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Epidemiological, Clinical and Radiological Features of ACL Aplasia:

a review of the literature

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We confirm that we have no financial or other interest in the subject which may be considered as constituting a real, potential or apparent conflict of interest

Introduction

- Anterior Cruciate Ligament (ACL) aplasia or hypoplasia or agenesis or absence is a rare congenital malformation, with a reported incidence of 0.017 per 1000 people.
- In 1956, Giorgi was the first author to describe such deformity. Since then, many other Authors have reported analogous cases.
- Its absence is more common than generally suspected and ca be often found in connection with other abnormalities of both bone (patella, tibia, fibula, femur dysplasia i.e) or soft tissue in the lower limb, as well as organ syndromes (i.e. thrombocytopenia absent radius syndrome (TAR), arthrogryposis).
- Since the body adapts to the congenital defects, many patients may not experience joint instability until a knee twisting injury occurs. Consequently, with the exception of patients displaying proven limb disorders or other organ diseases, past medical history is often silent and the patient is generally referred to the Orthopedic due to the instability felt after a trauma.

Purpose

The aim of the present study is to classify the epidemiological, clinical and radiological features most frequently related to ACL aplasia, in order to understand if a correct diagnosis may be feasible before the surgical approach.

Methods

- We have identified articles from Pubmed, Science Direct, Medline and Scopus databases, with the keywords “Anterior”, “Cruciate”, “Ligament”, “ACL”, “Aplasia”, Hypoplasia”, “Agenesis”, “Absence”, “Congenital”, “Knee”.
- We included all articles reporting about ACL deformities.
- We excluded studies reporting about other types of knee anomalies but ACL.
- We also excluded articles in languages not
- comprehensible by the authors.

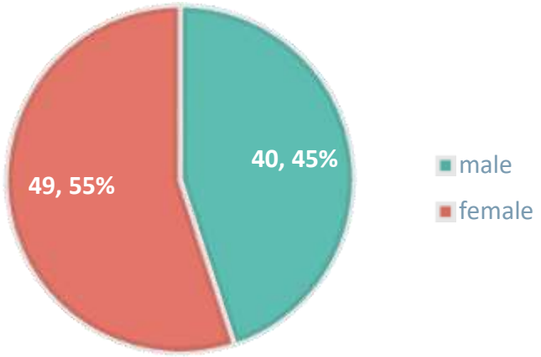
Results: Demographics*

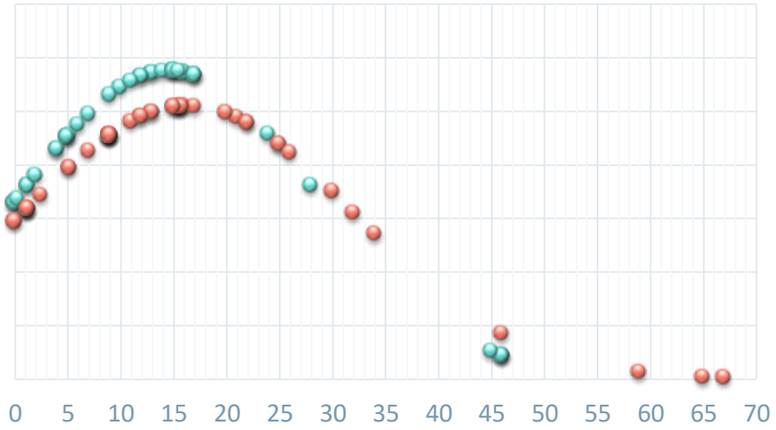
** We had to exclude the data collected from 2 articles from these results, as there were not reported the gender of the patients:*
(DeLee JC, Curtis R. Anterior cruciate ligament insufficiency in children. Clin Orthop Relat Res. 1983 Jan-Feb;(172):112-8. PMID: 6821976;
Sachleben BC, Nasreddine AY, Nepple JJ, Tepolt FA, Kasser JR, Kocher MS. Reconstruction of Symptomatic Congenital Anterior Cruciate Ligament Insufficiency. J Pediatr Orthop. 2019 Feb;39(2):59-64. doi: 10.1097/BPO.0000000000000940. PMID: 28178094.)

