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Proximal Femoral Focal Deficiency: Radiologic Analysis of 49 Cases[1]

Proximal femoral focal deficiency, an uncommon congenital anomaly, necessitates early radiologic classification for surgical planning and treatment. Objective radiographic criteria, including femoral length index, acetabular depth index, acetabular angle index, and shape of the proximal femur were determined in 49 patients before cartilaginous ossification of the femoral capital epiphysis; final classification was based on follow-up radiographs or findings at arthrography or surgery. These parameters were analyzed to determine the accuracy and contributions of each in classification. Correct classification into one of three groups was possible in 86% of cases with use of three of the parameters: femoral length index, acetabular depth index, and shape of the proximal femur. The acetabular angle was found to contribute insignificantly to classification. Magnetic resonance imaging, used in only one case, depicted the nonossified cartilaginous femoral capital epiphysis, thus obviating the need for invasive diagnostic procedures and facilitating early classification.

Index terms: Acetabulum, 442.159 • Children, skeletal system • Femur, abnormalities 443.1484, 443.1499 • Hip, abnormalities 442.1499 • Hip, MR Studies, 442.1214

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PROXIMAL femoral focal deficiency (PFFD) is a congenital partial absence of the proximal end of the femur with shortening of the entire limb. The etiology of this disorder is uncertain; it has received little attention in the radiologic literature. The diagnosis and classification have been based mainly on plain radiographic findings. This method does not permit definite classification during the 1st year of life. Because arthrography permits the presence or absence of a femoral head to be confirmed, it has been used as an adjunctive tool (1). Cineradiography can also be used to evaluate the degree of hip stability.

Several classification schemes based on the anatomic relationship between the acetabulum and the proximal end of the femur have been proposed. Aitken's system (2), which is probably the most often cited, designates four classes:

Class A: In this least severe type, the femoral head is present and attached to the shaft by the femoral neck. The femur is shortened (as in all types), and a coxa vara deformity is present. The cartilaginous neck is not seen on early radiographs but later ossifies. Occasionally, the cartilaginous connection between the neck and the shaft forms a subtrochanteric pseudarthrosis (Figs. 1, 2).

Class B: In this type, the acetabulum is "adequate" or moderately dysplastic and contains the femoral head. At maturity, no osseous connection is seen between the femoral head and the shaft. The femoral segment is short and usually has a bulbous bony tuft (Figs. 1, 3).

Class C: The acetabulum is severely dysplastic in this form. The femoral head is absent or is very small and not attached to the femoral shaft. The shortened femoral segment has a tapered proximal end (Figs. 1, 4).

Class D: This is the most severe form, with absence of the acetabulum and proximal femur. No proximal tuft is present (Fig. 1).

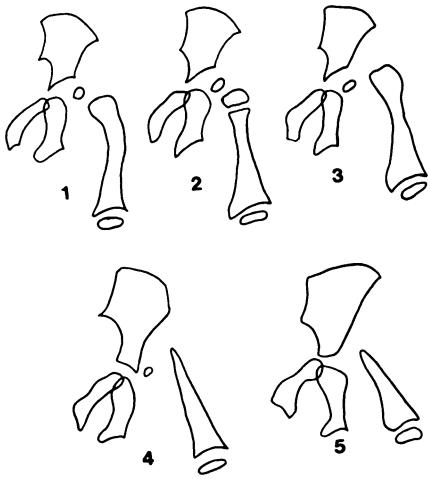

Figure 1. Amstutz's classification (3) of PFFD into five types based on the presence and location of the femoral head and neck and the degree of acetabular dysplasia. Types 1 and 2 correspond to Aitken's class A; type 3, to class B; type 4, to class C; and type 5, to class D.

Amstutz (3) further subdivided Aitken's classification into five types. He divided class A into types 1 and 2. Type 1 is reserved for the milder form with simple femoral shortening and coxa vara (Fig. 1). In type 2, a subtrochanteric pseudarthrosis is present (Fig. 2). The remaining types correspond to those of Aitken's classification.

Alternative classifications have been proposed by Gillespie and Torode (4) and Hamanishi (5) on the basis of eventual function and surgical approach. These researchers believe that the classification may change with the varying degree of ossification that occurs with growth.

