Article published online: 2025-12-22

Two Patients of Acute Leukemia With Underlying Klinefelter Syndrome (47,XXY): A Case Report with a Review of Literature

Pina J. Trivedi[1] Krishna Barad[1] Nidhi Patel[1]

[1] Cytogenetics Lab, Department of Cancer Biology, The Gujarat Cancer & Research Institute, Ahmedabad, Asarwa, Gujarat, India

Ind J Med Paediatr Oncol

Address for correspondence Pina J. Trivedi, PhD, Department of Cancer Biology, The Gujarat Cancer & Research Institute, NCH Campus, Asarwa, Ahmedabad, Gujarat 380016, India (e-mail: pina.trivedi@gcriindia.org; pjt1410@gmail.com).

Abstract

Klinefelter syndrome (KS), a common sex chromosome aneuploidy in men (47,XXY), is primarily known for causing infertility and endocrine issues but is also increasingly linked to a higher risk of *hematologic malignancies*. This abstract presents two cases of young men with KS who developed acute leukemia. Two young men with KS were diagnosed with acute leukemia: one with *acute* myeloid leukemia (AML-M4) and the other with B-cell acute lymphoblastic leukemia (B-ALL). Their cases highlight a notable association between KS and the development of these blood cancers. Cytogenetic analysis showed the constitutional 47,XXY karyotype in both patients, in addition to distinct, acquired chromosomal abnormalities within their leukemic cells. This suggests that the genetic instability inherent to KS may be a predisposing factor for these cancers. The KS-associated 47,XXY karyotype acts as a predisposing factor for leukemia, with the acquired chromosomal aberrations in the leukemic cells demonstrating a link between the congenital aneuploidy and genomic instability.

Keywords

- case report
- AML
- Klinefelter syndrome
- FISH
- conventional cytogenetics

Overview of KS and Its Clinical Features

Klinefelter syndrome (KS) is the most common sex chromosome aneuploidy in males and is typically characterized cytogenetically by the presence of an extra X chromosome, most frequently resulting in a 47,XXY karyotype. Clinically, individuals with KS often present with tall stature, small firm testes, hyper gonadotropic hypogonadism, gynecomastia, infertility due to azoospermia, and reduced secondary sexual characteristics.[1] Neurodevelopmental difficulties such as language delays, dyslexia, and executive dysfunction are also commonly reported, contributing to social and academic challenges.[2] Despite an estimated incidence of 1 in 500 to 1,000 live male births, KS remains significantly underdiagnosed, with only about 25 to 30% of affected individuals ever being clinically identified. The diagnosis is frequently delayed or made incidentally during evaluation for infertility or hematologic disorders.[3]

Beyond the classic features, KS is associated with a range of systemic complications. Studies over the past decade have expanded the clinical spectrum to include increased risk for metabolic syndrome, type 2 diabetes, osteoporosis, autoimmune diseases, and thromboembolic events.[4] In addition, there is a recognized predisposition to malignancies, particularly breast cancer and germ cell tumors (GCTs), attributed to the hormonal and chromosomal alterations in KS.[5] Although hematologic malignancies are relatively rare in these patients, a growing number of case reports have described associations between KS and both myeloid and lymphoid leukemia.[6]

KS and Malignancy Risk

Men with *KS*, a chromosomal disorder characterized by a 47, XXY karyotype, exhibit a unique and complex cancer risk profile that differs significantly from the general male population. This increased susceptibility is not uniform across all

DOI https://doi.org/10.1055/s-0045-1814440.
ISSN 0971-5851.

Thieme Medical and Scientific Publishers Pvt. Ltd., A-12, 2nd Floor, Sector 2, Noida-201301 UP, India

cancer types but is highly specific to certain malignancies, largely due to the hormonal, immunological, and genetic consequences of the extra X chromosome.[7] Regarding solid tumor malignancies; the most striking association is the dramatically elevated risk for male breast cancer. Men with KS have been reported to have a 40- to 50-fold increased risk of developing this rare cancer. The primary driver of this is the hormonal imbalance characteristic of KS, specifically the elevated serum Estrogen and reduced testosterone levels, which promote the proliferation of mammary ductal tissue and create an environment conducive to carcinogenesis.

