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ORIGINAL ARTICLE

Chromosomal abnormalities in patients with haematologic malignancies in the General Hospital of Mexico

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KEYWORDS
Cytogenetics;
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Abstract
Background: Haematologic malignancies are generated by alterations in haematopoietic stem cells. Chromosomal rearrangements are present in >50% of patients and are useful as diagnostic and prognostic factors.
Objective: In this study we describe the cytogenetic characteristics observed in patients with haematological malignancies in the Genetics Department during the period 2000–2014.
Material and Methods: The karyotype was performed on bone marrow (85%) and peripheral blood (15%) with conventional techniques in 9717 samples.
Results: The average age was 40 years (range 0.3–95) and the male/female distribution was 50.5%/49.5%. 352 cases (3.6%) were paediatric with a male/female distribution of 59/41%. The diagnosis was: acute leukaemia 4445 (45.7%), CML 2058 (20.4%), and MDS or some form of cytopenia 1573 (16%). Fewer than 5% of samples received were from AA, MM, CMPD, NHL, CLL, LPD and others. The distribution of acute leukaemia was: ALL 44%, AML 43% and unspecified 13%; the predominant subtypes were ALL-L2 at 50.7% and AML-M3 at 54.2%. Only 61% of the 9717 samples were processed. The karyotype was normal in 3956 (66.7%) samples, the rest (1972, 33.3%) had chromosomal abnormalities: 65% structural and 35% numerical. The changes observed most frequently were t(9;22)(q34;q11) 26%, hyperdiploidy/polyploidy 19.3%, diverse translocations 8.4%, hypodiploidy 8%, t(15;17)(q22;q12) 7.8%, and MDS-related disorders (del5q/-5/-7/+8) 7.7%. Different deletions, trisomy, monosomy and/or complex karyotype were present in smaller proportion (<7%).

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Conclusions: The karyotype remains useful to confirm the diagnoses, establish risk-based prognoses, and classify based on risk to patients; for example in cases with t(9;22) in CML or t(15;17) in M3.

PALABRAS CLAVE
Citogenética;
Cromosomas;
Leucemias

Alteraciones cromosómicas en pacientes con neoplasias hematológicas en el Hospital General de México

Resumen
Antecedentes: Las neoplasias hematológicas son generadas por alteraciones en células progenitoras hematopoyéticas. Presentan rearreglos cromosómicos en >50% de los pacientes de utilidad como factores de diagnóstico y pronóstico.
Objetivo: Describir las características citogenéticas observadas en los pacientes con enfermedades hematológicas recibidos en el Servicio de Genética durante el período 2000–2014.
Material y Métodos: El cariotipo se realizó en médula ósea (85%) y sangre periférica (15%) con técnicas convencionales en 9,717 muestras.
Resultados: La edad promedio fue 40 años (rango 0.3 a 95) y la distribución masculino/femenino 50.5/49.5%. Se estudiaron 352 casos (3.6%) pediátricos con una distribución masculino/femenino 59/41%. Por tipo de patología la distribución fue leucemias agudas, 4445 (45.7%), LMC 2058 (20.4%) y SMD o alguna citopenia 1573 (16%). Se recibieron <5% de muestras de AA, MM, SMPC, LNH, LLC, SLPC y otros. La distribución de las leucemias agudas fue: 44% LAL, 43% LAM y 13% sin especificar; los subtipos predominantes fueron LAL-L2 50.7% y LAM-M3 54.2%. Sólo 61% de las 9,717 muestras procesadas fueron factibles para el estudio. El cariotipo fue normal en 3,956 (66.7%) muestras, el resto (1,972; 33.3%) presentó alteraciones cromosómicas: 65% estructurales y 35% numéricas. Las alteraciones observadas con mayor frecuencia fueron la t(9;22)(q34;q11) 26%, hiperdploidía/poliploidía 19.3%, translocaciones variadas 8.4%, hipodiploidía 8%; t(15;17)(q22;q12) 7.8%; alteraciones relacionadas con SMD (del5q/-5/-7/+8) 7.7%. En menor proporción (<7%) se observaron diferentes deleciones, trisomías, monosomías y/o cariotipo complejo.
Conclusiones: El cariotipo sigue siendo de utilidad para confirmar diagnósticos como en los casos con t(9;22) en LMC o t(15;17) en M3 siendo muy útil como auxiliar para establecer pronósticos y clasificar con base en el riesgo a los pacientes.

Introduction

Haematologic malignancies are generated by alterations in haematopoietic stem cells and include leukaemias, lymphomas, myelomas, aplastic anaemia (AA), myelodysplastic syndromes (MDS) and chronic myeloproliferative disorders (CMPD). In general, lymphomas are classified by the type of cell of origin, while myeloid (AML/CML) or lymphoid (AML/CLL) leukaemias are either acute or chronic. Myelodysplastic syndromes (MDS) are a group of disorders characterised by one or more peripheral blood cytopenias, secondary to bone marrow dysfunction. Generally, CMPDs present a greater amount of mature myeloid cells in peripheral blood and include syndromes that share characteristics of MDS/CMPD and atypical chronic myeloid leukaemia (CML). The latter are diseases of adults with a frequent peak in the fifth and sixth decade of life; their global incidence is about 6–9 per 100,000 inhabitants. Globally, leukaemias and lymphomas are the most frequent haematologic malignancies, representing 2.8% of new cases of cancer. In Mexico, the Population-based Cancer Registry places haematologic malignancies in the first five places.[1,2]

Cytogenetic study in malignant and benign haemopathies is important for the characterisation of the disease, as it contributes to diagnosis and is a well-defined prognostic factor. In more than 50% of haematological malignancies, clonal chromosomal alterations have been characterised based on number of chromosomes (hyperdiploidy, trisomies or monosomies) or the structure of the chromosomes (translocations, inversions, deletions). The first chromosomal rearrangement described in cancer, and currently the best characterised in patients with CML, is t(9;22)(q34;q11), also known as the Philadelphia chromosome (Ph+). The genes involved in the Ph+ rearrangement are *ABL* and *BCR*, which, when fused, cause a *BCR/ABL* oncoprotein with tyrosine kinase activity. This protein is the target of the specific