Aitken's and Amstutz's classifications are the most widely used. Since the management of type 1 varies from that of type 2, a distinction between them is necessary (6). We, therefore, classified our cases using

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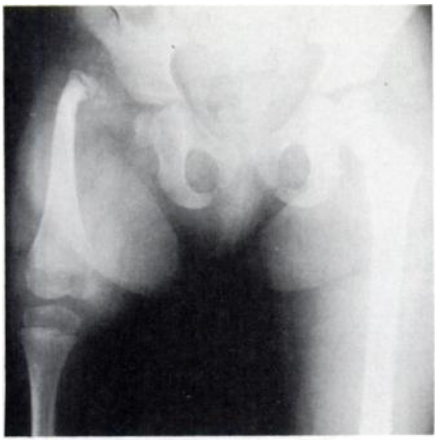
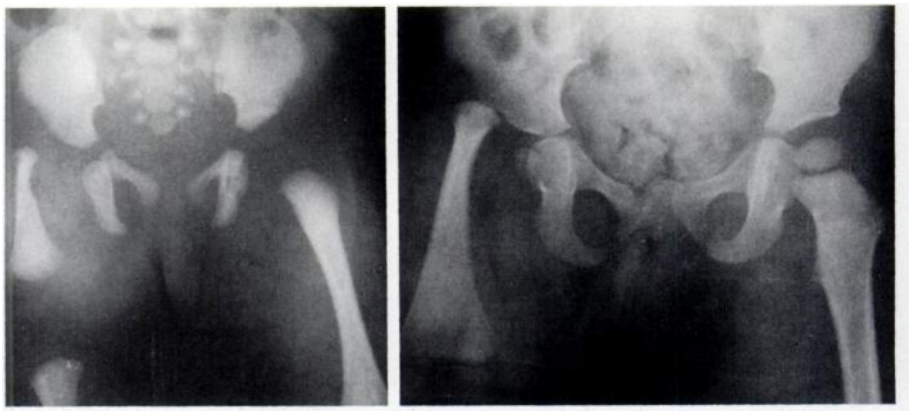
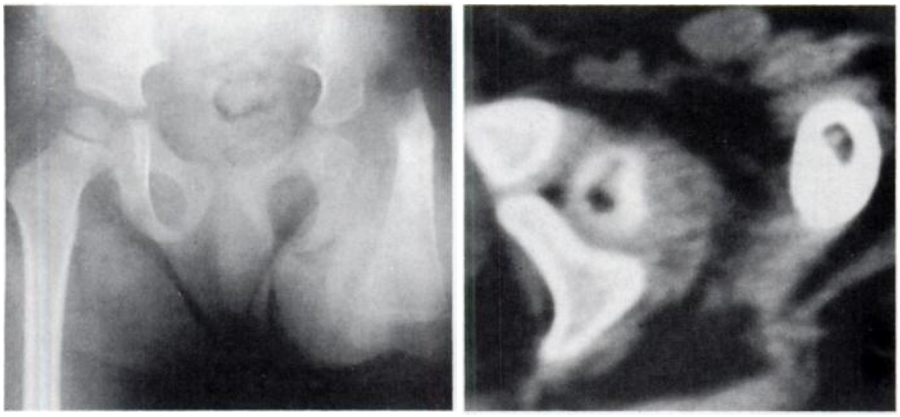


Figure 2. Radiograph of a 4-year-old girl with Amstutz type 2 deformity. Note coxa vara and subtrochanteric pseudarthrosis with sclerosis. Absent fibula is a commonly associated anomaly.



a. b.

Figure 3. Images of a child with Amstutz type 3 deformity. **(a)** Initial presentation at birth. Right femoral head is not ossified, and proximal femoral shaft is bulbous. Classification was uncertain at this time. **(b)** Later radiograph shows presence of ossified femoral head within acetabulum. No ossification of the femoral neck had occurred by age 4 years.



a. b.

Figure 4. **(a)** Left proximal femoral shaft in this 4½-year-old girl is tapered, and femoral head is not visualized. This might be either Amstutz type 4 or type 5 deformity. **(b)** Later CT scan of left hip at age 10 reveals presence of femoral head, allowing classification as type 4. No femoral neck ossification is seen.