Another notable risk is for extragonadal GCTs,[8] with men with KS having up to 50-fold increased risk, particularly for those arising in the mediastinum (the space between the lungs). These tumors are thought to originate from misplaced embryonic germ cells, and the extra X chromosome may play a direct role in their development. In contrast, the risk for prostate cancer appears to be decreased in men with KS, likely due to the inherent hypogonadism and chronically low testosterone levels typical of the syndrome.[9] However, this protective effect may be lost if the patient undergoes testosterone replacement therapy. Some studies also suggest a possible increased risk for other tumors, such as lung cancer, though the evidence is less conclusive than for breast cancer and GCTs.[10]

Knowledge Gap and Rationale

While KS is increasingly recognized for its association with various malignancies, the precise mechanisms underlying its link to hematologic cancers, particularly acute leukemia, remain incompletely understood. Existing literature, primarily consisting of isolated case reports, does not provide a comprehensive epidemiological or pathophysiological basis for this association.[11] Specifically, the role of chromosomal instability, X-linked gene dosage effects, and hormonal imbalances in driving leukemogenesis in KS patients requires further investigation. The diversity of leukemia subtypes observed in these cases (myeloid vs. lymphoid) further highlights the need for a more detailed understanding of how the 47,XXY karyotype influences different hematopoietic lineages.[12]

The present case report is rationalized by the scarcity of documented cases describing the co-occurrence of acute leukemia and KS. By detailing two distinct presentations—one with acute myeloid leukemia (AML) and another with B-cell acute lymphoblastic leukemia (B-ALL)—this report provides valuable clinical and cytogenetic data directly relevant to this knowledge gap. These cases, analyzed alongside a review of existing literature, aim to deepen the understanding of the clinical features, cytogenetic complexities, and potential shared pathophysiological mechanisms that may link these two conditions. The report underscores the importance of cytogenetic analysis in young male patients with leukemia, as it may lead to the incidental diagnosis of KS, which has significant implications for both treatment and long-term care.[13]

Case Details

Case 1: AML-M_4 in a 22-Year-Old Male Patient

Clinical presentation: A 22-year-old male patient presented with severe fatigue, generalized weakness, and a suspected infection.

Laboratory findings: Hematological examination showed severe anemia (Hb: 5.1 g/dL), leukocytosis (white blood cell: 35.32 $\times 10^3$/µL), thrombocytopenia (22×10^3/µL), and 70% circulating blasts on peripheral smear. Bone marrow aspiration was hypercellular, with 88% blasts exhibiting high nuclear-to-cytoplasmic ratios and granular cytoplasm. Erythroid and myeloid precursors were severely suppressed, and megakaryocytes were not seen.

Flow cytometry: Flow cytometry was conclusive of AML with AML-M4, with blasts positive for MPO (93%), CD13 (71%), CD33 (81%), CD117 (86%), CD64 (63%), CD15 (36%), HLA-DR (76%), and CD34 (50%), and aberrant Expression of CD7 (74%).

Cytogenetics: Cytogenetic analysis by G-banding using trypsin and Giemsa (GTG) banding showed a uniform 47, XXYc karyotype in 20 metaphases, making a diagnosis of KS. FISH testing for AML1-ETO and CBFB rearrangements was negative, ruling out prevalent AML-associated translocations.

Diagnosis: AML with AML-M4

Timeline: Bone marrow examination was performed on *Day 4 after admission*. Since cytogenetic testing is part of this examination to analyze chromosomes, it was carried out on day 5. This placed the testing at the beginning of the diagnostic process, which simultaneously confirmed the leukemia and led to the discovery of the underlying KS.

