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Leukaemia Section

Review

del(7)(p11-15) solely

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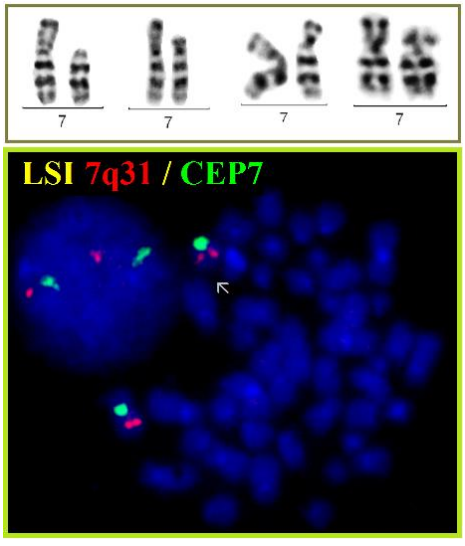
Abstract

Complete or partial loss of chromosome 7 is a recurrent cytogenetic abnormality that may be observed in both de novo and therapy-related hematopoietic disorders. It predominantly presents as monosomy 7 or deletion of the long arm of chromosome 7, often associated with an unfavorable prognosis. Deletions of the short arm of chromosome 7, originated from either a terminal or an interstitial deletion are less frequent and their pathological significance is less well characterized.

KEYWORDS
Monosomy 7; 7p deletions; gene deletions; tumor suppressor genes.

Identity



del(7)(p11-15) Partial karyotypes showing deletions of the short arms of chromosome 7. Hybridization with Vysis D7S486 (7q31)/CEP 7 probe (Abbott moleculars, US) showing deletion of 7p proximal to chromosome 7 centromere.

Clinics and pathology

Disease

Myeloid malignancies and acute lymphoblastic leukemia (ALL).

Etiology

Deletion of 7p as the sole abnormality occurs in disorders with myelodysplastic or myeloproliferative features and acute lymphoblastic leukemia, and may therefore affect early hematopoietic progenitor cells.

Epidemiology

Myeloproliferative disorders mainly: chronic myeloproliferative disorder in 9 (balanced sex ratio; 4 males and 5 females aged 53 to 79 years old, median age 63 years) (Bernstein et al., 1980; CML Yunis et al., 1986; Obeid et al., 1989; Palka et al., 1990; Rodjer et al., 1990; Ohyashiki et al., 1993; Pedersen-Bjergaard et al., 1993; Lezon-Geyda et al., 2001; Maciejewski et al., 2002) and AML in 10 patients (4 males and 6 females, aged 0 to 80 years, median age 0 years) (Bernstein et al., 1984; Testa et al., 1985; Mecucci et al., 1989; Abruzzo et al., 1992; Veldman et al., 1997; Chessells et al., 2002; Babicka et al., 2007; Blink et al., 2012). Male prevalence and prevalence of T-cell phenotype and in ALL (6 males and 2 females aged 3 to 50 years, median age 25 years) (Kaneko et al., 1989; Berger et al., 1990; Bernard et al., 2001; Berger et al., 2003; Douet-Guilbert et al., 2004; Mullighan et al., 2009; Russell et al., 2009; Safavi et al., 2015). 5 patients with myeloid malignancies had secondary disorders treated for a previous malignancy (Mecucci et al., 1989; Rodjer et al., 1990; Abruzzo et al., 1992; Pedersen-Bjergaard et al., 1993). Three patients were Down syndrome patients (Table 1).

(Table 1).

Chronic myeloproliferative disorders				
1	F/64	CMD	46,XX,del(7)(p11)	
2	M/71	RAEB	46,XY,del(7)(p11p22)	
3	F/62	CMMoL	46,XX,del(7)(p11)	
4	F/68	RAEB	46,XX,del(7)(p13)	
5	M/62	RA	45,XY,-10/46,XY,del(7)(p13p14)	**multiple myeloma, chemotherapy**
6	F	RA	46,XX,del(7)(p14)	
7	F/63	MDS	46,XX,del(7)(p11) 46,XX,-7,+21	**multiple myeloma, chemotherapy**
8	M/76	MDS	46,XY,del(7)(p12p15)	
9	M/53	RA	47,XY,del(7)(p13p22)	
Acute myeloid leukemia				
10	M/7	AML-M2	46,XY,del(7)(p11)	
11	M/51	AML-M1	46,XY,del(7)(p1?5)	
12	F/20	AML-M4	46,XX,del(7)(p14	
13	M/47	AML-M2	46,XY,del(7)(p12p21)	**Hodgkin disease, chemotherapy, radiotherapy**
14	F/80	AML-M4	46,XX,del(7)(p12p21)	**polycythemia vera, chemotherapy, radiotherapy**