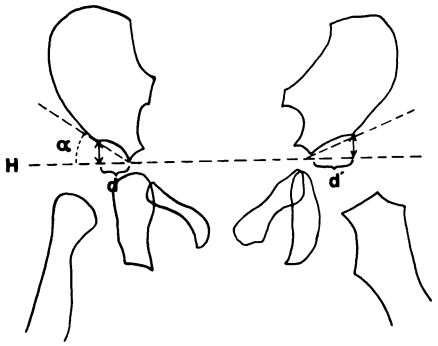


Figure 5. The distance (d) on Hilgenreiner's (9) line (H) from the triradiate cartilages to the perpendicular drawn from the edge of the acetabulum of the involved side divided by the distance on the normal side (d') was defined by Koman et al. (8) as the shelf index. This value was then normalized $[(d' - d)/(d' + d)]$ and defined as the acetabular depth index. The angle (α) between Hilgenreiner's line and the acetabular rim was measured.

the Amstutz method.

Classification of PFFD becomes important in planning treatment, which depends on the stability of the hip as determined by the presence of the femoral head and its relationship to the shaft. In patients with type 1 or 2 deficiency, a stable hip joint can be achieved. Surgical procedures are aimed at correction of the varus deformity, the pseudarthrosis, and the leg-length discrepancy. An osteotomy at the site of the subtrochanteric pseudarthrosis can correct the varus deformity and possible excessive anteversion of the femoral neck. King (7) reported favorable results with use of a metaphyseal-epiphyseal synostosis in patients with type 3 deficiency. In types 4 and 5, a stable hip joint cannot be obtained; fusion of the hip and, possibly, the knee joint is necessary. These conversions are combined with above- or below-the-knee amputations; leg-length inequality is then corrected with prostheses. The functional implications of associated anomalies, most commonly fibular hemimelia and/or foot deformities, must also be considered in surgical planning. Initial surgery is frequently performed before the age of 2 years to minimize psychological trauma to the patients and to allow them to adapt to the amputation. Obviously, such early surgery requires timely classification.

Objective criteria for early classification have been proposed in previous studies (8). Shelf index, acetabular index, and acetabular dysplasia have been used as predictors of pelvifemoral stability at maturity. We statistically analyzed these parameters in our patient population and, using discriminant analysis, determined the relevance of each. We also determined the frequency of misclassification based on these parameters. In addition, we discuss a case in which magnetic resonance (MR) imaging aided in early classification prior to ossification of the proximal capital femoral epiphysis, obviating the need for arthrography.

MATERIALS AND METHODS

The clinical charts and radiographs of 49 patients from the Philadelphia Shriners Hospital for Crippled Children were studied. This group includes all patients with PFFD treated at that institution from 1966 to 1985. The right femur was involved in 27 patients; the left, in 16 patients. Six patients had bilateral involvement. Thirty-three (67%) were boys, and 16 (33%) were girls. Two patients were black, eight were Hispanic, and one was Asian; the remaining 38 were white. The patients were initially evaluated for PFFD at varying ages. The majority (60%) were seen during the 1st or 2d year of life.

