Gait Pattern and Lower Extremity Alignment in Children With Diastrophic Dysplasia

Ilhan A. Bayhan, MD, Mehmet S. Er, MD, Tristan Nishnianidze, MD, Colleen Ditro, APN, Kenneth J. Rogers, PhD, ATC, Freeman Miller, MD, and William G. Mackenzie, MD

Background: The aim of this study was to describe the dynamic lower extremity alignment in children with diastrophic dysplasia (DD) by 3-dimensional gait analyses. Our main hypothesis was that gait kinematics and kinetics are different than the age-normalized population and patellar dislocation can alter the gait in patients with DD.

Methods: A retrospective review of clinical data and radiographs was conducted for patients with DD who had gait analysis before lower extremity skeletal surgery excluding foot procedures. Lower extremity range of motion was measured. The Pediatric Outcomes Data Collection Instrument (PODCI) was administered to parents to evaluate their children's functional status. Gait laboratory data were collected to compare the hip and knee kinematics in cases with and without patellar dislocation. Anteroposterior standing radiographs were taken for all patients to assess the correlation between measurements (clinical, radiologic, and gait) for coronal knee alignment.

Results: Thirty lower extremities of 15 children (7 females and 8 males) were evaluated. The mean age was 7.4 ± 3 years, the mean height was 97.7 ± 15 cm ($z = -5.1$), and the mean weight was 20.6 ± 6.2 kg ($z = -0.8$). The DD PODCI subscores were statistically significantly lower ($P < 0.05$) than the average stature for developing children, except for the happiness score. Gait analysis, compared between all DD and an age-normalized average stature group, showed decreased forward velocity, step length, and stride length with an increased average forward tilt of the trunk and pelvis, hip flexion, hip adduction, and internal rotation ($P < 0.001$). Delta hip and knee motion were also decreased ($P < 0.001$). The patella was dislocated in 19 (63.3%) and central in 11 (36.6%) knees. Comparison of the minimum knee and hip flexion at the stance phase demonstrated increased crouch gait in the patellar dislocation group ($P < 0.001$). Knee alignment measurements between clinical examination and gait analysis showed moderate correlation (r, 0.476; $P = 0.008$).

Conclusions: Children with DD demonstrated lower PODCI subscores except for happiness. Gait analysis showed limited lower extremity function of the children with DD in our study group. Patella dislocation group had increased crouch gait.

Levels of Evidence: Level III—diagnostic study.

Key Words: diastrophic dysplasia, gait, lower extremity alignment

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Diastrophic dysplasia (DD) is a rare, autosomal recessive inherited dysplasia that occurs from a genetic defect in chromosome 5q that encodes diastrophic dysplasia sulfate transporter protein.[1,2] "Diastrophic," meaning distorted, refers to the morphologic anomalies of the ears (cauliflower ear), spine, long bones, and hands and feet.[3] Although the phenotypic expression varies, in most cases, a typical pattern of deformity can be identified.[4] The extremities can show rhizomelic shortening. Cervical kyphosis is seen in one-third to one-half of the patients, and bifid posterior elements of the lower cervical spine are seen. Scoliosis can develop in at least one-third of the patients and can be very severe if it develops when the patients are below 3 years of age. The shoulders and radial heads may be subluxated. A short, proximally placed and triangular first metacarpal, named the hitchhiker thumb deformity, can also be seen at the first toe. The proximal interphalangeal joints of the fingers are often fused (symphalangism).[5,6] Neonatal hip dislocation can occur. The hips typically have a persistent flexion contracture with a short and broad femoral neck. The femoral head may be flat and often has a double-hump deformity.[6,7] Knees usually have flexion contractures with associated ligamentous contracture, epiphyseal narrowing, and irregularity.[8] Excessive genu valgum and knee instability can be observed.[9] The patellofemoral complex can be shifted laterally, with similar findings as congenital dislocation of the patella.[8–10] Children with progressive genu valgum, knee flexion contracture, and extensor lag typically have a lateral patellar dislocation. Foot deformities can be variable with equinus, equinovalgus, skewfoot, forefoot varus, etc.[3] Clinical appearance of the foot deformity can mimic the clubfoot; however, radiologically, it is not similar. Severe equinus can be seen at the hindfoot with a valgus-oriented subtalar joint and

From the Nemours/Alfred I. duPont Hospital for Children, Wilmington, DE.