Case 2: B-ALL in a 19-Year-Old Male Patient

Clinical presentation: A 19-year-old male patient was hospitalized with symptoms of weakness, fever, and abdominal pain.

Laboratory findings: Bone marrow aspirate showed hypercellularity with 90% blasts, suppression of the myeloid and erythroid series, and absence of megakaryocytes. There was significant pancytopenia with severely low hemoglobin and red blood cell count (4.9 – 5.1 g/dL), and critically low platelets ($22 - 27 \times 103$ Cells/µL). Marked leukocytosis was observed with a very high white blood cell count ($35.32 - 93.18 \times 103$ cells/µL). The manual differential count explicitly noted 70% blast cells, a key diagnostic feature of acute leukemia.

Flow cytometry: Immunophenotypic analysis established the diagnosis of B-ALL, with positive expression of CD19 (84%) and CD79a (45%), and positive expression of CD34 (87%) and HLA-DR (76%), but negative for myeloid markers.

Cytogenetics: Cytogenetic analysis identified a mosaic karyotype: 48, XXYc, +5 in 15 and 46, XY in 10 metaphases, representing KS and trisomy 5 in a leukemic subclone. FISH for gene *BCR-ABL* and *MLL* gene rearrangements was negative, further narrowing the genetic characterization of the disease.

Diagnosis: B-ALL (B-Cell Acute Lymphoblastic Leukemia)

Timeline: The patient was admitted on day 1 and the cytogenetic testing was performed on day 2 after **admission**,

Table 1 Shared pathophysiological thread

Feature	AML-M4* (Case 1)	B-ALL* (Case 2)	Common KS*-related Implications
Karyotype	47,XXYc*[20]	48,XXYc, + 5[15]/46,XY[10]	Genetic instability
Hematopoiesis	Myeloid dysplasia	Lymphoid hyperplasia	Stem cell dysfunction
Hormonal context	Hypogonadism (presumed in KS)	Hypogonadism (KS)	Reduced testosterone may impair normal immune and marrow regulation
Immune dysregulation	Less prominent	More prominent in ALL*	KS patients have altered T- and B-cell immunity

Abbreviations: AML-M4, acute myelomonocytic leukemia; B-ALL, B-cell acute lymphoblastic leukemia; KS, Klinefelter syndrome; 47,XXYc, 47,XXY constitutional.
Note: "*"Immune dysregulation appears more prominent in ALL because its lymphoid malignancy directly disrupts and remodels the already altered immune microenvironment from Klinefelter syndrome and hypogonadism.

concurrent with the bone marrow examination. This crucial diagnostic step allowed for the simultaneous diagnosis of both the acute leukemia subtype and the underlying KS, enabling clinicians to tailor the treatment plan based on a complete understanding of the patient's condition.

The institutional review board approved the present study, and the patient's general consent was obtained.

Shared Pathophysiological Thread

The two cases, AML-M4 and B-ALL, share a common pathophysiological thread rooted in the underlying KS. Both patients' karyotypes exhibit the presence of an extra X chromosome—47,XXYc in Case 1 and 48,XXYc, + 5 in Case 2—indicative of genetic instability. This instability, a hallmark of KS, likely contributes to the chromosomal abnormalities seen in both leukemias. Furthermore, the impaired hematopoiesis observed in both cases, manifesting as myeloid dysplasia in Case 1 and lymphoid hyperplasia in Case 2, points to broader stem cell dysfunction related to KS. Both cases occur within the context of KS-related hypogonadism, where reduced testosterone levels may impair normal immune and marrow regulation, creating a permissive environment for malignancy. While immune dysregulation was more prominent in the B-ALL case, KS patients are known to have altered T- and B-cell immunity, suggesting that this shared feature may be a contributing factor in both presentations (►**Table 1**).