Number of cases

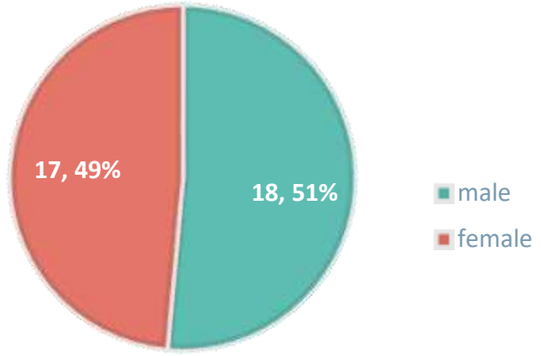
Age distribution at diagnosis

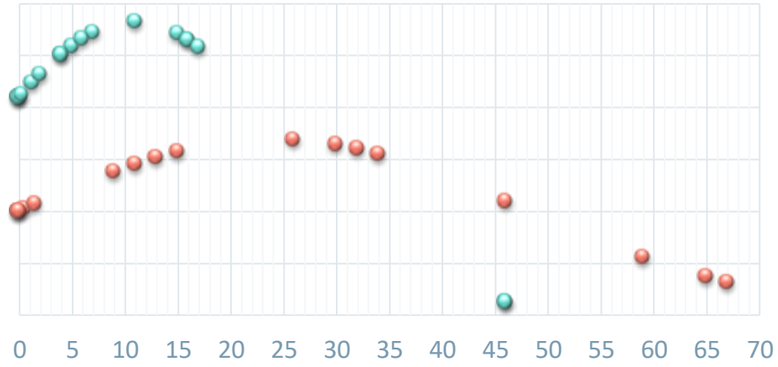
Total sample





Bilateral





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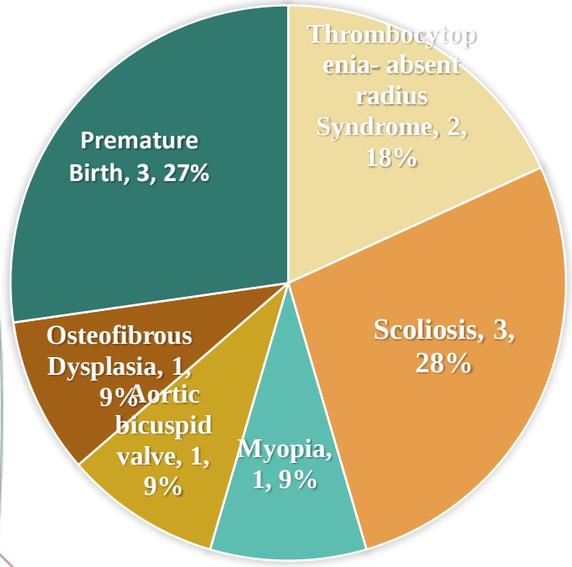
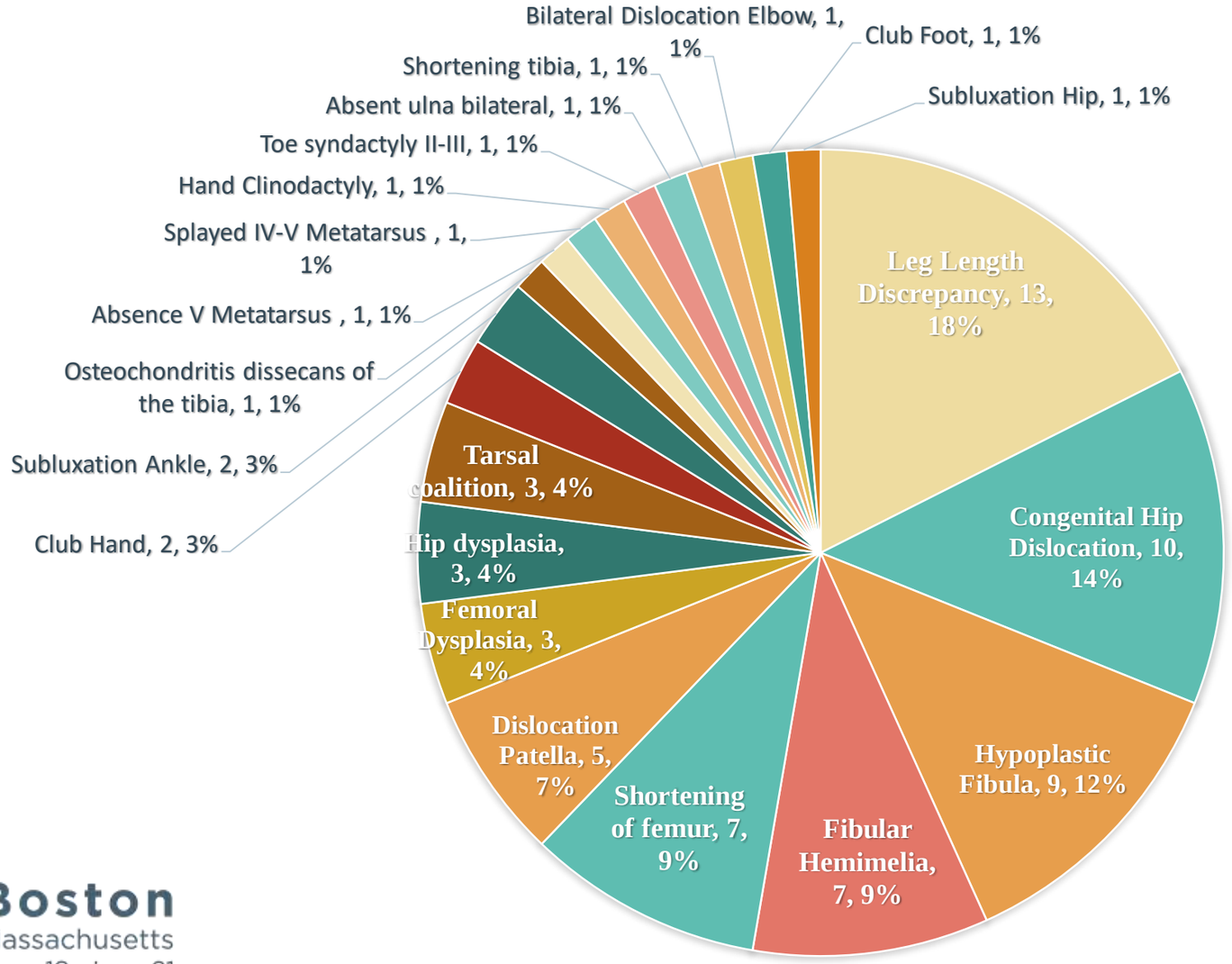
Results: Associated Conditions

