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CaseReport

BilateralVarusDeformityCorrectionandLegLengtheningwith anIlizarovFixatorinaFemalewithTrichorhinophalangealSyndromeType1(TRPSI)–CaseReport

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2. Keywords

Corticotomy; Trichorhinophalangeal syndrome; Osteochondromas

1.Introduction

Trichorhinophalangeal Syndrome Type I (TRPS I) is a rare multisystem disorder with autosomal dominantpatternofinheritance.ThedisorderwasfirstdescribedbyA.Giedionin1966[1].Patients with TRPS I present with facial, metacarpal, metatarsal, and phalangeal deformities. Characteristic featuresincludeahighforehead,largenosewithabulboustip,narrowupperlip,andsparse,thinhair [2,3].TRPSIiscausedbyamutationintheTRPS1genelocatedinchromosome8.[4]Therearethree subtypes of trichorhinophalangeal syndrome. TRPS type II is characterized by multiple osteochon- dromas, whereas TRPS type III features severe brachydactyly and more severe short stature than in type I [5-7]. Studies in mice showed decreased chondrocyte proliferation and reduced capacity for chondrocyte apoptosis in growth plates in animals with a disrupted TRPS1gene [8].

Skeletalabnormalitiesoftenincludeosteopenia,whichincreasestheriskofstressfractures.Digitsof thehandexhibitradialorulnardeviation.Radiographicfindingsincludecone-shapedbaseepiphyses and shortened phalanges (Figure 1 and 2). Nail plates are thin and dystrophic. Short stature is also a typical feature [7, 9].

TheexactincidenceofTRPSisunknown.Bytheyear2017,therewereapproximately100casereports ofTRPSIandIIIandapproximately100casereportsofTRPSII.However,manycasesmayremain undiagnosed due to a mild phenotype [9].

IlizarovfixationisagloballyestablishedmethodusedtotreatdeformedandshortenedlimbS[10-13]. To date, there have been no literature reports on bilateral varus deformity correction and leg lengthening in a patient with TRPS I.

3. Casereport

A36-year-oldfemalewithTRPSIpresentedtoourorthopedicoutpatientclinicduetoseverepainatthemedialcompartmentsofher knee joints. Neither non-steroidal anti-inflammatory drugs nor physiotherapyhadbeeneffective.Physicalexaminationrevealeda marked varus deformity. There were no signs of knee instability; meniscal signs were negative; the estimated range of motion was 0–130degreesbilaterally.Hipmobility(bilaterally)andgaitwere normal. Imaging studies showed bilateral varus deformity with mechanical axis deviation (MAD) of 23 mm at the right and 25 mmattheleftkneejoint.Therewasnoabsolutelengthdiscrepancy inthelowerlimbs(Figure3).

Thepatientwasqualifiedtoundergosurgicalcorrectionthedeformitywiththellizarovmethod.Two4-ringIlizarovexternalfixators weremounted,withthehingesbilaterallypositionedatthecenters of rotation and angulation (CORA) determined via earlier radiography.Corticotomywasperformedintheproximaltibiae.Thesurgical procedure was performed under subarachnoid (intrathecal) anesthesia,withouttheuseofischemiatourniquet.Thepostoperativeperiodwasuneventful.Patientverticalizationwasinitiatedon postoperativeday1,andshewaspreparedforfullweight-bearing withtheuseofcrutches.Onpostoperativeday5,gradualcorrec-

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tion of the deformity via distraction rods was started. The patient was discharged home on postoperative day 6.

Afollow-upvisitinanoutpatientsettingthreeweeksaftersurgery revealed no disturbances in skin healing or signs of wire-track infection. Radiographic images of lower limbs (Figure 4) demonstrated improved MAD (the improvement was more pronounced in the right lower limb). Subsequently, the rate of distraction was modified. The range of motion at the knee joints was the same as beforesurgery.

