ligamentous hyperlaxity

2020
Arthrogrypotic Syndrome is the Usual Misnomer in Children with Du Pan Syndrome
Objective: The term arthrogrypotic syndrome is of common usage by different medical disciplines. The hallmarks of acromesomelic dysplasia Du Pan Syndrome are characteristic facial features, severe growth deficiency and multiple contractures.

2020
Arthrogryposis is a descriptive term, not a specific disease entity: escobar syndrome is an Exampleticle
Background: Children born with multiple congenital contractures have been almost always given the diagnosis of arthrogryposis multiplex congenita. Arthrogryposis is a descriptive term, not a specific disease entity.

2020
Clinical and Genetic Heterogeneity in Six Tunisian Families with Horizontal Gaze Palsy With Progressive Scoliosis: A Retrospective Study of 13 Cases
Background: Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a rare autosomal recessive congenital disorder characterized by the absence of conjugate horizontal eye movements, and progressive debilitating scoliosis during childhood and adolescence.

2020
Tomographic Study of the Malformation Complex in Correlation With the Genotype in Patients With Robinow Syndrome: Review Article
We aimed to understand the etiology behind the abnormal craniofacial contour and other clinical presentations in a number of children with Robinow syndrome.

2019

2019
AJPS 90 17R5
Background: Trendelenburg's gait can be observed in Legg-Calvé-Perthes disease, antalgic gait observed in osteoarthropathy and waddling gait is usually seen in genu varum and circumduction gait in patients with genu valgum. Disabling pain was a prime manifestation in slipped capital femoral epiphysis (SCFE).

2019
Pseudorheumatoid and Os odArticle
We report the case of a girl-child who manifested the clinicoradiographic features of pseudorheumatoid arthritis. 3D-CT scan of the craniocervical junction showed distinctive features of dystopic type of os odontoideum.

2019
Acrodysostosis
Abstract Background Shortness of stature, dysmorphic facial features and intellectual disability associated with a broad spectrum of variable psychotic illnesses in a multi-generation family with acrodysostosis.

2019
Acrodysostosis in two female siblings revealed the recognition of several family subjects with a broad spectrum of Psychotic disorders
Background Shortness of stature, dysmorphic facial features and intellectual disability associated with a broad spectrum of variable psychotic illnesses in a multi-generation family with acrodysostosis.
2019
The Skeletal phenotype/ genotype in progressive pseudorheumatoid chondrodysplasia
Clinical Rheumatology pp 1–8 | Cite as Skeletal phenotype/genotype in progressive pseudorheumatoid chondrodysplasia

2019
Skeletal phenotype/genotype in progressive pseudorheumatoid chondrodysplasia
Background Axial and extra-axial deceleration in function and progressive joint pain with subsequent development of antalgic gait associated with swellings, and stiffness of the joints with loss of the physiological spine biomechanics were the natural history in this group of patients.

2019
Goldenhar syndrome-paper
Abstract Purpose Goldenhar syndrome consists of a combination of unilateral auricular appendages, auricular fistulas, and ocular epibulbar dermoids combined with a unilateral underdevelopment of the craniofacial structures and vertebral abnormalities.

2019
A child with a congenital hand anomaly
Unfortunately, Incomplete clinical documentation, has become a model and of common practice in most of the medical disciplines. Medical Journals main task is to expand the horizon of knowledge and to educate the readers with the best possible genuine methodology.

2019
Medicines Leri-Weill Dyschondrosteosis Syndrome: Analysis via 3DCT Scan
Background: Leri-Weill dyschondrosteosis (LWD) is a pseudoautosomal form of skeletal dysplasia, characterized by abnormal craniofacial phenotype, short stature, and mesomelia of the upper and lower limbs. Methods: We describe two female patients with LWD.

2019
Massive Axial and Appendicular Skeletal Deformities in Connection with Gorham-Stout Syndrome
Background: Etiological understanding is the corner stone in the management of skeletal deformities. Methods: Multi-centre study of patients with deformities in connection with diverse etiological backgrounds.

2019
Unilateral lytic changes over the weight-bearing joint causing severe destruction of ankle joint (atypical Charcot joint) in a girl with congenital insensitivity to pain without anhidrosis (hereditary sensory and autonomic neuropathy type V): Case report and literature review - Background. The presence of Charcot arthropathies, joint dislocations, infections and fractures in a child without evidence of neurological abnormality should give rise to a suspicion of congenital insensitivity to pain (hereditary sensory and autonomic neuropathy).