15	F/11	AML	46,XX,del(7)(p12) **mature B-cell neoplasm, chemotherapy**
16	F/57	AML-M7	47,XX,del(7)(p?),**+21c**
17	F/0	AML-M7	46,XX,del(7)(p15)
18	M/55	AML-M0	46,XY,del(7)(p12)/46,idem,t(2;2)(q31;q35)
19	F/1	AML	47,XX,del(7)(p11),**+21c**
Acute lymphoblastic leukemia			
20	M/14	T-ALL	46,XY,del(7)(p15)
21	M/25	T-ALL	46,XY,del(7)(p13p15)
22	M/25	T-ALL	46,XY,del(7)(p13)
23	M/29	T-ALL	46,XY,del(7)(p11p15)
24	M/50	T-ALL	46,XY,del(7)(p12)
25	F	ALL	47,XX,del(7)(p13p15),**+21c**
26	F/3	B-ALL	47-48,XX,+17,del(20),+21,inc/46,XX,del(7)(p11)
27	M/30	T-ALL	46,XY,del(7)(p13)

Abbreviations: M, male; F, female; CMD, chronic myeloproliferative disorder; RAEB, refractory anemia with excess of blasts; MDS, myelodysplastic syndrome; CMMoL, chronic myelomonocytic leukemia; RA, refractory anemia; AML-M2, acute myeloblastic leukemia with maturation; AML-M1, acute myeloblastic leukemia without maturation; AML-M4, acute myelomonocytic leukemia; AML, acute myeloid leukemia; AML-M7, acute megakaryoblastic leukemia; AML-M5, acute monoblastic leukemia; AML-M0, acute myeloblastic leukemia with minimal differentiation, ALL, acute lymphoblastic leukemia/lymphoblastic lymphoma.
1. Bernstein et al., 1980; 2. Yunis et al., 1986; 3. Obeid et al., 1989; 4. Palka et al., 1990. 5. Rodjer et al., 1990; 6. Ohyashiki et al., 1993; 7. Pedersen-Bjergaard et al., 1993; 8. Lezon et al., 2001; 9. Maciejewski et al., 2002; 10. Bernstein et al., 1984; 11. Testa et al., 1985; 12-14. Mecucci et al., 1989; 15. Abruzzo et al., 1992; 16. Veldman et al., 1997; 17. Chessells et al., 2002; 18. Babicka et al., 2007; 19. Blink et al., 2012; 20. Kaneko et al., 1989; 21. Berger et al., 1990; 22. Bernard et al., 2001; 23. Berger et al., 2003; 24. Douet-Guilbert et al., 2004; 25. Mullighan et al., 2009; 26. Russell et al., 2009; 27. Safavi et al., 2015.

Cytogenetics

Represents as interstitial (9 cases) or terminal deletion (18 patients) with various breakpoints.

Prognosis

Deletion of 7p, appears to confer increased risk of treatment failure and inferior outcome, same as it observed with monosomy 7 in myeloid malignancies. Similarly, event-free survival and survival for patients with monosomy 7 or del(7p), but not of patients with del(7q), were significantly worse in ALL patients than those lacking these abnormalities suggesting the critical region of loss may be on the p-arm (Mecucci et al., 1989; Heerema et al. 2004).

Genes involved and proteins

Germinal Mutations Deletions of the short arm of chromosome 7 that contains the GLI3 (GLI family zinc finger 3) gene at 7p14.1 are responsible for some cases of Greig cephalopolysyndactyly contiguous gene deletion syndrome, a disorder that affects development of the limbs, head, face that may be associated with intellectual disability. Chromosomal deletion or mutation within the TWIST1 (twist family bHLH transcription factor 1) gene at 7p21.1 is observed in Saethre-Chotzen syndrome associated with intellectual disability and developmental delay.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

Chromosome 7p deletion is a recurrent cytogenetic abnormality that may be observed at initial presentation or may emerge during disease progression. Terminal or interstitial 7p deletions result in loss of the deleted material, supporting the existence of tumor-suppressor genes in that region. While no consistent region of loss was identified, these chromosomal deletions may contain areas harboring multiple functionally linked genes whose loss of function is thought to be the major event in disease initiation or progression. Most 7p losses involve large chromosomal regions, therefore it is likely that a cluster of cooperating tumor-suppressor genes collectively promote oncogenesis in dosage-dependent manner.