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Reprints: William G. Mackenzie, MD, Department of Orthopaedics, Nemours/Alfred I. duPont Hospital for Children, P.O. Box 269, Wilmington, DE 19899. E-mail: william.mackenzie@nemours.org.

laterally displaced navicular on the talus.[4] Joint range of motion can diminish as early as 5 years of age,[9] with early osteoarthritis of weight-bearing joints.[11]

Instrumented gait analysis is a helpful tool for the evaluation of lower extremity alignment.[12,13] The aim of this study was to describe the dynamic lower extremity alignment in children with DD who also had clinical and radiologic evaluation. Our main hypothesis was the gait kinematics and kinetics would be different than the normal population and that patella dislocation can alter the gait in patients with DD. We also evaluated the correlation between measurements (clinical, radiologic, and gait) for the coronal knee alignment.

METHODS

The study was approved by the institutional review board. Patients younger than 18 years of age and diagnosed with DD who had gait analysis before lower extremity bony surgeries from 2005 to 2014 were identified. A retrospective review of clinical examinations, gait analysis, and radiographs was conducted.

Gait Analysis

Cleveland clinic marker sets and 8-camera motion analysis system (Motion Analysis Corp, Santa Rosa, CA) with realtime and orthotrack software were used for kinetic and kinematic measurement and calculations. Ten to 20 gait cycles for each patient, while he or she was walking at a self-selected pace, were recorded. Data from 2 AMTI force plates (AMTI, Newton, MA) were used to calculate kinetic data from kinematics. Kinetic data were not available for patients with short stride length who were using assistive devices. By using kinematic variables, dynamic hip and knee joint flexion-extension angles were compared in cases with and without patellar dislocations.

Clinical and Functional Measurements

Assistive devices used were noted. Physical examination was performed by a physical therapist who measured the range of motion of the lower extremity joints before gait analysis.

Health-related Quality of Life Outcome Score

The Pediatric Outcomes Data Collection Instrument (PODCI) was completed by parents of patients before gait analysis to evaluate their children's functional status.[14] The PODCI scores of 2 children could not be located.

Radiologic Measurements

Anteroposterior standing radiographs were taken for all patients. Coronal plane alignment was measured using digital templating software (TraumaCad, Novapacs, Salt Lake City, UT)[15] on a light picture archiving and communications system workstation (Koninklijke Philips Electronics, Eindhoven, the Netherlands). Knee varus and valgus malalignment was measured using the anatomic axis of the femur and the anatomic axis of the tibia.

Statistical Analysis

Statistical analysis was performed using SPSS version 22 (SPSS Inc., Armonk, NY). Normality of the variable distribution was tested with the Kolmogorov-Smirnov test. Continuous variables were compared using the Student t test. Pearson correlation coefficient was used to test for the correlation of knee alignment between radiologic, clinical, and gait variables. Statistical significance was set as $P < 0.05$.

RESULTS

Thirty-six children with DD were identified; 30 lower extremities of 15 children (7 females and 8 males) were evaluated. Age, height, and weight means at gait analysis were 7.4 ± 3 ($z = -5.15$), 97.7 ± 15 cm ($z = -5.1$), and 20.6 ± 6.2 kg ($z = -0.8$), respectively. Surgeries before gait evaluation are listed in Table 1. Assistive device use in daily life included the following: walker (8 patients, 53%) and cane (1 patient, 6.6%).

PODCI Scores

The PODCI scores were compared with scores of average stature children as described by Haynes and Sullivan.[16] The mean scores for children with DD were lower than those of average stature children. The difference was statistically significant in all domains except the happiness score (Table 2).

Passive Range of Motion

Lower extremity passive range of motion measurements are listed in Table 3. The mean flexion contractures at the hip, knee, and ankle were 18.7 ± 11.7, 19.1 ± 16.4, and 20 ± 25.6, respectively.

