Pathogeny and natural history of congenital dislocation of the hip

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A R T I C L E   I N F O

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- Congenital dislocation
- Natural history
- Pathogenesis

A B S T R A C T

Based on a review of the literature, the authors have made a critical study of several etiological factors. Endogenous factors such as acetabular dysplasia, increased anteverision of the femoral neck, and capsular laxity support the genetic theory but are neither constant nor necessary and are only facilitating factors. The major factor seems to be a mechanical one linked to the position in the uterus: hyperflexion with adduction and external rotation constituting the dislocating foetal posture combined with abnormal pressure on the greater trochanter and leading to expulsion of the head upward and backward. This theory can explain the natural history of CDH which is first, at birth a hip instability followed by two possible evolutions: either persistent luxation becoming irreducible or spontaneous stabilisation leading sometimes to complete healing or to residual abnormalities (subluxation or dysplasia). This concept suggests practical conclusions: the importance of an early diagnosis, the selection of the signs of the hip at risk, the pattern of prevention, the role for non-clinical investigations, the principles of the treatment based on postures, the indications for the different types of treatment.

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Although many factors have been incriminated in the aetiology of congenital hip subluxation, dislocation, and dysplasia, none of the pathogenic hypotheses is fully satisfactory. The confusion resulting from this uncertainty generates a number of issues:

- terminological issues, since the classic term “congenital dislocation of the hip” (CDH) is widely considered to be inappropriate and is often replaced by “dislocating malformation”, “dislocating dysplasia” or, more recently, “developmental displacement of the hip” (DDH)[Klisic [30]];
- diagnostic issues, as the meaning of the clinical instability remains controversial and no consensus exists about the role for radiography and, above all, for ultrasonography [Bonnard [5], Graf [25], Téot and Deschamps [57,58]];
- public health issues, because depending on the pathogenic concept the strategy should focus either on prevention [Klisic [29,30]] or on screening;
- and treatment issues, since the natural history of CDH largely dictates the therapeutic indications. The main concern is the risk of overtreatment that places a costly burden on society and can induce iatrogenic complications (avascular necrosis of the femoral head). In addition, improved knowledge of the cause of the dislocation facilitates the conduct of the treatment by providing a sound underlying rationale, thereby diminishing the reduction failure rate.

Based on published work by Le Damany [34], Faber [21], Ortolani [37], Salter [43], and Tachdjian [56], CDH is usually viewed as a hip development abnormality that starts in utero and becomes apparent at birth or within the first few post-natal months (Table 1).

As clearly demonstrated by Gardner [23], true dislocation cannot occur in the embryo before the formation of the articular cleft. Clavert [12] administered antimitotic agents to pregnant rabbits during the period of hip embryogenesis and obtained a marked increase in the prevalence of CDH, suggesting that CDH might be due to an overall hypoplastic malformation of the hip region. This hypothesis is not confirmed, however, by everyday clinical practice.

Davies [16], Dyson [20], Ilfeld [28], and Tredwell [61] concurred with Wynne Davies [66] that CDH may exist as two distinct aetiological forms: a form due to joint hyperlaxity that can be detected at birth based on perception of a positive Ortolani manoeuvre and a form related to subluxating acetabular dysplasia, which escapes detection by neonatal screening and is diagnosed only at a later age.

However, our experience with neonatal CDH screening and efforts to identify clinical signs that are more subtle than the palpable clunk during the Ortolani manoeuvre [44]; our anatomic [45], radiographic [46–48] and, more recently, ultrasonographic [49] studies; and a critical review of the literature lead us to suggest a unifying theory of the pathogenesis of CDH.


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1. Study of the various aetiological factors

The aetiological factors fall into two categories, endogenous or constitutional factors and exogenous or mechanical factors.