References

Abruzzo LV, Jaffe ES, Cotelingam JD, Whang-Peng J, Del Duca V Jr, Medeiros LJ. T-cell lymphoblastic lymphoma with eosinophilia associated with subsequent myeloid malignancy. Am J Surg Pathol. 1992 Mar;16(3):236-45

Babicka L, Ransdorfova S, Brezinova J, Zemanova Z, Sindelarova L, Siskova M, Maaloufova J, Cermak J, Michalova K. Analysis of complex chromosomal rearrangements in adult patients with MDS and AML by multicolor FISH. Leuk Res. 2007 Jan;31(1):39-47

Berger R, Dastugue N, Busson M, Van Den Akker J, Pérot C, Ballerini P, Hagemeijer A, Michaux L, Charrin C, Pages MP, Mugneret F, Andrieux J, Talmant P, Hélias C, Mauvieux L, Lafage-Pochitaloff M, Mozziconacci MJ, Cornillet-Lefebvre P, Radford I, Asnafi V, Bilhou-Nabera C, Nguyen Khac F, Léonard C, Speleman F, Poppe B, Bastard C, Taviaux S, Quilichini B, Herens C, Grégoire MJ, Cavé H, Bernard OA. t(5;14)/HOX11L2-positive T-cell acute lymphoblastic leukemia. A collaborative study of the Groupe Français de Cytogénétique Hématologique (GFCH). Leukemia. 2003 Sep;17(9):1851-7

Berger R, Le Coniat M, Vecchione D, Derré J, Chen SJ. Cytogenetic studies of 44 T-cell acute lymphoblastic leukemias. Cancer Genet Cytogenet. 1990 Jan;44(1):69-75

Bernard OA, Busson-LeConiat M, Ballerini P, Mauchauffé M, Della Valle V, Monni R, Nguyen Khac F, Mercher T, Penard-Lacronique V, Pasturaud P, Gressin L, Heilig R, Daniel MT, Lessard M, Berger R. A new recurrent and specific cryptic translocation, t(5;14)(q35;q32), is associated with expression of the Hox11L2 gene in T acute lymphoblastic leukemia. Leukemia. 2001 Oct;15(10):1495-504

Bernstein R, Macdougall LG, Pinto MR. Chromosome patterns in 26 South African children with acute nonlymphocytic leukemia (ANLL). Cancer Genet Cytogenet. 1984 Feb;11(2):199-214

Blink M, van den Heuvel-Eibrink MM, Aalbers AM, Balgobind BV, Hollink IH, Meijerink JP, van der Velden VH, Beverloo BH, de Haas V, Hasle H, Reinhardt D, Klusmann JH, Pieters R, Calado RT, Zwaan CM. High frequency of copy number alterations in myeloid leukaemia of Down syndrome. Br J Haematol. 2012 Sep;158(6):800-3

Chessells JM, Harrison CJ, Kempski H, Webb DK, Wheatley K, Hann IM, Stevens RF, Harrison G, Gibson BE. Clinical features, cytogenetics and outcome in acute lymphoblastic and myeloid leukaemia of infancy: report from the MRC Childhood Leukaemia working party. Leukemia. 2002 May;16(5):776-84

Douet-Guilbert N, Morel F, Le Bris MJ, Herry A, Le Calvez G, Marion V, Abgrall JF, Berthou C, De Braekeleer M. Cytogenetic studies in T-cell acute lymphoblastic leukemia (1981-2002) Leuk Lymphoma 2004 Feb;45(2):287-90

Heerema NA, Nachman JB, Sather HN, La MK, Hutchinson R, Lange BJ, Bostrom B, Steinherz PG, Gaynon PS, Uckun FM; Children's Cancer Group. Deletion of 7p or monosomy 7 in pediatric acute lymphoblastic leukemia is an adverse prognostic factor: a report from the Children's Cancer Group Leukemia 2004 May;18(5):939-47