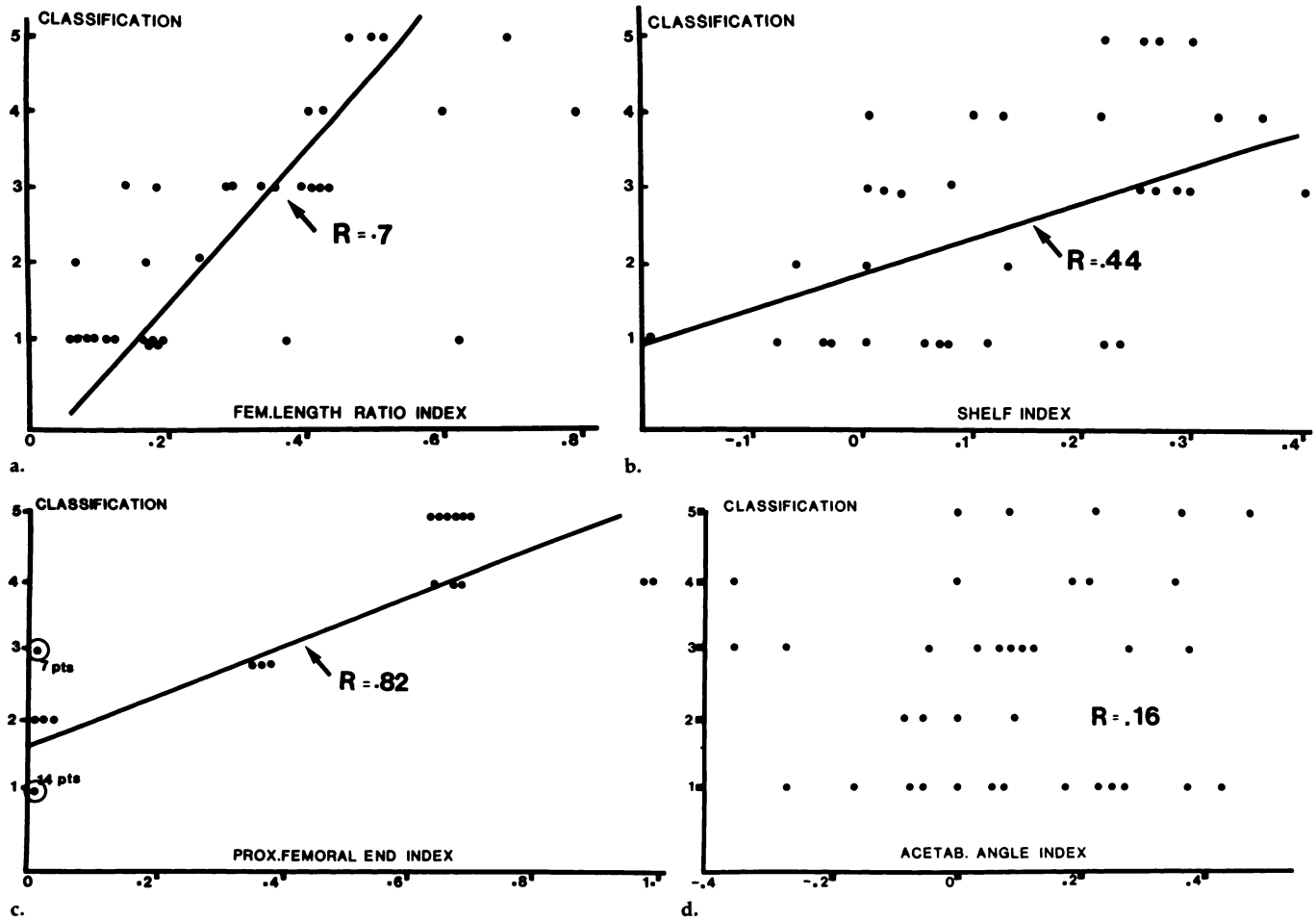


Figure 6. **(a)** The index of femoral length correlated well with the Amstutz type (r = .7). **(b)** The acetabular depth index had a poorer correlation (r = .44). **(c)** The shape of the proximal femur (proximal femoral end index) correlated well with type. **(d)** The acetabular angle index correlated poorly with type.

Some patients had been referred from other institutions and were seen when they were older. Several patients had been referred for surgery from the Shriners Clinic in Puerto Rico. All of these patients had undergone early radiography, usually shortly after birth, and these images were used for analysis.

Radiographs from the patients' initial examinations were evaluated. Several parameters were analyzed to determine the accuracy of each in classifying the disease. These included the femoral length, acetabular angle, acetabular depth (shelf index), and shape of the proximal femur. The contribution of each parameter alone and in combination with the other parameters was determined. The femoral lengths (the distance from the most proximal ossified bone to the inferior border of the medial condyle) of the normal side and of the abnormal side were measured. The acetabular angle and the acetabular depth were measured (Fig. 5). The shape of the proximal femur was classified as bulbous, tapered, or absent. Occasionally, the distinction between bulbous and tapered was indeterminate. These subjective parameters were assigned the following point values: bulbous = 3, indeterminate = 2, tapered = 1, absent = 0. The ratio of the difference between the normal and abnormal sides to the sum of each side [(normal − abnormal)/(normal + abnormal)] was determined for each parameter, except in the case of acetabular angle, for which the ratio (abnormal − normal)/(normal + abnormal) was determined. These normalized values were considered indexes of the parameter. The bilateral cases were eliminated from this analysis.