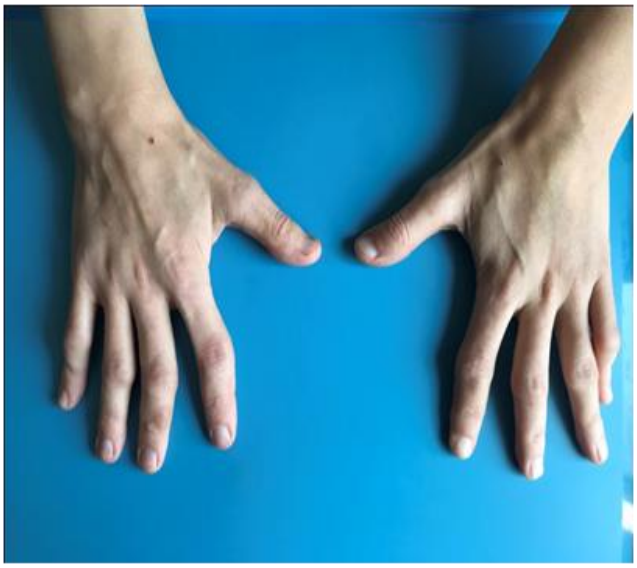

Figure1:Skeletal abnormalities often include osteopenia, which increasesthe risk of stress fractures. Digits of the hand exhibit radial or ulnardeviation.

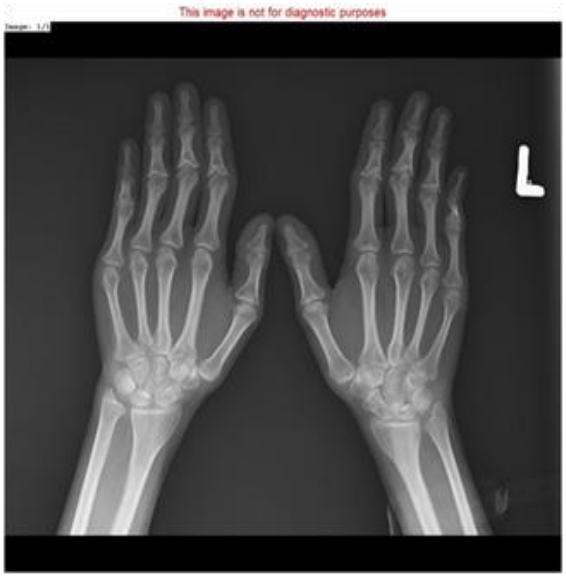

Figure2:Radiographic findings include cone-shaped base epiphy-ses and shortened phalanges

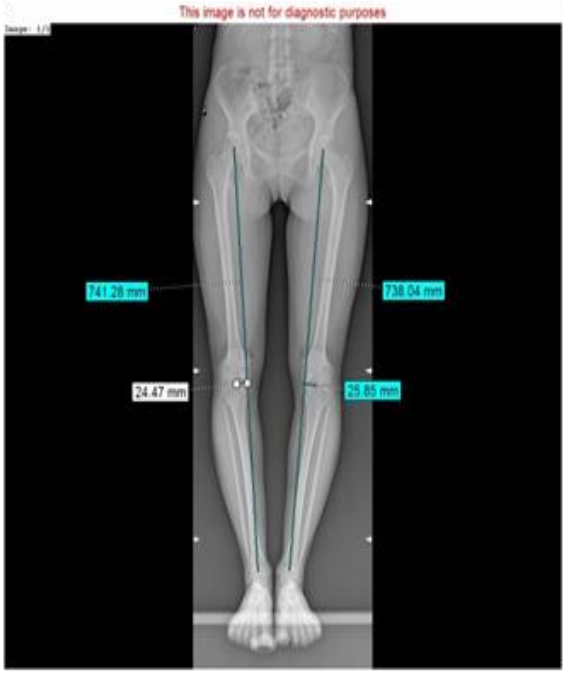

Figure3:Imagingstudiesshowedbilateralvarusdeformitywithmechanicalaxisdeviation(MAD)of23mmattherightand25mmattheleftkneejoint.Therewasnoabsolutelengthdiscrepancyinthe lower limbs