1.1. Endogenous factors

These consist in a primary hip abnormality that may involve the acetabulum, femur, or joint capsule. The existence of endogenous factors suggests a role for genetic susceptibility, which may explain the greater frequency of CDH in girls (5 girls for 1 boy) and in certain geographic areas, ethnic groups, and families (3% to 12% of cases). When one twin has CDH, the frequency of CDH in the other twin is 40% for monozygotic twins and 3% for dizygotic twins [Carter [8], Idelberger [27]].

Thus, although genetic factors cannot fully explain the occurrence of CDH, they play an undeniable role [Fuhrmann [22]]. The genetics of CDH in France have been thoroughly investigated by Le Marec and Roussey [35], who ruled out monogenic inheritance but also found that a multifactorial theory was unsatisfactory. Nevertheless, they indicated that the table developed by Stalder [54], which implies a multifactorial pathogenic process, is an easy means of estimating the genetic risk: about 10% in siblings of a boy with CDH and 3% in those of a girl with CDH, and about 5% in the first-born child when one of the parents has CDH.

1.1.1. Acetabular dysplasia

The hip dysplasia theory advocated by Faber [21], Courtois [15], and Klisic [30] long prevailed. This theory constitutes the rationale for the ultrasound studies conducted by Graf [25]. Nevertheless, many lines of evidence argue against it.

• Experimental studies in animals during the growth period have established that hip dislocation induced by abnormal lower limb position [Asplund [2], Michelsson [36], Salter [42], and Yamamura [52]] or by damage to the connecting structures of the hip joint [Langenskiold [32], Sibrandj [50] and Smith [52]] cause secondary acetabular deformities. The acetabular is distorted into an oval shape, and wear creates a dislocation groove located either in the antero-superior sector if the hip was extended (Salter) or in the postero-superior sector if the hip was left in flexion (other experiments). The acetabular dysplasia is reversible if the femoral head is returned to its normal position in the cavity. In all these experiments, the dysplasia was induced by the dislocation, contraindicating the theory that dysplasia causes the dislocation.

• In studies on the natural history of hip dysplasia without dislocation in infants [Coleman [13], Geiser [24], Pratt [38], Seringe and Hass [47]], none of the patients progressed to subluxation or gradual dislocation (Fig. 1).

• Comparisons of the outcomes of treated and un-treated hips [Seringeand Hass [47]] established a distinction between secondary dysplasia (due to the dislocation), which was reversible; and primary dysplasia, which was not influenced by the treatment (Fig. 2).

Table 1: Classical view of the natural history of CDH (from Salter).

<table>
<thead>
<tr>
<th>In utero</th>
<th>Shallow acetabulum</th>
<th>Dysplasia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth</td>
<td>Femoral anteversion</td>
<td>Dislocatable hip</td>
</tr>
<tr>
<td></td>
<td>Hip no longer flexed</td>
<td></td>
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<tr>
<td></td>
<td>Joint laxity</td>
<td></td>
</tr>
<tr>
<td>After birth</td>
<td>Gradual hip extension</td>
<td>Dislocation</td>
</tr>
<tr>
<td></td>
<td>Tight swaddling</td>
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</tr>
</tbody>
</table>

Fig. 1. Natural history. (A) At 4 months of age, bilateral acetabular dysplasia is visible and the proper centring of the femoral heads is in doubt (in part due to the external rotation of the femurs). (B) Appearance at 6 months of age. (C) Continued spontaneous improvement at 15 months of age. (D) The hips are normal at 6 years of age.
Studies of foetal ultrasonograms by Téot and Deschamps [57,58] support a mechanical cause to the acetabular dysplasia, consisting of abnormal pushing forces through the femoral head in the postero-superior quadrant of the acetabulum.