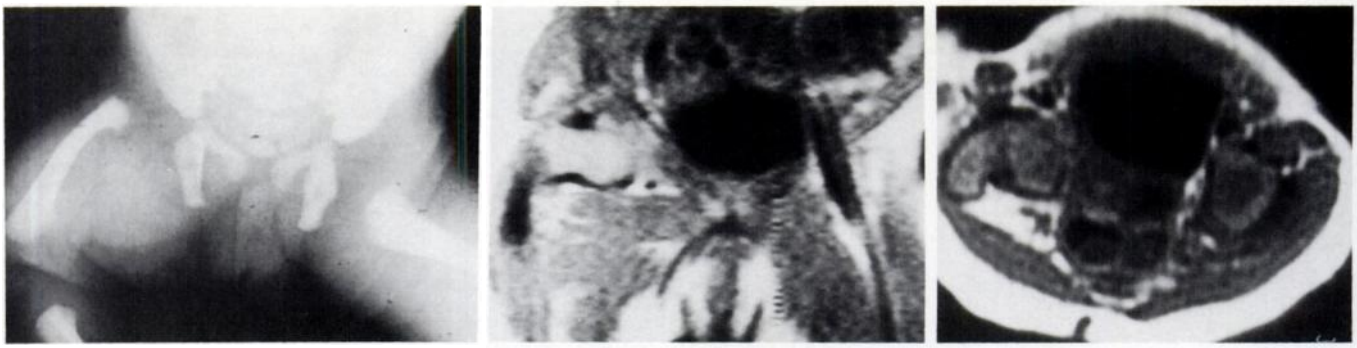
Patients were classified into Amstutz's groups on the basis of their final radiologic and clinical outcome. All images and information obtained while the patient was evaluated at Shriners Hospital were studied. Cases from more recent years were included only if early classification could be established (i.e., less severe types). Comparison of the initial type with the "final" one was made.

Stepwise discriminant analysis was performed on these four parameters with use of the SPSSX program on an IBM (Atlanta) 4381 computer. A "confusion matrix" was generated to analyze misclassification. Correlation was made between each index classification and the final classification.

RESULTS

Of the 15 patients with type 1 disease, nine were correctly classified on initial presentation. On retrospective evaluation, only one of the five type 2 patients, three of the ten type 3 patients, one of the eight type 4 patients, and three of the five type 5 patients were correctly classified. Often, the classification was not mentioned in the patients' charts or radiology reports. Although patients were examined at various ages, ratios comparing the abnormal and normal sides were obtained to minimize discrepancies because of growth. The growth ratio between the abnormal and normal femur has been shown to be constant (3).

Two patients had paternal relatives with a "short leg." We were unable to determine if these relatives had PFFD. One patient's mother had "clubfoot." No other genetic basis or predisposing factors were identified. Associated anomalies were detected in 30 of the 49 patients. The most common was ipsilateral fibular hemimelia, which was present in nine patients. Metatarsus adductus was present in six patients. Various other anomalies were present in 12 patients, including cleft palate, upper extremity hemimelias, scoliosis, and



a. b. c.

Figure 7. **(a)** Plain radiograph of a 1-year-old infant with a short right femur. Proximal femoral end is bulbous. This may be Amstutz type 1, 2, or 3. On coronal **(b)** and axial **(c)** T1-weighted 500/28 (repetition time msec/echo time msec) MR images, cartilaginous femoral head and neck are clearly visible; thus deformity is Amstutz type 1 or 2.

the Pierre Robin syndrome.

Statistical analysis of the femoral length index showed that it varied linearly with type (r = .7) (Fig. 6a). The acetabular angle index was found to be an unreliable indicator of type (r = .16) (Fig. 6d). Acetabular depth index correlated somewhat better with type (r = .44) (Fig. 6b). The shape of the proximal end of each abnormal femur was found to correlate well with type (r = .82) (Fig. 6c).