Cytogenetic Analysis

Cytogenetic analysis was performed on bone marrow aspirates and peripheral blood lymphocyte cultures from both patients using the GTG-banding technique. For each bone marrow sample, well-spread metaphases were examined microscopically to determine the karyotype. Subsequently, peripheral blood lymphocyte cultures were performed using standard cytogenetic protocols to differentiate between constitutional and acquired chromosomal abnormalities. The lymphocytes were cultured, harvested, and stained, and a consistent number of metaphases were analyzed to confirm the constitutional nature of the observed chromosomal complements.

Case 1: Cytogenetic analysis of the bone marrow sample from Case 1 revealed a uniform karyotype of 47,XXYc in all 20 metaphases analyzed (►**Fig. 1**). This finding, diagnostic of KS, was further confirmed by a consistent 47,XXYc karyotype in a subsequent peripheral blood lymphocyte culture, verifying the constitutional nature of the chromosomal abnormality.

Case 2: Cytogenetic analysis of the bone marrow sample showed two distinct cell lines. Fifteen of the 25 metaphases analyzed revealed an abnormal karyotype of 48,XXYc, + 5, consistent with KS and an additional trisomy 5 (►**Fig. 2**). The remaining 10 metaphases showed a normal male karyotype of 46,XY. Analysis of a peripheral blood lymphocyte culture confirmed the constitutional 47,XXYc karyotype, and the absence of trisomy 5 in this sample confirmed the acquired (neoplastic) nature of the trisomy 5 clone.

Discussion

KS is the most prevalent sex chromosome aneuploidy in males, cytogenetically defined by the presence of one or more supernumerary X chromosomes, most commonly resulting in a 47,XXY karyotype. This chromosomal disorder occurs in approximately 1 in 600 live male births and presents with high phenotypic variability, including hypergonadotropic hypogonadism, infertility, tall stature, gynecomastia, and cognitive or behavioral challenges.[14] Although KS is relatively common, it is significantly underdiagnosed—clinical identification is achieved in only 25 to –30% of cases, primarily due to its subtle and variable presentation.[15]

Cytogenetically, KS represents a constitutional chromosomal abnormality that may influence hematopoiesis through gene dosage effects and genomic instability. Overexpression of genes located in the pseudoautosomal regions (PAR1 and PAR2) of the extra X chromosome, which escape X-inactivation, may affect the transcription of genes involved in immune regulation and cell cycle control, thereby promoting a tumorigenic environment.[16] In the first presented case, conventional cytogenetic analysis of bone marrow revealed a 47,XXY karyotype in all 20 metaphases analyzed, without evidence of acquired chromosomal changes. This confirmed the presence of a constitutional aneuploidy rather than a leukemic clone. However, even in the absence of acquired abnormalities, constitutional chromosomal