Gait Analysis

Temporal Spatial Data

Forward velocity, step length, and stride length were decreased compared with those of the normal population with statistical significance ($P < 0.001$). Similar results were observed when the parameters were height-adjusted and normalized ($P < 0.001$, Table 4).

Kinematics

The average forward tilt of the trunk and pelvis, hip flexion, hip adduction, and internal rotation were increased with statistical significance ($P < 0.001$). Delta hip

TABLE 1. Surgeries Before Gait Evaluation

Surgery	N (%)
CSTR	8 (26)
LCS	2 (6.6)
SMO	2 (6.6)
MFO	2 (6.6)
Hip reduction	1 (3.3)
GRI	3 (20)

CSTR indicates complete subtalar release; GRI, spinal growing rod instrumentation; LCS, lateral column shortening; MFO, mid foot osteotomy with external fixations; SMO, supramalleolar osteotomy.

TABLE 2. Pediatric Health Assessment (Parent)—13 Patients

	DD Standardized		
	Mean	SD	*P*
Extremity and physical functioning	61.7	27.2	< 0.0001
Transfer and basic mobility	67	25.5	< 0.0001
Sports and physical functioning	75	41.1	0.048
Pain/comfort	59	29.4	< 0.0001
Happiness	75	14.5	0.065
Global functioning	57.3	23.6	< 0.0001

DD indicates diastrophic dysplasia.

motion (maximum hip flexion and maximum hip extension during stance) was decreased compared with the delta hip motion of the normal population ($P < 0.001$, Table 4). Knee flexion, valgus, and external rotation angles were increased ($P < 0.001$). Delta knee motion (maximum knee flexion and maximum knee extension during stance) was also decreased similar to the hip joint ($P < 0.001$). Ankle dorsiflexion was decreased compared with that of the normal population as well ($P < 0.001$, Figs. 1, 2).

The average age was 6.4 years in patients with patellar dislocation (19 knees, 63.4%) and 9 years in patients in the nondislocated group (11 knees, 36.6%). Comparison of the minimum hip and knee flexion in the stance phase showed significant difference between children with and without patellar dislocation. The average hip flexion was 52.3 ± 14 in the dislocated and 20.2 ± 18 in the nondislocated group ($P < 0.001$). Higher average knee flexion for the dislocated group (42.2 ± 16) was observed compared with the nondislocated group (12.6 ± 11.4, $P < 0.001$).

Kinetics

Kinetics values were collected from only 11 lower extremities of 6 children. Knee and ankle flexion moments were decreased with a statistical significance ($P < 0.001$). The same results were observed at the hip ($P = 0.004$) and ankle powers ($P < 0.001$, Table 5).

Correlation Analysis

Lower extremity alignment angles are listed in Table 6. The mean knee valgus angle was 11.7 ± 13 on standing radiograph, 13 ± 7 on clinical examination, and 12.9 ± 8 on gait analysis. Correlation between the groups was significant only in clinical examination and gait analysis with moderate strength (r, 0.476; $P = 0.008$).

DISCUSSION

The primary objective of this study was to compare dynamic gait evaluation of the children with DD with the normal population. It is common for this patient population to have surgical interventions for their foot deformities in early life.[3,4,17] Ninety percent of our patients had foot surgeries before their gait analysis.

Sixty percent of the patients were using an assistive device (walker, cane, etc.) for walking. Sponseller and colleagues reported 12% using a wheelchair; however, for longer distances, 44% used an assistive device (scooter, wheelchair, walker etc.).

The PODCI was developed by the American Academy of Orthopaedic Surgeons, American Academy of Pediatrics, Pediatric Orthopaedic Society of North America, and Shriners Hospitals for Children to assess pediatric musculoskeletal conditions.[14] To our knowledge, no classification system has been described to assess the functional status of children with DD. In all domains except happiness, PODCI scores were lower than those of average stature children. We believe that the PODCI is a useful tool for not only comparing with the average stature population but also evaluating surgical outcomes.