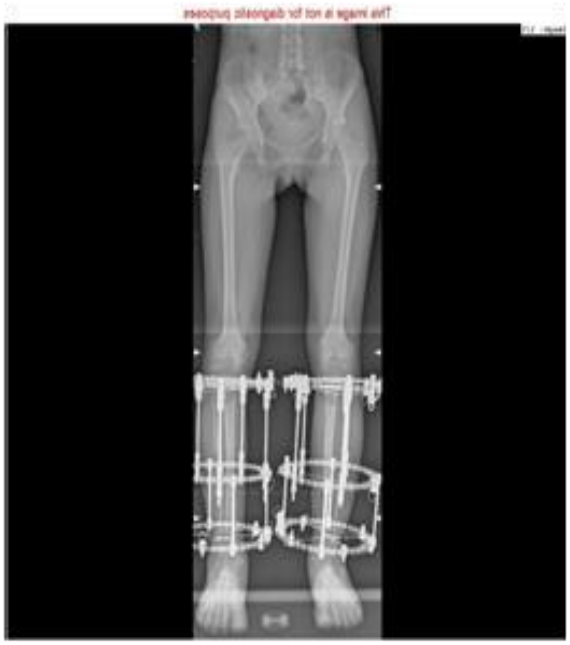

Figure 4: The improvement was more pronounced in the rightlowerlimb

A follow-up assessment three months after surgery showed bone unionatbothregenerates.TheIlizarovfixatorswerescheduledto beremovedonemonthapart.Followingtheremovalofonefixator, the patient was advised not to bear full weight.

Apanoramiclower-limbradiographtakenthreemonthsfollowing theremovaloftheotherIlizarovfixatorshowedbilateraldistalfib-

ularstressfracturesintheprocessofhealing(Figure5).Thetreat- ment resulted in no limb length discrepancy, and the mechanical axesshowedimprovement.Thepatientcouldwalkwithouttheuse of walking aids.

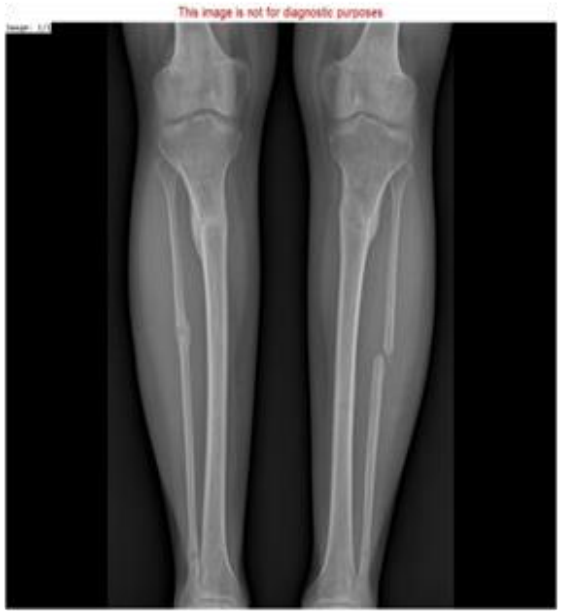

Figure 5: A panoramic lower-limb radiograph taken threemonthsfollowingtheremovaloftheotherIlizarovfixatorshowe dbilateral distal fibular stress fractures in the process of healing

Six months following Ilizarov treatment, both tibial regenerates showedgoodboneremodeling(Figure6).Thepatientcouldwalk withoutpain,bilaterallydemonstratedphysiologicalrangeofmo- tioninthekneeandanklejoints,andplannedtoresumework.Fol- low-upwasdiscontinuedtwelvemonthsfollowingtheremovalof thesecondIlizarovfixator.Atthefinalfollow-upvisit,thepatient could walk without pain and had resumed her work as a waitress.