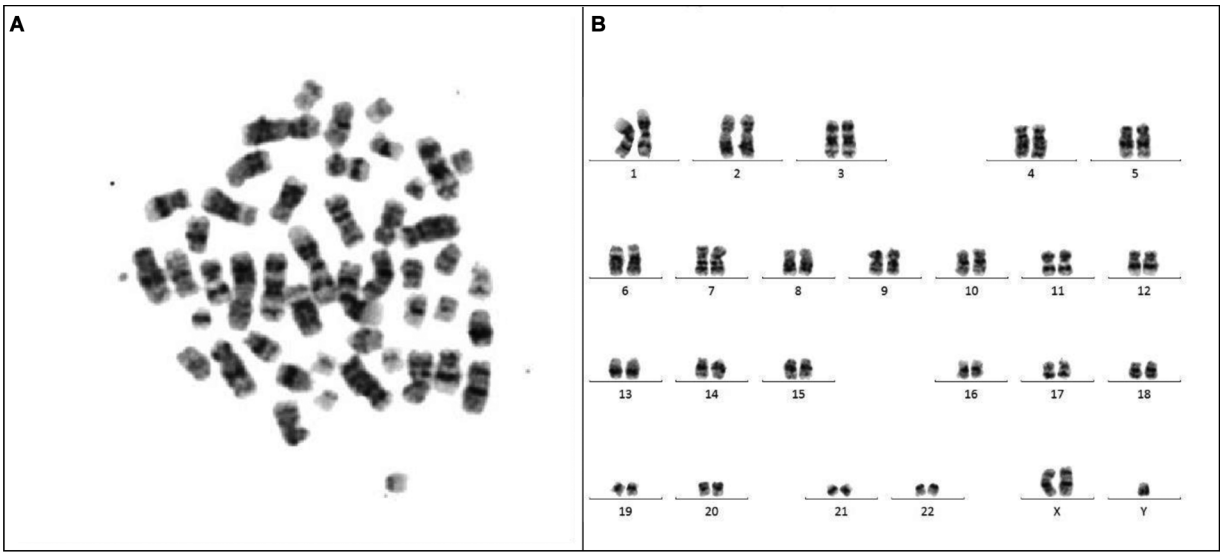

Fig. 1 Representative images of conventional cytogenetic results of GTG banded karyotype showing 47,XXYc [Case 1] (**A**) karyotype image and (**B**) metaphase indicating an Extra X chromosome. GTG, G-banding using trypsin and Giemsa.

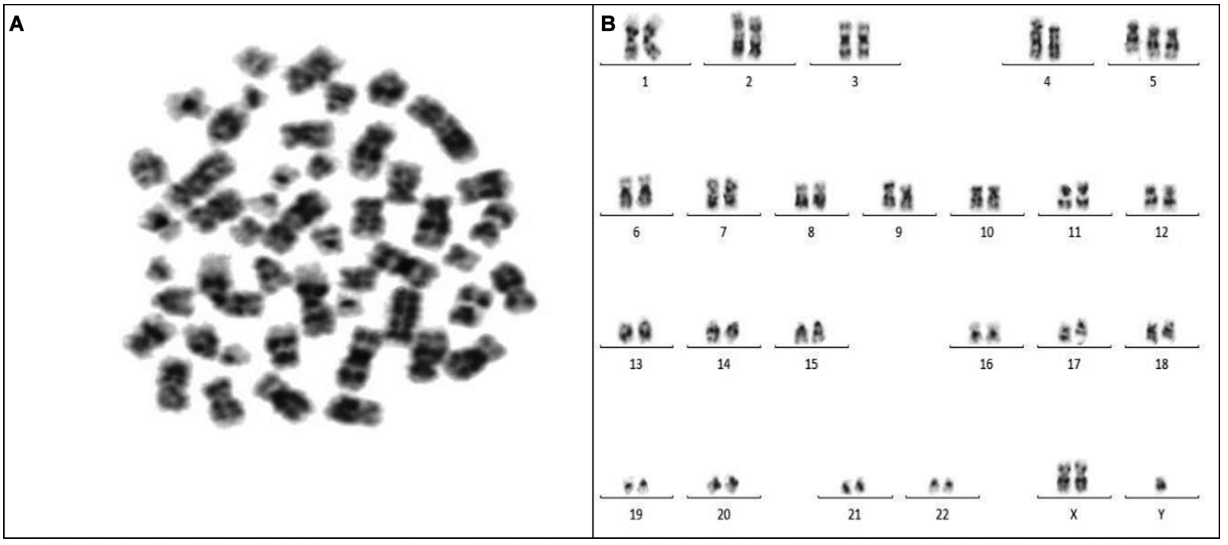

Fig. 2 Representative images of conventional cytogenetic results of GTG banded karyotype showing 48,XXYc, + 5 [Case 2] (**A**) Karyotype image and (**B**) Metaphase which indicates an extra copy of X chromosome with trisomy 5. GTG, G-banding using trypsin and Giemsa.