In patients with osteochondrodysplasias, early joint degeneration is one of the major causes of disability.[18] In DD, delayed ossification and distortion of the epiphysis causes early degeneration and deformation of the hip and knee joints.[9] Range of motion begins to decrease as early as 5 years of age.[7,9,19] Most patients need hip and knee replacement in early adulthood.[20,21]

Defining knee flexion-extension, varus-valgus, and hip rotation in these children with complex deformities in

TABLE 3. Passive Joint Range of Motion Recorded Before Gait Analysis

Movements (deg.)	Mean	SD	Maximum	Minimum
Hip flexion	117.0	13.7	140	90
Hip extension	−18.7	11.7	5	−40
Hip abduction	24.3	14.3	50	−7
Hip internal rotation	21.2	24.8	90	−18
Hip external rotation	42.7	19.9	85	0
Knee flexion	134.0	19.6	150	95
Knee extension	−19.1	16.4	5	−48
Ankle dorsiflexion (with knee flexion)	−18.0	25.7	25	−70
Ankle dorsiflexion (with knee extension)	−20.0	25.6	20	−70
Ankle plantar flexion	42.9	17.9	75	5
Ankle inversion	33.7	19.4	60	0
Ankle eversion	20.9	26.3	75	−30
Tibial transmalleolar axis	14.6	17.6	50	−20
Forefoot adduction	−10.8	11.0	10	−32
Calcaneal inversion	11.2	13.5	35	−20
Calcaneal eversion	4.5	13.2	30	−22

TABLE 4. Kinematic Results

Variables	Diastrophic Dysplasia Mean	SD	Normal Population Mean	SD	*P*
Forward velocity (cm/s)	61	25.5	115	5.5	< 0.001
Normalized forward velocity (L/s)	0.61	0.23	0.95	0.08	< 0.001
Step length (cm)	25.7	8.4	49.3	5.5	< 0.001
Normalized step length	0.25	0.06	0.40	0.01	< 0.001
Stride length (cm)	51.7	16.1	98.3	10.9	< 0.001
Normalized stride length	0.52	0.12	0.81	0.01	< 0.001
Trunk lateral tilt (deg.)*	2.7 shoulder up	4.1	1.88 shoulder up	1.01	0.25
Trunk lateral tilt (deg.)†	4.9 shoulder down	4.4	2.2 shoulder down	1.03	< 0.05
Trunk forward tilt (deg.)‡	14.9 forward	12.2	2.02 forward	1.43	< 0.001
Pelvis rotation (deg.)*	12.1 hip forward	8.14	6.89 hip forward	0.99	0.001
Pelvis rotation (deg.)†	12.2 hip trailing	8.3	7.0 hip trailing	0.86	0.001
Pelvis forward tilt (deg.)‡	26.6 anterior	5.7	13.6 anterior	1.81	< 0.001
Hip flexion (deg.)†	40.5 flexion	22.4	−3.7 extension	2.29	< 0.001
Hip flexion (deg.)*	65.5 flexion	16.3	40.6 flexion	2.95	< 0.001
Hip abduction (deg.)‡	1.16 abduction	10.3	5.2 adduction	1.45	0.002
Hip rotation (deg.)‡	17.4 external	13.8	5.9 external	3.4	< 0.001
Delta hip motion (deg.)	24.9 flexion	8.3	44.3 flexion	1.6	< 0.001
Knee flexion (deg.)†	31.3 flexion	20.7	5.5 flexion	2.0	< 0.001
Knee flexion (deg.)*	56.3 flexion	22.6	35.7 flexion	2.3	< 0.001
Knee abduction (deg.)‡	12.9 valgus	8.58	5.14 valgus	2.17	< 0.001
Knee abduction (deg.)*	7.07 valgus	9.01	1.7 valgus	2.41	0.004
Knee abduction (deg.)†	17.3 valgus	7.64	9.4 valgus	2.29	< 0.001
Tibial torsion (deg.)‡	14.2 external	19.8	2.8 external	3.5	0.004
Delta knee motion (deg.)	24.9	6.9	30.1	1.11	< 0.001
Ankle flexion (deg.)*	0.83 dorsiflexion	15.9	13.3 dorsiflexion	1.43	< 0.001
Ankle flexion (deg.)†	9.9 plantarflexion	12.9	5.4 plantarflexion	1.27	0.072
Ankle rotation (deg.)‡	12.9 internal rotation	9.5	0.9 external rotation	2.48	< 0.001
Foot orientation (deg.)‡	14.3 external	21.2	7.2 external	1.81	0.075