However, as underlined previously by Le Damany [34] and subsequently by Ralis and McKibbin [40], the depth of the acetabulum deserves consideration. The acetabulum seems to be deeper in black Africans than in Caucasians [Skirling [51]]. This fact probably explains the lower frequency of CDH among blacks (0.49%) than among Caucasians (1.53%) reported by Arzt in New York City [1]. Thus, an excessively shallow acetabulum may be a constitutional factor that increases the risk of dislocation. Instead of “primary dysplasia”, we therefore prefer the term “dysmorphism”, which points to a specific acetabular shape [4,64]. In any case, acetabular depth is not a major causative factor.

1.1.2. Excessive femoral neck anteversion

The abnormality is located in the femoral shaft, and not in the neck. Excessive femoral neck anteversion is not a consistent feature of CDH (Seringe and Kherrat [45,53]) and consequently cannot be a major causative factor.

Experimental studies in animals have been inconclusive.

Variations in femoral anteversion had no noticeable effects on the acetabulum in studies by Smith [52], Cahuza [7], in contrast, found that the induction of excessive femoral anteversion in growing dogs resulted in deformities affecting the antero-superior portion of the acetabulum, with progression to early osteoarthritis but not to dislocation.

Wilkinson [65] reported that imm gelation in tight flexion and external rotation (as in the frank breech presentation) was followed by posterior dislocation with femoral neck retroversion (due to an intra-cervical deformity), a condition that shared no similarities with clinical observations.

We believe, nevertheless, that excessive femoral anteversion can play a non-negligible role in the mechanical theory of hip dislocation, as it can be likened to external rotation of the hip (whose importance in foetal positions associated with hip dislocation will be discussed below).

1.1.3. Joint hyperlaxity

Joint hyperlaxity is clearly not the inciting cause, since hip dislocation is not a consistent feature of Ehlers-Danlos syndrome or idiopathic generalised hyperlaxity.

Furthermore, the hyperlaxity does not affect the entire capsule. Instead, the postero-superior part of the capsule is stretched (Seringe and Kherrat [45]). Similar to the acetabular dysplasia, this abnormality seems secondary to the femoral head displacement and not the cause of the dislocation.

The hypothesis that hyperlaxity may be related to a hormone (relaxin) [Thieme [59], Tonnis [60], Von Rosen [63]] has led to numerous investigations, whose results are conflicting. However, in animals, Wilkinson [65] showed that the induction of hip dislocation by a specific joint position was easier after the parenteral administration of sex steroids. This result was confirmed by Suzuki and Yamamuro [55].

The frequency of inguinal hernias is increased 5-fold in girls and 3-fold in boys with CDH compared to the rest of the population [Uden [62]]. This fact suggests a connective tissue abnormality that simultaneously explains the development of CDH and that of inguinal herniation.

We believe that joint hyperlaxity is a predisposing factor whose relationship to hormonal factors remains unproven. Joint hyperlaxity might be due to a genetic abnormality of the hip connective tissue, which might explain both the potential hyperlaxity of the capsule and the increased malleability of the labrum (with more or less reversible deformities).

1.2. Exogenous or mechanical factors

The mechanical and postural theory defended by D. Browne [6], Laurence [33], and Dunn [17] appears the most convincing since, in contrast to genetic factors, it can explain the increased frequency of CDH in first-born babies; babies born in the breech presentation; babies with high birth weights; and babies with foetal deformities, torticollis, oligohydramnios, or foeto-maternal disproportion [17]. A well-documented case-report by Kohler [31] describing CDH after a tubal pregnancy further supports this theory.

1.2.1. Mechanical factors active in utero

Mechanical factors are clearly active in fetuses that are in the frank breech position (with the knees extended), since CDH is extremely common in this situation (20% to 25%). Two different explanations have been put forward:

- Wilkinson [65] underlined the deleterious effect of external rotation of the lower limb, which was confirmed in experimental animal studies;
- and studies from Finland [36] and Japan [55,67] emphasized the role for the tension applied to the hamstring muscles in the frank breech position, which may cause dislocation of the hip.

Regardless of their validity, these hypotheses apply, according to their authors, only in the event of a frank breech position. Therefore, they are unsatisfactory, as we need an explanation that applies also to the complete breech position (with the knees flexed) and to the cephalic position.

