

# Loss of the Y chromosome in male patients with Myeloid Neoplasms

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## Introduction

Loss of the Y chromosome (LOY) is describe as a both a normal age - related event and a marker of a neoplastic clone in haematological diseases.<sup>1,2,3,4,5,6,7,8</sup>

## Objective

To understand the relationship between LOY chromosome and the different myeloid neoplasms.

## Methods

### Population

We retrospectively analysed cytogenetic results of 891 males' patients, from 1995 to 2016 with myeloid neoplasms.

### Cytogenetics Analyses

Chromosome analysis has done, from cultured cells of bone marrow, without stimulation.

Numerical and structural chromosomal abnormalities were documented for each patient and subdivided according to the number of additional abnormalities.

## Results & Discussion

### Population characterization

120 male patients were removed from this study because they were repeated, remaining 771, which were divided into different pathologies (Figure 1).

### Discussion

Median age calculated for each disease is similar to the ones present in the bibliography.

We have 1,9% of cases with myeloid neoplasms in which the loss of chromosome Y is present in more than 75% of cells (Figure 3).

If we remove patients with LOY and more than 80 years old we don't have any significant difference between the median ages of LOY population and the population with chromosome Y.

This results suggest that loss of chromosome Y is a anomaly characteristic of myeloid neoplasms and in this cases is not associated with age.<sup>3,4,5</sup>

The observation of Table 2 suggest that the loss of chromosome Y in this group of patient is associated with the translocation between chromosome 9 and 22. If exist the loss of chromosome Y, is also present the t(9;22), this finding suggest that the lost of Y is associated with the pathology (Chronic Myeloid Leukemia). But only with these information we can't explain it's mechanism of action.

For patients with myeloid neoplasms it remains uncertain whether the loss of Y chromosome is a casual effect, or is directly associated with the disease process, or is merely transient object in the process.<sup>7</sup>

For more precise conclusions it should be done more studies.

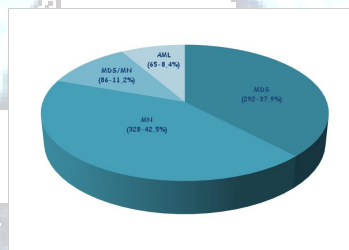


Figure 1: Population characterization. Between brackets are the number of cases for each diseases and respective percentage. MN - Myeloproliferative Neoplasms; MDS - Myelodysplastic Syndromes; AML - Acute Myeloid Leukemia MDS/MN - Myelodysplastic/Myeloproliferative Neoplasms

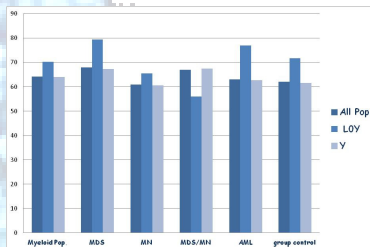


Figure 2: Median Age calculated: ■ - in all population of each group/disease; ■ - in LOY population for each group /diseases; ■ - in the population with chromosome Y for each group /diseases.

Table 1: Frequencies and median age calculated for all population and for each disease analyzed when the Y chromosome is present (Y) /absent (LOY) in more than 75% of the cells observed. The last row is the median age found in the bibliography.<sup>1</sup>

	Myeloid Pop.	MDS	MN	MDS/MN	AML	group control
%						
LOY	1,9	1,7	2,4	1,7	1,5	0,4
Y	98,1	98,3	97,6	98,3	98,4	99,6
All Pop.	64,2	68	61	67	63	62
Median Age						
LOY	70,3	79,4	65,5	56	77	71,7
Y	64,1	67,3	60,6	67,5	62,8	61,5
Bbiblog	-	>70	50-60	>60	>60	-

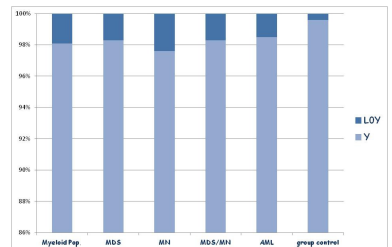


Figure 3: Percentage of males patients without chromosome Y (LOY - ■) and with chromosome Y (Y - ■) found in each disease analyzed.

Table 2: Karyotype from male patient with Chronic Myeloid Leukemia at initial diagnosis and follow-ups.

Patient Number	Sample number	Cariotype
I	A	45,X,-Y,t(5;8)(q11.2;q22),t(9;22)(q34;q11.2)[30]
	B	46,XY,t(5;8)(q11.2;q22)[23]/46,X,-Y,t(5;8),t(9;22)(q34;q11.2)[7]
	C	46,XY,t(5;8)(q11.2;q22)[30]
	D	46,XY,t(5;8)(q11.2;q22)[30]
II	A	45,X,-Y,t(9;22)(q34;q11.2)[16]/46,XY[14]
	B	45,X,-Y,t(9;22)(q34;q11.2)[13]/46,XY[17]
	C	46,X,-Y,t(9;22)(q34;q11.2)[3]/46,XY[27]
	D	45,X,-Y,t(9;22)(q34;q11.2)[1]/46,XY[29]
III	A	45,X,-Y,t(9;22)(q34;q11.2)[28]/46,XY[2]
	B	46,XY[30]
	C	46,X,-Y,t(9;22)(q34;q11.2)[1]/46,XY[29]
	D	46,XY,t(9;22)(q34;q11.2)[1]/46,XY[29]
	E	46,XY[30]

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