Pathologies



Limb Anomalies



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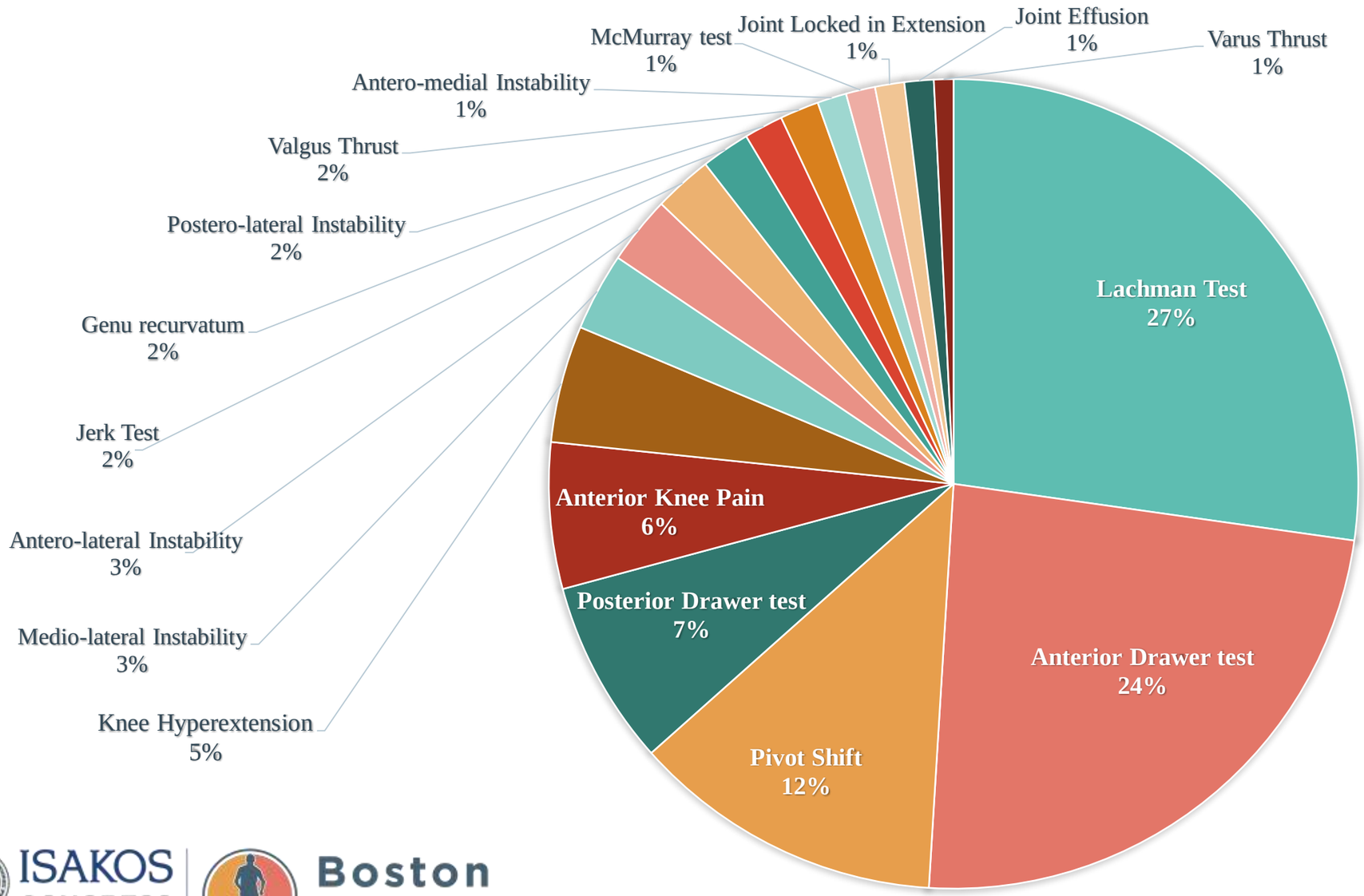
Results: Physical examination