imbalances such as those seen in KS have been associated with increased susceptibility to hematologic malignancies due to ongoing genomic stress and immune dysfunction. Additionally, research suggests that sex chromosome abnormalities, particularly involving the X chromosome, may contribute to leukemogenic risk in younger individuals with bone marrow failure syndromes or unexplained cytopenias.[17]

The second case was cytogenetically more complex, with mosaicism involving two cell lines: 48,XXY, + 5 in 15 metaphases and a normal male karyotype (46,XY) in 10 metaphases. Trisomy 5 is a recognized acquired cytogenetic aberration often seen in both myeloid and lymphoid neoplasms, and it is associated with poor prognosis in some leukemia subtypes.[11] The co-occurrence of KS and trisomy 5 supports the theory that individuals with constitutional chromosomal aberrations may have an increased propensity for acquiring leukemogenic somatic abnormalities. Mosaicism further underscores the importance of analyzing an adequate number of metaphases to differentiate between inherited and acquired changes.[18]

The risk for hematologic malignancies is also significantly heightened in men with KS. A strong association has been established with non-Hodgkin lymphoma, for which men with KS have a three- to four-fold increased risk. The exact mechanism for this is not fully understood, but it is speculated to be related to the immune system dysfunction and the potential for increased genomic instability resulting from the extra X chromosome.[19] The link to leukemia is less certain; while some studies have reported an increased risk, others

Table 2 Cytogenetic and clinical characteristics of reported leukemia cases in patients with Klinefelter syndrome

Author, Year	Age/ex	Leukemia subtype	KS karyotype	Additional cytogenetics	Treatment	Outcome/note
Slavcheva et al, 2010[8]	41 M	AML-M4 (myelomonocytic)	47,XXY	t(8;21)(q22;q22)	Induction chemotherapy	Achieved remission
Lim et al, 2010[24]	Adult M	AML with RUNX1:RUNX1T1	47,XXY	t(8;21)(q22;q22)	Standard AML induction	Association of KS with recurrent AML lesion
Ljubić et al, 2010[25]	Adult M	Acute basophilic leukemia (AML subtype)	47,XXY	None specific	AML protocol	First KS case with this rare AML subtype
Kjeldsen, 2022[26]	3 M	B-ALL	47,XXY	≥2 acquired aberrations; shortened telomeres	Standard ALL therapy	Highlighted chromosomal instability in KS + ALL
Gürgey et al, 1994 (series)[27]	Children (various M)	B-ALL/ALL	47,XXY (some mosaic)	Variable (extra aberrations in several cases)	Pediatric ALL protocols	Several pediatric ALL cases in KS summarized

Abbreviations: AML, acute myeloid leukemia; KS, Klinefelter syndrome.

suggest that the association may be coincidental, with the KS diagnosis made during the cytogenetic evaluation for leukemia.[20]

The presence of Trisomy 5 in a subset of the leukemic cells in Case 2 is a significant finding. While the constitutional 47, XXY karyotype of KS is a predisposing factor for hematologic malignancies, it is the acquired clonal abnormalities, such as trisomy 5, that directly influence the prognosis.[21] The findings in Case 2 align with patterns reported in the Mitelman Database, where trisomy 5—although uncommon—has been documented as a recurrent acquired abnormality in acute leukemia, particularly in aggressive or complex karyotypes.[22] In the context of B-ALL, trisomy 5 is a common chromosomal aberration; however, its specific prognostic impact can be complex and depends on the presence of other concurrent genetic abnormalities. Generally, the presence of multiple chromosomal aberrations often suggests a more aggressive disease course and may be associated with a less favorable outcome compared to cases with a normal karyotype or a single, simple chromosomal change.[23] A comparison of the literature review of KS with AML and B-ALL is provided in ►**Table 2**.