However, our clinical studies [44] have led to the following observations:

- the hamstrings of a baby born in the frank breech position are consistently hypotonic and slack and, consequently, cannot play a role in the genesis of hip dislocation;
- in neonates with hip instability, external rotation of the lower limb with forced hip flexion dislocates the femoral head.
posteriorly, whereas internal rotation reduces the dislocation (regardless of the degree of flexion-extension of the knee);

- reconstitution of the foetal position in babies with CDH allows the identification of three postures associated with dislocation (Fig. 3): knees in extension or forced extension and external rotation, knees in semi-flexion and external rotation, and knees in forced flexion and neutral rotation and in contact with each other (but with excessive femoral anteversion, which is equivalent to external rotation). This study extends the work by Wilkinson [65] and the older investigations by Roser [41].

A dynamic study of hip instability on a cadaver specimen [45] allowed us to confirm this “dislocating foetal posture” concept, with little or no abduction or even and external rotation (or excessive anteversion) (Figs. 4 and 5). In addition, the bitrochanteric diameter of the foetus decreases when the lower limbs are in external rotation, suggesting that the position in external rotation is a direct consequence of the foeto-maternal mechanical conflict [45].

1.2.2. Mechanical factors during delivery

As shown by Dunn [18], Gardner [23], Walker [64], and our group [45], the normal neonatal hip is perfectly stable, and manual traction on the lower limb during delivery cannot cause CDH. Instead, manual traction would cause either separation of the proximal femoral epiphysis or a femoral shaft fracture.

Although Cheetman and Garrow [11] believe that dislocation is caused by the manoeuvres performed to screen for hip instability, it is worth pointing out that, to induce dislocation in a cadaver, continuous pressure must be applied to the femur for 3 to 6 hours (Hjelmstedt [26]). The duration and repetition of the clinical examination are never sufficient to induce dislocation of a normal hip.

1.2.3. Post-natal mechanical factors

Rabin [39] in 1965 and subsequently Salter [43] and Coleman [14] found a high frequency of hip dislocation in populations that use traditional swaddling techniques (tight wrapping with the lower limbs extended). Several authors such as Klisic [29] felt this finding might explain dislocations acquired during the first few post-natal months and recommended a form of prevention by routine swaddling with the hips abducted. This measure decreased the frequency of late diagnoses of hip dislocation.

However, studies by Barlow [3], Dunn [19], Rabin [39], and our group [44,49] on the natural history of hip instability have established that the instability resolves spontaneously in at least half the cases (Fig. 6). In addition, in our experience with the wait-and-see management of hip dysplasia without dislocation [47,49] and of congenital pelvic asymmetry [48], progression toward dislocation does not occur (Fig. 7).

The increased frequency of hip dislocation associated with traditional swaddling techniques may be ascribable to another factor: instead of causing hip dislocation, the forced extension of the lower limbs may prevent the spontaneous reduction of a hip that was dislocated in utero and remains unstable. This explanation is consistent with studies from Japan on the role for the psoas and hamstring muscles [55,67]. Similarly, the traditional practice of carrying infants on the mother’s back or hip with the lower limbs abducted explains the low frequency of hip dislocation in certain African ethnic groups (Skirving [51]).

2. Pathogenic theory

The mechanical theory seems to prevail. Before discussing its role in the pathogenesis of CDH, we will attempt to answer the following question:

2.1. How can mechanical factors cause hip dislocation?

Based on the experimental model of acquired hip dislocation in patients with neuro-muscular diseases and on experiments in animals [2,42,50,55,67] and post-mortem neonates [26], two factors must be present in combination:

- a position of the femur such that the head is not oriented toward the acetabular fossa but instead towards the edge of the cavity and the capsule: this is the dislocating position;
- a force that expels the femoral head and that may originate in active or spastic muscles in the case of paralytic dislocation, in passive muscle tensions created by casting in animal experiments, or in an external force applied to the femur in post-mortem studies of neonates.