Kaneko Y, Frizzera G, Shikano T, Kobayashi H, Maseki N, Sakurai M. Chromosomal and immunophenotypic patterns in T cell acute lymphoblastic leukemia (T ALL) and lymphoblastic lymphoma (LBL) Leukemia 1989 Dec;3(12):886-92

Lezon-Geyda K, Najfeld V, Johnson EM. Deletions of PURA, at 5q31, and PURB, at 7p13, in myelodysplastic syndrome and progression to acute myelogenous leukemia Leukemia 2001 Jun;15(6):954-62

Maciejewski JP, Risitano A, Sloand EM, Nunez O, Young NS. Distinct clinical outcomes for cytogenetic abnormalities evolving from aplastic anemia Blood 2002 May 1;99(9):3129-35

Mecucci C, Van Orshoven A, Boogaerts M, Michaux JL, Van den Berghe H. Characterization of deletions of chromosome 7 short arm occurring as primary karyotypic anomaly in acute myeloid leukaemia Br J Haematol 1989 Jan;71(1):13-7

Mullighan CG, Collins-Underwood JR, Phillips LA, Loudin MG, Liu W, Zhang J, Ma J, Coustan-Smith E, Harvey RC, Willman CL, Mikhail FM, Meyer J, Carroll AJ, Williams RT, Cheng J, Heerema NA, Basso G, Pession A, Pui CH, Raimondi SC, Hunger SP, Downing JR, Carroll WL, Rabin KR. Rearrangement of CRLF2 in B-progenitor- and Down syndrome-associated acute lymphoblastic leukemia Nat Genet 2009 Nov;41(11):1243-6

Obeid D, Gould C, Creasy M. Deletion of the short arm of chromosome 7 in chronic monocytic leukaemia undergoing blast cell transformation Br J Haematol 1989 Apr;71(4):561-2

Ohyashiki K, Iwabuchi A, Sasao I, Ohyashiki JH, Ito H, Toyama K. Clinical and cytogenetic significance of myelodysplastic syndromes with disease evolution Cancer Genet Cytogenet 1993 May;67(1):71-8

Palka G, Spadano A, Calabrese G, Parruti G, Guanciali Franchi P, Di Sante O, Recchia A, Di Lorenzo R, Torlontano G. Cytogenetic survey of sixty-one patients with preleukemic syndrome including myeloproliferative and myelodysplastic diseases Ann Genet 1990;33(4):214-8

Pedersen-Bjergaard J, Philip P, Larsen SO, Andersson M, Daugaard G, Ersbøll J, Hansen SW, Hou-Jensen K, Nielsen D, Sigsgaard TC, et al. Therapy-related myelodysplasia and acute myeloid leukemia Cytogenetic characteristics of 115 consecutive cases and risk in seven cohorts of patients treated intensively for malignant diseases in the Copenhagen series Leukemia

Russell LJ, Capasso M, Vater I, Akasaka T, Bernard OA, Calasanz MJ, Chandrasekaran T, Chapiro E, Gesk S, Griffiths M, Guttery DS, Haferlach C, Harder L, Heidenreich O, Irving J, Kearney L, Nguyen-Khac F, Machado L, Minto L, Majid A, Moorman AV, Morrison H, Rand V, Strefford JC, Schwab C, Tönnies H, Dyer MJ, Siebert R, Harrison CJ.

Deregulated expression of cytokine receptor gene, CRLF2, is involved in lymphoid transformation in B-cell precursor acute lymphoblastic leukemia Blood 2009 Sep 24;114(13):2688-98

Safavi S, Hansson M, Karlsson K, Biloglav A, Johansson B, Paulsson K. Novel gene targets detected by genomic profiling in a consecutive series of 126 adults with acute lymphoblastic leukemia Haematologica 2015 Jan;100(1):55-61

Testa JR, Misawa S, Oguma N, Van Sloten K, Wiernik PH. Chromosomal alterations in acute leukemia patients studied with improved culture methods Cancer Res 1985 Jan;45(1):430-4

Veldman T, Vignon C, Schröck E, Rowley JD, Ried T. Hidden chromosome abnormalities in haematological malignancies detected by multicolour spectral karyotyping Nat Genet 1997 Apr;15(4):406-10

Yunis JJ, Rydell RE, Oken MM, Arnesen MA, Mayer MG, Lobell M. Refined chromosome analysis as an independent prognostic indicator in de novo myelodysplastic syndromes Blood 1986 Jun;67(6):1721-30

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