The key interaction here is that the underlying genetic instability of KS (due to the extra X chromosome) likely creates a permissive environment for the development of these secondary neoplastic chromosomal aberrations, such as Trisomy 5. This suggests that KS itself does not directly worsen the prognosis in the same way that a high-risk mutation would, but it contributes to the development of the aggressive clonal abnormalities that do have a negative prognostic impact.[13] This unique combination of sex chromosome aneuploidy and a recurrent leukemic abnormality may have a synergistic leukemogenic effect, particularly on the lymphoid lineage—as seen in this case of B-ALL. Testosterone deficiency in KS has also been implicated in altering the bone marrow microenvironment and impairing hematopoietic stem cell function and immune surveillance, thus facilitating clonal evolution and malignant transformation. These genetic and hormonal disruptions are believed to play a central role in both the initiation and aggressiveness of hematologic malignancies in KS patients.[12] Altogether, these findings highlight the clinical importance of performing cytogenetic testing in young male patients who present with hematologic malignancies and clinical features suggestive of KS—such as gonadal dysfunction, tall stature, or neurodevelopmental delays.

Conclusion

Both cases highlight a possible association between KS and acute Leukemia development, which is most likely the result of chromosomal instability induced by the extra X chromosome. This evidence supports a multidisciplinary approach and emphasizes the need for additional studies to define the role of KS in leukemia predisposition. Cytogenetic analysis is crucial for both diagnosing leukemia and uncovering underlying constitutional chromosomal disorders like KS. This dual function provides essential information for patient counseling, risk stratification, and long-term care.

Patient's Consent

The institutional review board approved the present study, and the patient's general consent was obtained.

Funding

The authors acknowledge the funding and financial support provided by the Gujarat Cancer Society and Gujarat Cancer & Research Institute (GCRI), which made it possible to carry out this study. Their contribution played a crucial role in facilitating the investigation and completion of this work.

Conflict of Interest

None declared.

Acknowledgments

The authors sincerely thank the Gujarat Cancer Society (GCS) and the Gujarat Cancer & Research Institute (GCRI) for their invaluable support and resources provided during the course of this case study.