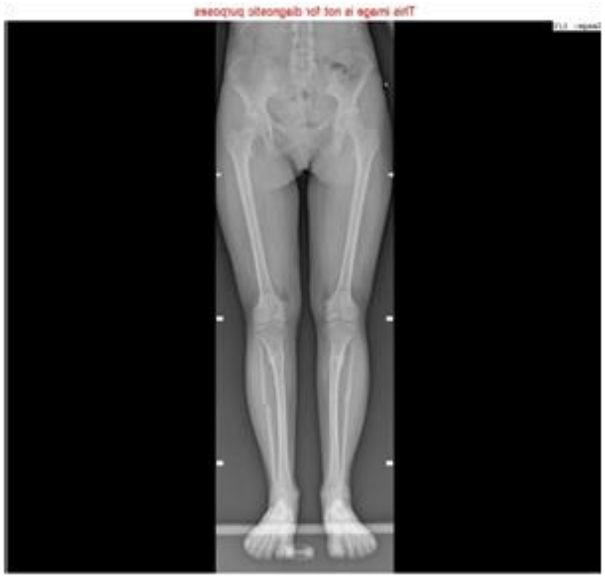

Figure6:SixmonthsfollowingIlizarovtreatment,bothtibialregene-rates showed good bone remodeling

4. Discussion

TRPS is a rare skeletal dysplasia, with autosomal dominant pat- ternofinheritance.TheconditioninvolvesadefectingeneTRPS1, which encodes a transcription factor for a zinc-finger protein that stabilizesnucleicacidmolecules.Hisgeneticdefectdisruptschon- drocyte proliferation and apoptosis leading to growth retardation and limb deformities [3, 14, 15].

TherearethreeknowntypesofTRPS.TypesIandIIIproducesim- ilarphenotypesanddifferonlyinseverity.Typicalfeaturesinclude short stature; sparse, thin hair; a characteristic pear-shaped nose; and a long philtrum. Skeletal abnormalities include phalangeal deformities,proximalfemurgrowthdisturbance(coxavara),coxa magna, and deformities mimicking those in Legg-Calvé-Perthes disease. TRPS type II is characterized by multiple osteochondro- mas and mental retardation [14].

Patients with TRPS require multispecialist care, as the syndrome maybeassociatedwithendocrinedisorders:hypothyroidism,idio- pathichypoglycemia,orgrowthhormonedeficit[14].Growthhor- monetherapymaybeeffectiveintreatingshortstature,providedit is initiated sufficiently early [3].

The patient described in this case report presented at our orthope- dic outpatient clinic with bilateral knee pain located at the medial compartmentsofthekneejoints.Physicalexaminationrevealeda marked varus deformity of both legs, with no signs of instability in any of the lowerlimb joints. The rangesof motion were within normallimits.Imagingstudiesshowednoabnormalitiesoftheslig- aments,articularcartilage,ormenisci.Thepatientwasqualifiedto undergo a gradual deformity correction, with lengthening of both legs.Inlightofthelackofliteraturereportsontreatingbonedefor- mitiesinTRPSpatientswiththeIlizarovmethod,weplannedlimb lengtheningofupto3cmbasedonourownexperienceintreating skeletaldysplasias.

The surgical technique of limb elongation was developed by Co- divillaandpopularizedbyWagnerinthe1970s.Initially,thetech- nique was associated with a number of complications [16]. The discovery of distraction osteogenesis by Prof. G.A. Ilizarov wasa breakthrough in skeletal dysplasia treatment; and introductionofaringexternalfixatorgreatlyimprovedtreatmentoutco mesall over the world. Although it has already been over 65 years since thepublicationofdistractionosteogenesisprinciples,inthehands of experienced doctors Ilizarov external fixators still are effective tools for treating challenging deformities, bone nonunion, and bone defects [17-20].

Therehavebeennumerousreportsonthetreatmentoflowerlimb deformitieswiththeIlizarovmethod.Theyemphasizethefactthat achieving good treatment outcomes is dependent on careful patientqualificationfortreatment,patients'collaboration,andcommitmenttotheinconveniencesassociatedwithtreatment.Skeletal dysplasia treatment should be provided only in specialist centers staffed by experienced personnel and having access to rehabilitation equipment [21-23].

The case study presented above is the first report on distraction osteogenesis used in the treatment of this rare genetic syndrome. The Ilizarov method seems to help achieve good results in treating limb shortening and limb deformities in patients with TRPS. Further studies and a recruitment of more patients with TRPS are needed to establish the standards of orthopedic treatment for this condition.

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