Univariate analysis revealed that there were significant differences (P < .05) in the mean variables for shape of the proximal femur, femoral length index, and acetabular depth index among each of the five types. The acetabular angle index was not significantly different among the types. Multivariate F statistics were used to compare the group means as part of discriminant analysis. The four variables were analyzed; when the shape of the proximal femur and acetabular depth index were selected by the discriminant technique, all types differed significantly (P < .05) except for types 1 and 2. Evaluation of the five types with a "confusion matrix" yielded a correct classification for 60% of the patients.

Because surgical management is dependent on hip stability, it is clinically important to separate patients into three groups: stable hip (types 1 and 2), unstable hip (types 4 and 5), and indeterminate status of the hip (type 3). To achieve this and to increase the sample size of the groups tested, types 1 and 2 were combined to form a new group I; type 3 was renamed group II; and types 4 and 5 were combined to form group III. There were 20 patients in group I, ten in group II, and 13 in group III. The patients with bilateral PFFD were not considered in the analysis. With this new structure, each pair of groups differed significantly (P < .02) when the shape of the proximal femur, femur length index, and acetabular depth index were included in the analysis.

A "confusion matrix" was determined for the three groups (Table 1). Eighty-six percent of the cases were correctly classified with the acetabular depth index, femur length index, and shape of the proximal femur. The latter two were the strongest discriminant variables.

Table 1
Confusion Matrix Based on Shape of the Proximal Femur, Acetabular Depth Index, and Femoral Length Index

Actual Group	No. of Cases	Predicted Group Membership (%)		
		I	II	III
I	20	85	15	0
II	10	30	70	0
III	13	0	0	100

DISCUSSION

PFFD is an uncommon disorder infrequently seen by most radiologists. Accurate early classification can be difficult because it is frequently based on the subjective evaluation of plain radiographic findings. Using our relatively large patient population, we evaluated objective parameters and determined the accuracy of classification into class types. Koman et al. (8) discussed the use of shelf index, acetabular index, and the degree of acetabular dysplasia to predict function at maturity. We found the acetabular index (r = .16) to be only minimally helpful in separating less severe types.

Using discriminant analysis, we found that if patients were classified into three groups, accurate classification could be made on the basis of the acetabular depth index, femur length index, and shape of the proximal femur in 86% of cases. With a larger population, it may be possible to further differentiate them into five groups. However, we were unable to do this with any significant predictive value. We found that the shape of the proximal femur and the femoral length index were the most useful parameters in classification. The presence of an acetabulum does indicate that a femoral head will ossify eventually. This is most useful in distinguishing between Amstutz types 4 and 5 since, by definition, no acetabular development occurs in type 5. Types 1 and 2 cannot be distinguished radiographically at an early age.

Amstutz (3) has shown that the growth ratio between the normal femur and the abnormal femur is constant. We also found the femoral length index to be a good predictor of final classification. The initial shape of the proximal femoral shaft had the best correlation (r = .82). We believe that, radiographically, the type of femoral end, the length of the femur, and the amount of acetabular dysplasia (based on the depth of the acetabulum) are the most useful parameters in early classification.

Several authors (10–12) have found that hip arthrography and cineradiography are useful aids in evaluating hip stability. Only one of our patients underwent arthrography. Several were examined with "push-pull" films. Three patients were examined with computed tomography (CT). CT was useful in evaluating acetabular dysplasia. However, it was found to be generally unhelpful in determining the presence of an unossified femoral head or neck. Arthrography is invasive and involves the inherent

risks of joint infection, trauma, and irradiation. We examined one of our infants with MR imaging (Fig. 7). Axial and coronal 500/28 images demonstrated a cartilaginous head and its connection to the femoral shaft. These features were not visible on the plain radiograph. The stability of the femur and hip can, therefore, be determined before ossification occurs. MR imaging provides optimal contrast for differentiating cartilage from other soft tissues. We believe that this will obviate arthrography and may eliminate the need to change classifications with growth, as has been suggested by other authors (4, 5), because one will be able to detect early the presence of a femoral head and connecting neck. We suggest that MR imaging may become the method of choice in the early classification of PFFD. ■

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