The application of these facts to CDH is simple:

- the dislocating foetal posture combines the usual forced flexion, some degree of external rotation (or excessive femoral anteversion), and adduction;
- the force that expels the femoral head originates in two factors: muscle activity, which promotes dislocation in the above-described position (psoas, adductors, hamstrings, and rectus femoris muscles) and support on the greater trochanter, in keeping with the hypothesis put forward by Chandler as early as 1926 [10].

This simple and rational concept is in contradiction with the classical explanations put forward by Le Damany [34] and D. Browne [6], which were not satisfactory (Figs. 8 and 9).

2.2. Pathogenesis of CDH

We suggest the following pathogenic mechanism based on all the facts discussed above:

- Genetic factors are probably inconsistent. They may confer increased susceptibility related either to joint hyperlaxity (diminished mechanical strength of the labrum and capsule) or to a shallow acetabulum (acetabular dysmorphism).
- Mechanical factors play the preponderant role and may consist in application of a force to the greater trochanter while the femur is in a position conducive to dislocation. For all three dislocating postures, the two lower limbs may be symmetric or asymmetric, which may explain the occurrence of bilateral and unilateral...
dislocations. In the breech presentation, the force applied to the greater trochanter may come from contact with the matern- nal pelvic inlet, which may explain that the dislocation is often bilateral. In the cephalic presentation, the force may come from contact with the maternal lumbar spine, explaining the higher frequency of unilateral dislocation of the left hip, since the foetal back is usually on the left side [Dunn [17]].

Fig. 4. (A) Dislocating effect of external rotation of the femur when the hip is in forced flexion (foetal position); (B) horizontal cross-section through the pelvis showing posterior subluxation of the femoral heads.

Thus, dislocation of the hip may develop towards the end of the pregnancy (within the last few weeks or days or perhaps even during labour) under the influence of several mechanical factors, which may be fairly often combined with genetic susceptibility fac- tors. After birth, the hip is released from the forces applied in utero and tends to improve spontaneously. The clinical finding in this situation is hip instability (either reducible dislocation or dislocat- able hip). If the instability is sustained, the dislocation persists and gradually becomes irreducible. In about half the cases, the hip becomes stable spontaneously and the outcome is either a full recovery or the persistence of residual abnormalities, i.e., residual dysplasia and subluxation (Fig. 10).

3. Conclusions – Practical implications

The pathogenic process related to intra-uterine mechanical fac- tors and the improved understanding of the natural history of CDH increase our ability to resolve the issues related to this con- dition.

3.1. What terms should be used?

We believe the classical designation “congenital hip disloca- tion/subluxation” is appropriate, as it is defined by a variable degree

Fig. 5. (A) Re-centring effect of neutral or internal rotation (depending on the degree of femoral anteversion). (B) On the other hand, the foetal bitrochanteric diameter is increased (85 mm versus 68 mm in external rotation).
of prenatal femoral head displacement relative to the acetabular cavity. In contrast, the term “hip dysplasia”, used without qualifiers, is too vague, as pointed out by Catterall [9], and should be avoided. Nevertheless, the term “acetabular dysplasia” remains well suited to the acetabular abnormalities visualised by radiography or ultrasonography, although this term encompasses several clearly separate entities [Geiser [24], Seringe et al. [46]] (Table 2).

The adjective “teratological” is sometimes used to describe certain forms of hip dislocation but should be discarded, as it implies either an error in embryonic development or the development of hip dislocation in a baby with multiple malformations or deformities (severe oligohydramnios). Most of the cases classified as being ‘teratological’ are actually CDH cases that develop early during foetal development and are therefore already ‘irreducible’ at birth.