2019
The Managment of cervical spine abnormalities in children with spondyloepiphyseal dysplasia congenita: Observational study
Spondyloepiphyseal dysplasia congenita (SEDC) is an autosomal dominant disorder, characterized by disproportionate dwarfism with short spine, short neck associated with variable degrees of coxa vara. Cervical cord compression is the most hazardous skeletal deformity in patients with SEDC which requires special attention and management.Ten

2018

2018
Bilateral cox vara and Tibia vara Pediatric Traumatology, Orthopaedics and Reconstructive Surgery. In most instances, a toddler is seen with unilateral varus of the tibia, usually the deformity appearing slightly more distal than the knee joint. Radiographs of the focal fibrocartilaginous dysplasia show a characteristic abrupt varus at the metaphyseal –diaphyseal junction of the tibia

2018
Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Abstract Analysis of tissue from a 34-years-old male patient from Austrian origin with a history of multiple fractures associated with painful episodes over the carpal, tarsal and at the end of the long bones respectively is presented.

2018
Bilateral Coxa Vara and Tibia Vara associated with severe short stature in a girl manifesting a constellation of bone lesions with exclusive involvement of the lower limbs. In most instances, a toddler is seen with unilateral varus of the tibia, usually the deformity appearing slightly more distal than the knee joint. Radiographs of the focal fibrocartilaginous dysplasia show a characteristic abrupt varus at the metaphyseal diaphyseal junction of the tibia.

2018
Re- Shoulder pain in Smokers (BMJ-Case reports)

2018
Schmid's Type of Metaphyseal Chondrodysplasia: Diagnosis and Management: Objectives There are several types of metaphyseal chondrodysplasia and various clinical types have been differentiated. The Schmid type of metaphyseal chondrodysplasia is the most common.

2018
Is attention deficit/Hyparactivity disorder (ADHD) a diagnosis or a symptomcomplex: Experience from pediatric orthopedic practice

2018
Turning the backbone into an ankylosed concrete-like structure: Case report Rationale: Progressive restriction of the spinal bio-mechanics is not-uncommon deformity encountered in spine clinics.

2018
BMJ-Rapid Response-Re-deep vein thrombosis
Figure 1provided by the author, showed deep vein thrombosis in the right leg of a patient with leg swelling and erythema which are visible, but the author did not comment on the skin over the knees.

2018
BMJ-Rapid Response

2018
BMJ-Plain radiography of the skull after investigation for raised calcium

2018
BMJ- RAPID RESPONSE-Chest and neck pain

2018
BMJ-RAPID RESPONSE-LIMPING CHILD

2018
BMJ-RAPID-RESPONSE-Re a baby ..

2018
Carpal Fusion in Ellis-Van Creveld Syndrome
Carpal anomalies occur in a variety of syndromic associations. In many of these the carpal findings may be characteristic of the disorder. Carpal fusion may occur as an isolated anomaly or related to syndromic association.

2017

2017
Are parents of children with Cockayne syndrome manifesting features of the disorder: Case reports Rationale: Postnatal growth failure and progressive neurologic dysfunction and increasing multiorgan involvement are the main clinical features of Cockayne syndrome (CS).

2017
Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays
In five separate families, we identified nine individuals affected by a previously unidentified syndrome characterized by growth retardation , spine malformation, facial dysmorphisms, and developmental delays.

2017
Chronic arsenicosis and cadmium exposure in wild snowshoe hares (Lepus americanus) breeding near Yellowknife, Northwest Territories (Canada), part 2: Manifestation of bone abnormalities and osteoporosis

2017
How frequent is osteogenesis imperfecta in patients with idiopathic osteoporosis: Case reports

Rationale The term idiopathic osteoporosis itself is quite a non-specific disease label, which fails to address the etiological understanding. Bone mineral density alone is not a reliable parameter to detect patients at high risk of fracture

2017
A novel mutation in ATRX associated with intellectual disability, syndromic features, and osteosarcoma

2017
Craniosynostosis, Scheuermann's disease, and intellectual disability resembling Shprintzen-Goldberg syndrome: A report on a family over 4 generations Case report

2017
Abstract rationale: Craniosynostosis is a disorder characterized by premature fusion of cranial sutures with subsequent development of abnormal craniofacial contour associated with variable skeletal and extra-skeletal abnormalities

2017
Muscle Weakness: A Misleading Presentation in Children With Distinctive Syndromic Entities (Clinical Case Reports) Marked ligamentous hyperlaxity and muscle weakness/wasting associated with awkward gait are the main deficits confused with the diagnosis of myopathy. Seven children (6 boys and 1 girl with an average age of 8 years) were referred to our department because of diverse forms of skeletal abnormalities.

2017
AL KAISSI SYNDROME (ALKAS) Patent
Al Kaissi syndrome is an autosomal recessive developmental disorder characterized by growth retardation, spine malformation, particularly of the cervical spine, dysmorphic facial features, and delayed psychomotor development with moderate to severe intellectual disability (summary by Windpassinger et al., 2017).

