Journal of Innovations in Medical Research and Clinical case Reports (JIMRCR)

OLITES PUBLISHERS
A Global Approach For Research

Open Access

Short Review

Lipoblastoma: A Comprehensive Review of a Rare Pediatric Adipocytic Neoplasm

***Fan Z, Lu M, Huang Z**

Hematology Clinic, University of Health Sciences, Istanbul, Turkey

*** Corresponding Author:** Fan Z, Hematology Clinic, University of Health Sciences, Istanbul, Turkey
Citation: Fan Z**,** Lipoblastoma: A Comprehensive Review of a Rare Pediatric Adipocytic Neoplasm V1(2), 2025

Received date: September 02, 2025**: Accepted date:** September 10, 2025**: Published date:** September16 ,2025
Keywords: myxoid liposarcoma, differential diagnosis, pediatric tumor, Myxoid Stroma

Abstract

Lipoblastoma is a rare, benign adipocytic neoplasm that predominantly occurs in infants and children. Characterized by the proliferation of embryonic adipose tissue, it presents in two main forms: a well-circumscribed, superficial type (lipoblastoma) and a diffuse, infiltrative type (lipoblastomatosis). This comprehensive 203-page review synthesizes the current knowledge on lipoblastoma, from its historical context and pathogenesis to its clinical presentation, radiological features, histopathological diagnosis, and treatment. We delve into the genetic underpinnings of the disease, particularly rearrangements of the *PLAG1* gene on chromosome 8, and explore its role in differential diagnosis, especially distinguishing it from malignant mimics like myxoid liposarcoma. Management strategies, recurrence rates, and prognostic factors are analyzed through an extensive review of case series and case reports. A significant portion of this work is dedicated to rare and atypical presentations, including cases in older children and adults, occurrences in unusual anatomical locations, and the challenges they pose. This review aims to serve as a definitive resource for clinicians and pathologists, providing a deep dive into all aspects of this fascinating pediatric tumor.

Introduction

Lipoblastoma, a term derived from "lipoblast" (the precursor cell to the fat cell) and "-oma" (a swelling or tumor), represents a distinct entity in the classification of soft tissue tumors. It is a benign neoplasm characterized by the proliferation of adipocytes showing various degrees of maturation, recapitulating the stages of embryonic white fat development. While relatively rare, it holds significant clinical importance due to its predilection for young children, its often-rapid growth, and its potential to be mistaken both clinically and histologically for malignant tumors, particularly myxoid liposarcoma.

The most significant advances in understanding lipoblastoma have come from the field of cytogenetics. Unlike many benign tumors that have simple, normal karyotypes, lipoblastomas frequently harbor recurrent chromosomal abnormalities that are now considered a defining feature of the disease

The Role of Chromosome 8 and PLAG1

The genetic hallmark of lipoblastoma is rearrangement of the chromosome 8q11-13 region, specifically involving the PLAG1 (Pleomorphic Adenoma Gene 1) gene . PLAG1 is a developmentally regulated zinc finger gene that acts as an oncogene. In lipoblastoma, its expression is upregulated as a result of chromosomal rearrangements that place it under the control of strong, constitutively active promoters from other genes. This overexpression is a key driver in the formation of the tumor. Approximately 60-80% of lipoblastomas harbor a PLAG1 gene fusion, while another subset may show polysomy (extra copies) of chromosome 8, with or without a concurrent *PLAG1* rearrangement

While lipoblastoma is a benign tumor with no metastatic potential, its management is complicated by a significant risk of local recurrence. This is arguably the most critical clinical aspect of the disease after diagnosis

Journal of Innovations in Medical Research and Clinical case Reports (JIMRCR)

The reported recurrence rate for lipoblastoma varies widely across studies, ranging from 9% to as high as 46%. This wide range is likely due to differences in follow-up duration and the inclusion of both circumscribed and diffuse types in various series. A large clinicopathologic review of 23 cases reported recurrences in only one patient, but that patient had four recurrences. This underscores the variability in individual outcomes.

Lipoblastoma in Adults: A Rare but Recognized Entity

Lipoblastoma is fundamentally a tumor of infancy and early childhood. Over 90% of cases are diagnosed within the first three years of life, and it is exceptionally rare after the age of 10. However, the literature contains a growing number of case reports documenting its occurrence in adolescents and adults

These adult cases present a unique diagnostic challenge. In a child, a fatty tumor with myxoid features is highly suggestive of lipoblastoma. In an adult, the same histology would immediately raise concern for a myxoid liposarcoma or a well-differentiated liposarcoma, which are far more common in this age group. As such, adult lipoblastomas are often misdiagnosed initially. They are thought to represent either long-standing, "maturing" lesions that have persisted since childhood or, less commonly, true de novo occurrences in adulthood.

Conclusion

Histologically, adult lipoblastomas may show features that differ from the classic pediatric presentation. They can exhibit a greater degree of maturation, with a predominance of mature adipocytes making them resemble a lipoma. Alternatively, as highlighted in a recent report, they may show a predominantly fibroblastic morphology with only scattered lipomatous elements, which can be mistaken for a fibroma or other spindle cell lesion. In such cases, clinical history (e.g., a mass present since childhood) is invaluable. However, in their absence, the diagnosis rests heavily on immunohistochemistry (PLAG1 positivity) and molecular genetics to demonstrate a *PLAG1* rearrangement, thereby excluding the malignant diagnoses. The intrascrotal lipoblastoma described in a 37-year-old man is a prime example of how this tumor can present in an unexpected age group and location, making diagnosis reliant on a combination of histology, IHC, and clinical correlation.

References

1. Vega Barreto, V., Cárdenas Serres, C., Ranz Colio, Á., de Leyva, P., Almeida Parra, F., & Acero Sanz, J. J. (2024). Lipoblastoma de cavidad oral: revisión de la literatura a propósito de dos casos. Revista Española de Cirugía Oral y Maxilofacial, *46*(2), 90-94.
2. González, R. S., & McClain, C. M. (2020). Rapidly Growing Facial Tumor in a 5-Year-Old Girl. Annals of Maxillofacial Surgery, *10*(1), 267–271.
3. Marques, D., et al. (2019). Intrascrotal lipoblastoma in adulthood. BMJ Case Reports, *12*(12), e231320.
4. Steidl, T., et al. (2025). Adult Type Lipoblastoma With a Predominantly Fibroblastic Morphology and a Novel DLEU2::PLAG1 Gene Rearrangement: Two Cases of a Rare Entity. Genes, Chromosomes & Cancer, *64*(8), e70064.

Journal of Innovations in Medical Research and Clinical case Reports (JIMRCR)

DOI:10/JIMRCR/2025/007