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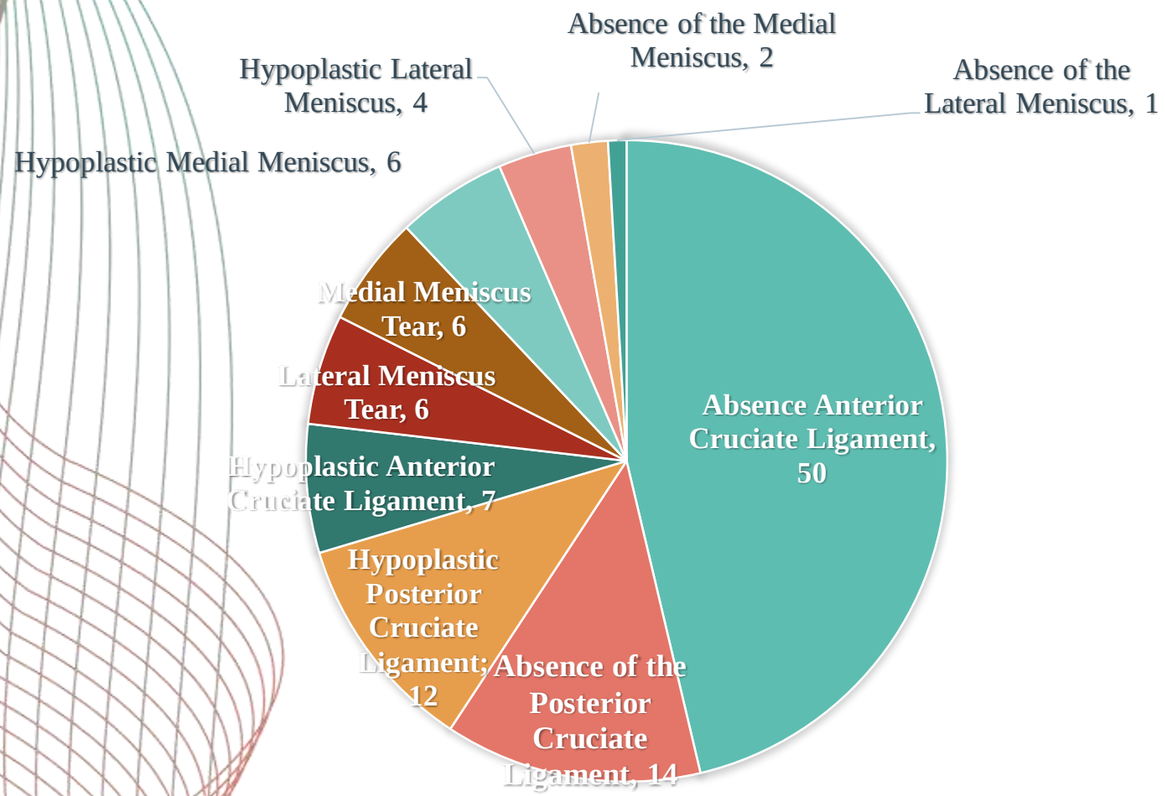
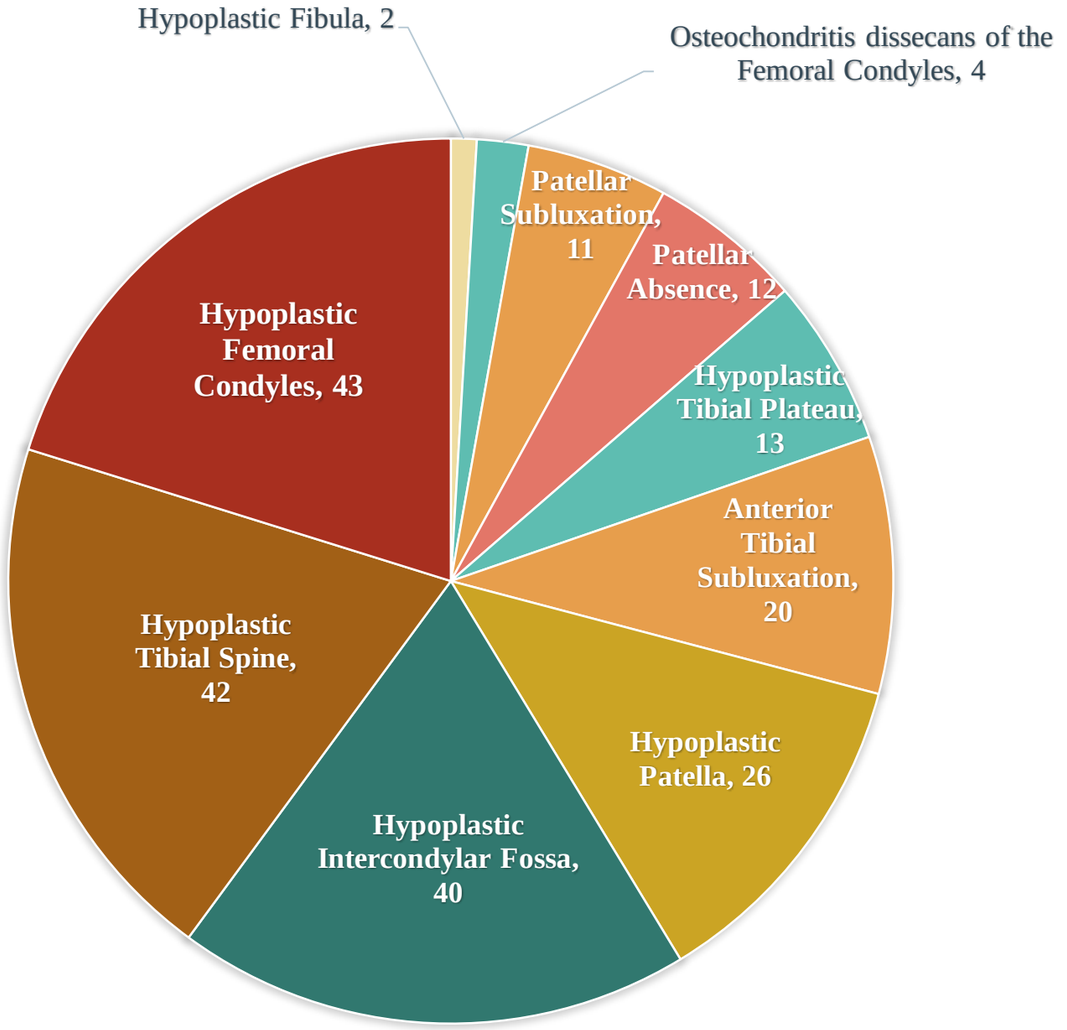
Results: Instrumental examination

MRI Findings



Radiographic Findings



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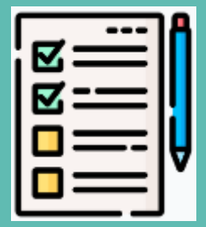
Conclusions



- A preoperative diagnosis of ACL aplasia is undoubtedly challenging
- In the males affected, the diagnosis tends to be done earlier than in the females
- Since the physical examination seems not diriment, a medical history reporting about lower limb anomalies, affecting in particular either the ipsilateral femur, hip or knee, can arise the suspect
- In doubtful cases, based on a physical examination positive for knee instability, standard X-rays seem more objective than MRI
- Treatment of ACL Aplasia remains an open debate among Authors (conservative vs. surgical treatment), therefore an early diagnosis may help the physician in the proper management of these patients, especially those with organ syndromes.

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