2016

2016
Spine malformation complex in 3 diverse syndromic entities: Case reports
Rationale: Clinical and radiographic phenotypic characterizations were the base line tool of diagnosis in 3 syndromic disorders in which congenital cervico-thoracic kyphosis was the major deformity.

2016
Can Multiple Hereditary Exostoses Overlap With Mesomelic Dysplasia
Background: We studied an unusual combination of severe short stature, mesomelia (Leri-Weill dyschondrosteosis syndrome), and multiple exostosis in several family subjects over three generations.

2016
The constellation of skeletal deformities in a family with mixed types of mucopolysaccharidoses: Case report Introduction: A 13-year-old child was clinically diagnosed with mucopolysaccharidosis type VI-Maroteaux-Lamy syndrome (MPS VI) at the age of 5 years, and the diagnosis was confirmed biochemically and genetically (homozygous mutation in ARSB gene).

2016
Lower limbs deformities in patients with McCune-Albright syndrome: Tomography and treatment Background: The skeletal changes in McCune-Albright disease are usually severe because of the polyostotic form of the disease. Trendelenberg gait and limited mobility are the most common presenting features.

2016
Age of diagnosis of fibrodysplasia ossificans progressiva has a variable onset and a misleading phenotype Background: The clinical presentation and the clinical phenotypic characterization and the natural history of Fibrodysplasia Ossificans Progressiva (FOP) is diverse and the natural history of the disease is to certain extent different from one patient to another.

2016
Corrections of Diverse Forms of Lower Limb Deformities in Patients with Mucopolysaccharidosis type IV A (Morquio Syndrome)

2016
Corrections of diverse forms of lower limb deformities in patients with mucopolysaccharidosis type IVA (Morquio syndrome) Background: Thoracolumbar kyphosis has been considered as the first presenting deformity and is often a key diagnostic clue noted in children with mucopolysaccharidosis (MPS) type IV (Morquio's syndrome).

2016
The Diversity of the Clinical Phenotypes in Patients with Fibrodysplasia Ossificans Progressiva Background: The clinical presentation, phenotypic characterization and natural history of fibrodysplasia ossificans progressiva (FOP) are diverse and the natural history of the disease is, to a certain extent, different from one patient to another.

2016
Progressive Collapse of the Thoracic Cage

2015

2015
Distinctive Skeletal Abnormalities with No Microdeletions or Microduplications on Array-CGH in a Boy With Mohr Syndrome (Oro-Facial-Digital Type II)

2015
We describe a constellation of distinctive skeletal abnormalities in an 8-year-old boy who presented with the full clinical criteria of oro-facial-digital (OFD) type II (Mohr syndrome)

2014

2014
A child with split-hand/foot associated with tibial hemimelia (SHFLD syndrome) and thrombocytopenia maps to chromosome region 17p13.3. Am J Med Genet A

2014
Swellings over the Limbs as the Earliest Feature in a Patient with Osteogenesis Imperfecta Type V. Case Rep Orthop.

2014
Reconstruction of bilateral tibial aplasia and split hand-foot syndrome in a father and daughter. Afr J Paediatr Surg.

2014
Agenesis of the Corpus Callosum and Skeletal Deformities in Two Unrelated Patients: Analysis via MRI and radiography . Case Rep Orthop.

2014
Distinctive spine abnormalities in patients with Goldenhar syndrome: tomographic assessment. Eur Spine J.

2013

2013
Windswept lower limb deformities in patients with hypophosphataemic rickets. Swiss Medical J.

2013
Is webbing (pterygia) a constant feature in patients with Escobar syndrome Orthop Surg.

2013
Craniovertebral malformation complex in a child with Weismann-Netter-Stuhl syndrome. J Pediatr (Rio J)

2013
Progressive congenital torticollis in VATER association syndrome. Spine Journal

2013
Progressive non-infectious anterior vertebral fusion, split cord malformation and situs inversus visceralis; a novel malformation complex in a female child. BMC Musculosceletal Disorders

2013
Atlanto-axial rotatory fixation in a girl with spondylocarpotarsal synostosis syndrome. Scoliosis

2013
Asymmetrical skull, ptosis, hypertelorism, high nasal bridge, clefting, umbilical anomalies, and skeletal anomalies in sibs: is carnevale syndrome a separate entity? Am J of Med Genet Part A

2013
Unusual facies, thumb hypoplasia, distinctive spinal fusions and extraspinal mobility limitation, in a pair of monozygotic twins. Clinical Dysmorphology

2013
Vertebral hyperostosis, ankylosed vertebral fractures and atlantoaxial rotatory subluxation in elderly patient with a history of infantile idiopathic scoliosis. Journal of Medical Case Reports

2013
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