*Maximum during stance.
†Minimum during stance.
‡Average during stance.

all 3 planes can be difficult. We follow the widely accepted model that the knee joint functions close to a hinge therefore this axis of principle motion is first defined, the range of motion around this axis is defined as flexion-extension, the residual orthogonal angle with maximum extension is varus-valgus, and the rotation of the knee axis relative to the pelvis is termed hip internal and external rotation. This is the same model for the physical examination and kinematics. It is somewhat difficult though in some children with DD to accurately define this knee joint axis.

Vaara and colleagues found 23 degrees of hip flexion contractures, which was similar to our study. However, for the knee joint, Peltonen and colleagues found a

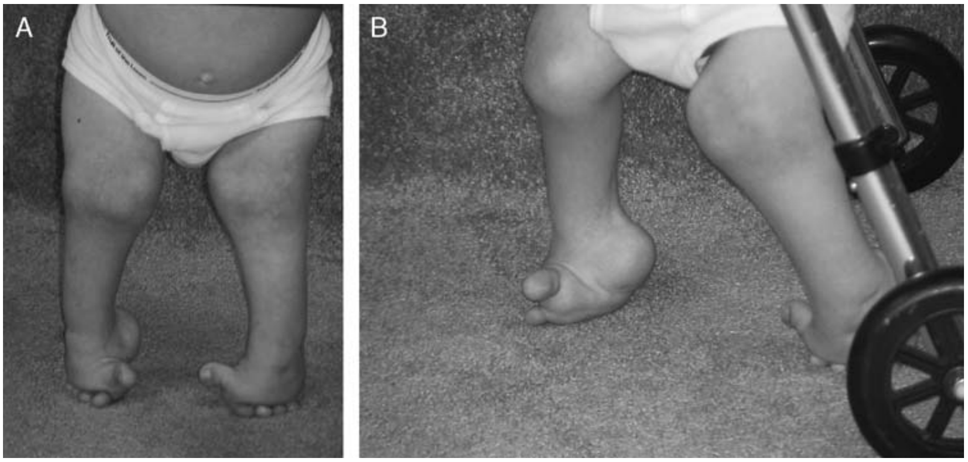

FIGURE 1. Lower extremity alignment in diastrophic dysplasia patient. Coronal (A) and Sagittal plane (B).