References

1 Hiramatsu H, Morishima T, Nakanishi H, et al. Successful treatment of a patient with Klinefelter's syndrome complicated by mediastinal germ cell tumor and AML(M7). Bone Marrow Transplant 2008;41(10):907–908
2 Zitzmann M, Aksglaede L, Corona G, et al. European academy of andrology guidelines on Klinefelter syndrome endorsing organization: European Society of Endocrinology. Andrology 2021;9 (01):145–167
3 Rau RE, Carroll AJ, Heerema NA, et al. Klinefelter syndrome and 47,XYY syndrome in children with B cell acute lymphoblastic leukaemia. Br J Haematol 2017;179(05):843–846
4 Bojesen A, Juul S, Gravholt CH. Prenatal and postnatal prevalence of Klinefelter syndrome: a national registry study. J Clin Endocrinol Metab 2003;88(02):622–626
5 Tartaglia N, Howell S, Davis S, et al. Early neurodevelopmental and medical profile in children with sex chromosome trisomies: background for the prospective eXtraordinarY babies study to identify early risk factors and targets for intervention. Am J Med Genet C Semin Med Genet 2020;184(02):428–443
6 Kanakis GA, Nieschlag E. Klinefelter syndrome: more than hypogonadism. Metabolism 2018;86:135–144
7 Bchir M, Ayed W, Neji HB, et al. Leukemia in patients with Klinefelter syndrome: a report of two cases. Indian J Hematol Blood Transfus 2016;32(Suppl 1):66–68
8 Slavcheva V, Lukanov T, Balatsenko G, et al. Clinical case of acute myeloblastic leukemia with t(8;21)(q22;q22) in a patient with Klinefelter's syndrome. Hematol Rep 2010;2(01):e11
9 Carroll PR, Morse MJ, Koduru PPK, Chaganti RSK. Testicular germ cell tumor in patient with Klinefelter syndrome. Urology 1988;31 (01):72–74
10 Keung YK, Buss D, Chauvenet A, Pettenati M. Hematologic malignancies and Klinefelter syndrome. a chance association? Cancer Genet Cytogenet 2002;139(01):9–13
11 Bonomi M, Rochira V, Pasquali D, Balercia G, Jannini EA, Ferlin AKlinefelter ItaliaN Group (KING) Klinefelter syndrome (KS): genetics, clinical phenotype and hypogonadism. J Endocrinol Invest 2017;40(02):123–134
12 Corona G, Pizzocaro A, Lanfranco F, et al; Klinefelter ItaliaN Group (KING) Sperm recovery and ICSI outcomes in Klinefelter syndrome: a systematic review and meta-analysis. Hum Reprod Update 2017;23(03):265–275
13 Vawter MP, Harvey PD, DeLisi LE. Dysregulation of X-linked gene expression in Klinefelter's syndrome and association with verbal cognition. Am J Med Genet B Neuropsychiatr Genet 2007;144B (06):728–734
14 Cook B, Nayar S, Filson S, Yap T. The incidence of male breast cancer in Klinefelter syndrome and its proposed mechanisms. Breast 2024;78:103827
15 Liu S, Akula N, Reardon PK, et al. Aneuploidy effects on human gene expression across three cell types. Proc Natl Acad Sci U S A 2023;120(21):e2218478120
16 Turriff A, Levy HP, Biesecker B. Prevalence and psychosocial correlates of depressive symptoms among adolescents and adults with Klinefelter syndrome. Genet Med 2011;13(11): 966–972
17 Swerdlow AJ, Schoemaker MJ, Higgins CD, Wright AF, Jacobs PAUK Clinical Cytogenetics Group. Cancer incidence and mortality in men with Klinefelter syndrome: a cohort study. J Natl Cancer Inst 2005;97(16):1204–1210
18 Xu J, Sun J, Zheng SL. Prostate cancer risk-associated genetic markers and their potential clinical utility. Asian J Androl 2013; 15(03):314–322
19 Rafikova G, Gilyazova I, Enikeeva K, Pavlov V, Kzhyshkowska J. Prostate cancer: genetics, epigenetics and the need for immunological biomarkers. Int J Mol Sci 2023;24(16):12797
20 Los E, Ford GA. Klinefelter syndrome. Nih.gov StatPearls Publishing; 2019. Accessed May 05, 2025 at: https://www.ncbi.nlm.nih.gov/books/NBK482314/
21 Vaswani PPM, Dumagay TE. Trisomy 5 as the sole chromosomal anomaly in acute lymphoblastic leukaemia. BMJ Case Rep 2018; 2018:bcr-2018–226006
22 Mitelman F, Johansson B, Mertens F. Mitelman database of chromosome aberrations and gene fusions in cancer. 2023. Accessed December 9, 2025 at: https://mitelmandatabase.isb-cgc.org
23 Smadja N, Varette C, Louvet C, Gramont A, Krulik M. A case of trisomy 5 in a Philadelphia positive leukemia. Cancer Genet Cytogenet 1992;61(02):216–217
24 Lim G, Kim SY, Cho SY, et al. A novel case of acute myeloid leukemia with RUNX1/RUNX1T1 rearrangement in Klinefelter syndrome. Leuk Res 2010;34(09):e259–e260
25 Ljubić N, Lang N, Skelin IK, et al. Klinefelter syndrome and acute basophilic leukaemia—case report. Coll Antropol 2010;34(02): 657–660
26 Kjeldsen E. Congenital aneuploidy in Klinefelter syndrome with B-cell acute lymphoblastic leukemia might be associated with chromosomal instability and reduced telomere length. Cancers (Basel) 2022;14(09):2316
27 Gürgey A, Kara A, Tuncer M, Alikaşifoğlu M, Tunçbilek E. Acute lymphoblastic leukemia associated with Klinefelter syndrome. Pediatr Hematol Oncol 1994;11(02):227–229