3.2. How can CDH be diagnosed early in life?

- At birth and during the first few post-natal weeks, the method of choice is the physical examination to look for hip instability. Dynamic sonography is a valuable adjunct in difficult cases.

| Table 2: Different patterns of acetabular dysplasia. |
|----------------|--------------------------------------------------|
| Types           | Interpretation                                  |
| Pseudo-dysplasia | Radiographic projection or congenital pelvic asymmetry |
| Primary dysplasia | Genetic dysmorphism                              |
| Secondary dysplasia | Consequence of the dislocation or subluxation |
| Residual dysplasia | Dysplasia that persists after treatment (and may have a primary component) |
3.3. What public health recommendations should be made?

Prevention, strictly speaking, is not feasible for this congenital disorder. Routine swaddling with the hips abducted was performed in many maternity wards in the 1970s but failed to substantially decrease the frequency of late diagnoses CDH. The only effective approach has been screening by repeated physical examination: the age at diagnosis has decreased with this approach, and additional decreases can be expected to occur. The physical examination should focus on detecting hip instability, whose presence confirms the diagnosis; and on identifying high-risk hips, which require particularly close monitoring. The criteria defining high-risk hips (Table 3) were selected based on pathogenic considerations. Apart from a family history suggesting a genetic factor, all these criteria are based on the intra-uterine postural theory.

Table 3
High-risk hips.

- Family history of CDH
  (Definite diagnosis in a first-degree relative)
- Breech presentation
- Other suggestive postures
  (Genu recurvatum, torticollis)
- Limited abduction
  (Less than 60°)
- Difficult abduction
- Asymmetric abduction

- Asymmetric abduction

Fig. 8. Early concepts of the mechanisms responsible for in utero hip dislocation. (a) Le Damany suggested that abnormal antero-posterior loading with the knee flexed might convert the femur into a class 1 lever via abnormal contact with the antero-superior iliac spine, driving the femoral head laterally and anteriorly, away from the acetabular fossa. (b) According to D. Browne, axilary pressure on the thigh might expel the femoral head downwards and posteriorly.

Fig. 9. Current concept of the mechanisms responsible for in utero hip dislocation. Pressure on the greater trochanter of the femur in external rotation (or with excessive anteversion) may drive the femur upwards and behind the acetabulum.

- Starting at 3–4 months of age, the physical examination remains of value but is less likely to detect instability, whereas the limitation of abduction is more marked. A standard radiograph of the pelvis to look for abnormal femoral head centring is an essential adjunct.

Fig. 10. Natural history of CDH according to the current concept.
Cases that are diagnosed late should no longer be classified as dislocating dysplasia cases that are not detectable by physical examination. Instead, they should be viewed as failures of screening related to several factors: insufficient muscle relaxation during testing for instability, failure to repeat the physical examination, or inadequate knowledge of certain clinical signs (piston movement or instability with no clunk, hypertonic adductors, and congenital pelvic asymmetry).

3.4. What are the principles of treatment?

The role for dislocating postures in the pathogenesis of CDH clearly supports the induction of postures associated with centering of the head. In babies, a position in hip flexion at 90°, abduction at 60° to 70°, and either neutral or moderate internal rotation is appropriate. Care should be taken to avoid external rotation, which prevents re-centering of the head. Pathogenic considerations also explain the need for continuous and sufficiently prolonged use of an abduction device to obtain retraction of the capsular dislocation pocket, avoid recurrent dislocation, and correct the secondary acetabular dysplasia. The constraints associated with the therapeutic position can impair the blood supply to the femoral head, thereby inducing a risk of avascular necrosis of the femoral head.

For this reason, and because a number of mild hip abnormalities can improve spontaneously, abduction therapy should be considered only in patients with a definite diagnosis of CDH or marked subluxation. In doubtful cases, a better strategy consists in clinical monitoring combined with sonography or radiography depending on the age of the infant.

Disclosure of interest

Authors’ disclosure of conflict of interest was not requested when the article was originally published.

References