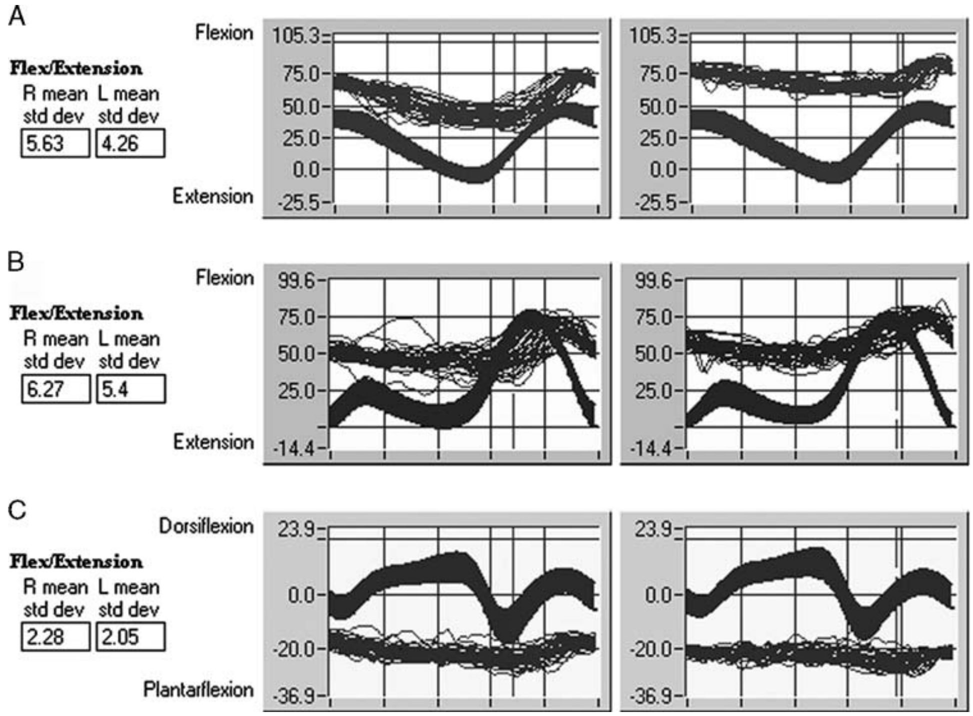

FIGURE 2. Diastrophic dysplasia. Hip (A), knee (B), ankle (C) kinematic graphics. Graphics shows flexion contractures with insufficient joint movements.

considerable amount of variation in the degree of flexion contracture. In our study, the mean knee flexion contracture was 19.1degrees.

Knees with patellar dislocation showed significant hip and knee flexion contractures. We believe lateralization of quadriceps extensor moment causes early progression of joint deformity. In the case series of Langenskiold and Ritsila,[10] children with surgical treatment of congenital dislocation of the patella showed improvement in function. We believe that early surgical alignment of the extensor patellar mechanism can slow the progression of the joint deformities. Also the valgus alignment of the lower extremities should be corrected either with guided growth techniques or with osteotomies.

Interpretation of radiographs, especially at the knee joint, is difficult because of joint contracture, delayed ossification, and an abnormal epiphysis. Vaara et al[22] also mentions that information provided by plain radiographs is often inadequate. In this study, the valgus deformity measured on standing radiographs did not correlate with the clinical and gait analysis measurements. We believe that the radiographs may include rotational problems in which the knee is not projected in a true anteroposterior view, which can cause measurement errors. Three-dimensional

TABLE 5. Kinetic Results

	Diastrophic Dysplasia		Normal Population		
Variables	**Mean**	**SD**	**Mean**	**SD**	***P***
Hip flexion moment (N m/kg)*	0.65 flexion	0.27	0.70 flexion	0.04	0.593
Hip abduction moment (N m/kg)*	0.63 abduction	0.266	0.62 abduction	0.04	0.983
Knee flexion moment (N m/kg)*	0.215 flexion	0.12	0.56 flexion	0.059	< 0.001**
Knee abduction moment (N m/kg)†	0.01 varus	0.15	0.05 valgus	0.009	0.192
Knee abduction moment (N m/kg)*	0.145 valgus	0.10	0.20 valgus	0.01	0.072
Knee abduction moment (N m/kg)‡	0.19 varus	0.18	0.11 varus	0.006	0.151
Ankle flexion moment (N m/kg)*	0.065 flexion	0.04	1.14 flexion	0.09	< 0.001**
Hip power (W/kg)*	0.84	0.37	1.26	0.04	0.004**
Ankle power (W/kg)*	0.56	0.47	2.64	0.24	< 0.001**

*Maximum during stance.
†Average during stance.
‡Minimum during stance.
**$P < 0.01$.

TABLE 6. Lower Extremity Alignment Angles

	mLPFA	mLDFA	mMPTA	mLDTA	JCLA
Mean (deg.)	96.8	95.3	92.8	81.93	7.77
Min-Max (deg.)	58-146	71-136	75-126	60-102	0-28

JCLA indicates joint line congruency angle; mLDFA, mechanical lateral distal femoral angle; mLDTA, mechanical lateral distal tibial angle; mLPFA, mechanical lateral proximal femoral angle; mMPTA, mechanical medial proximal tibial angle.

gait analyses can be helpful for correct measurement as demonstrated in previous studies.[12,13]

This study includes the lower extremity analyses of the patients with DD before their corrective surgeries. Although our patient numbers were adequate to demonstrate the gait pattern of this group of patients, a larger sample size would permit a more accurate representation of the broader population of patients with DD.

In conclusion, the work presented in this study is useful for showing the dynamic lower extremity alignment and limited extremity function of children with DD as compared with the normal population. We believe that our study will provide data to guide surgical interventions, compare outcomes improvement, and maintain mobility.

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