

Leukaemia Section

Mini Review

+5 or trisomy 5

Edmond SK Ma, Thomas SK Wan

Hematology Division, Department of Pathology, The University of Hong Kong, Queen Mary Hospital, Hong Kong, P.R. China (ESKM, TSKW)

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Clinics and pathology

Disease

Trisomy 5 is found in both acute lymphoblastic leukaemia (ALL) and acute myeloid leukaemia (AML), but very rarely as the sole karyotypic abnormality.

Clinics

In childhood ALL, an extra chromosome 5 is commonly encountered in cases with hyperdiploidy >50 chromosomes. The presence of trisomy 5 in high hyperdiploid childhood ALL is associated with a less favourable clinical outcome. Trisomy 5 as a sole abnormality in ALL is exceedingly rare and described in only 3 cases, including 2 adult ALL and 1 paediatrics case occurring in a 12-year old girl.

Trisomy 5 has been described in 19 cases of AML. Gain of chromosome 5 usually occurs in association with other cytogenetics aberrations, although very rarely it may exist as the sole abnormality. A case of AML-M2 with normal karyotype at diagnosis showed trisomy 5 as the sole abnormality at relapse, and a case of AML-M5 showed trisomy 5 as the only chromosome aberration in 3% of 59 metaphases at presentation. A further case of AML-M4 showed a clone with trisomy 5 as the sole abnormality together with a second clone with trisomy 5 and evolutionary change. In the other 16 cases, trisomy 5 was found in association with numerical (n=4), structural changes (n=4), or both numerical and structural changes (n=8).

A number of interesting observations with respect to AML and trisomy 5 should be noted. First, an association between trisomy 5 and t(8;21) (n=3) and trisomy 8 (n=6) is observed. Second, five out of 6 cases with concurrent trisomies 5 and 8 show monocytic differentiation and are diagnosed as either AML-M4 or M5. Finally, trisomy 5 has been described in all FAB

subtypes of AML except acute promyelocytic leukaemia. Given the rarity of trisomy 5 in AML, it is possible that the associated cytogenetic aberrations such as t(8;21) or trisomy 8 and not trisomy 5 per se that predicts for the myeloid phenotype.

Prognosis

Trisomy 5 in childhood ALL with hyperdiploidy >50 chromosomes is associated with a poorer clinical outcome. Among the 3 cases of ALL with trisomy 5 as the sole karyotypic abnormality, 2 (one case each of adult and paediatric ALL) showed short survival whilst one adult ALL case showed event free survival of 4 years off chemotherapy.

Among 19 AML patients with trisomy 5, complete remission was achieved in 8 and the median overall survival was 15 months. For the remaining 11 patients, 2 achieved partial remission and died, 4 did not attain remission at all, 1 was not treated, and the status of 4 patients was unknown.

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