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Clinical and Cytogenetic Characterization of der(1;7)(q10;p10) in Haematological Disorders: Is It Unique to Myelodysplastic Syndrome?

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Introduction

- Der(1;7)(q10;p10) **(**der(1;7)**)** is a derivative chromosome generated by an unbalanced translocation between chromosomes 1 and 7, which results in the gain of the long arm of chromosome 1 (+1q) and the loss of the long arm of chromosome 7 (-7q).
- Recent literature has proposed der(1;7) as a distinct cytogenetic entity within MDS.

Aim

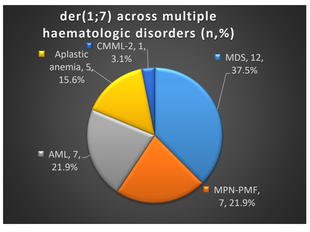
- To describe the clinical and cytogenetic features of der(1;7)(q10;p10) in haematological disorders.

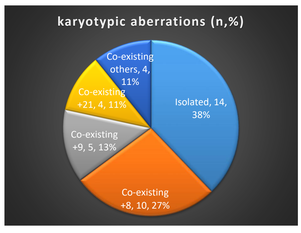
Methods

- All cases with der(1;7) seen at our center, between February 2006 and March 2025, were retrospectively analyzed for their clinical and cytogenetic profiles.
- The MDS cases were classified according to ICC and WHO-Haem5 criteria.
- To further characterize and establish the distinct characteristics of der(1;7), we compared these cases with MDS cases exhibiting isolated monosomy 7.

Results

der(1;7) (February 2006 -March 2025) (n=32)





Baseline characteristics of der(1;7) (n=32)

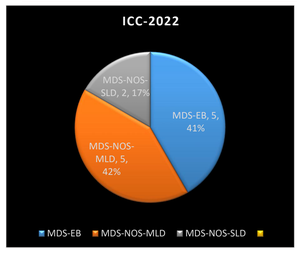
Parameters	Overall (n=32)	MDS (n=12)	AML (n=7)	PMF (n=7)	AA (n=5)	CMML-2 (n=1)
Age in years, Median (range)	53.5 (11-88)	50.5 (28-64)	54 (30-88)	51 (31-64)	32 (11-55)	69
Gender						
Male, **n (%)**	26 (81.3)	9 (75)	5 (71.4)	6 (85.7)	5 (100)	1 (100)
Female, **n (%)**	06 (18.7)	3 (25)	2 (28.6)	1 (14.3)	0 (0)	-
Hb, g/dl, median (range)	8.35 (2.8-14.4)	8.2 (2.8-13.3)	8.6 (5.8-14.4)	8.3 (6.6-11)	9.8 (5.2-10.6)	6.1
WBC x10^9/L, median (range)	3.5 (1.1-82.9)	2.5 (1.3-4.6)	3.5 (1.1-82.9)	3.3 (2.6-9.7)	3.6 (2.0-4.1)	8.0
Platelet x10^9/L, median (range)	42 (7-185)	31 (7-154)	40 (12-94)	46 (20-182)	87 (18-158)	185
ANC, uL (%)	1150 (19-29844)	358 (96-2944)	999 (19-29844)	868 (676- 7130)	738 (220-1886)	2880
PB Blast%, n (%)						
0	14 (43.7)	7 (58.3)	0	1 (14.3)	5 (100)	1
2-4	6 (18.7)	4 (33.3)	0	2 (28.6)	0 (0)	-
5-19	11 (34.4)	1 (8.3)	6 (85.7)	4 (57.1)	0 (0)	-
≥20	01 (3.1)	0 (0)	1 (14.3)			
BMA Blast%, n (%)						
0-4*	24 (68.8)	12 (100)	1 (14.3)#	7 (100)	4	-
5-9	0 (0)	0	0	0 (0)	0	-
10-19	3 (9.4)	0	1 (14.3)	0 (0)	1	1 (100)
≥20	7 (15.6)	0	5 (71.4)	0 (0)	0	-

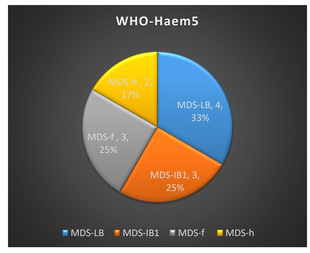
*includes diluted marrow #myeloid sarcoma

References

1. Lang W, Luo Y, Wang L, et al. The der(1;7)(q10;p10) defining a distinct profile from -7/del(7q) in myelodysplastic syndromes: A systematic review and meta-analysis. Cancer Med. 2024;13(1):e6890.
2. Slovak ML, O'Donnell M, Smith DD, Gaal K. Does MDS with der(1;7)(q10;p10) constitute a distinct risk group? A retrospective single institutional analysis of clinical/pathologic features compared to -7/del(7q) MDS. Cancer Genet Cytogenet. 2009;193(2):78-85.

MDS Subtypes as per ICC-2022 and WHO-Haem5





Aplastic Anemia (n=5)

- One case had a germline *GATA2* mutation.
- One case was diagnosed with Fanconi anemia and transformed to acute myeloid leukemia within one year of diagnosis.
- One case progressed to myelofibrosis (possibly related to eltrombopag) within 1 year and 8 months of diagnosis, with acquisition of *ASXL1* and *RUNX1* mutations.

Acute Myeloid Leukemia (n=7)

- Five cases were diagnosed as acute myeloid leukemia with myelodysplasia-related changes
- One case was diagnosed as AML presenting with extramedullary sarcoma of the left humerus and leukemia cutis
- One case developed AML secondary to MDS.

Primary Myelofibrosis (n=7)

- Five cases had *JAK2*V617F mutation, of which two progressed to blast phase transformation
- Two cases had *CALR* mutation (one with *CALR* type 1 and one with *CALR* type 2). The case with *CALR* type 1 mutation progressed to blast phase transformation.

Comparison of der(1;7) with Isolated Monosomy 7

Parameters	Monosomy 7 (n=70)	MDS (n=12)	p-value
Age in years, Median (range)	50.5 (6-76)	50.5 (28-64)	0.94
Gender, n (%)			0.65
Male	48 (68.6)	9 (75%)	
Female	22 (31.4)	3 (25%)	
Hb, g/dl, median (range)	7.5 (2.7-13)	8.2 (2.8-13.3)	0.31
WBC x10^9/L, median (range)	2.6 (0.3-18.7)	2.5 (1.3-4.6)	0.77
Platelet x10^9/L, median (range)	25 (2-443)	31 (7-154)	0.19
ANC, uL (%)	512 (10-15708)	358 (96-2944)	0.98
PB Blast%, n (%)			
0	67 (95.7)	7 (58.3)	
2-4	2 (2.9)	4 (33.3)	
5-19	1 (1.4)	1 (8.3)	
BMA Blast%, n (%)			0 .33
0-4	57 (81.4)	12 (100%)	
5-9	9 (12.9)	0	
10-19	4 (5.7)	0	

Discussion

- Consistent with existing literature, der(1;7) is predominantly seen in males, most frequently in MDS, often as a sole karyotypic abnormality, and commonly co-occurs with +8 and +9.
- While der(1;7) appears to have distinct clinical and cytogenetic features, it is not unique to MDS.
- The limited sample size, absence of long-term follow-up, and lack of molecular profiling preclude definitive classification as a distinct MDS entity.