

## Trisomy 2

### Identity



GTG-banded partial karyotype showing 47,XY,+2

### Clinics and Pathology

#### Note

Trisomy 2 (+2) is a rare yet recurrent finding in [myelodysplastic syndrome](#) (MDS) but occurs more frequently in [acute myeloid leukaemia](#) (AML) in combination with other chromosomal abnormalities. It is a recognised chromosomal change in other neoplasms, in particular [hepatoblastomas](#) and has been described in [fibrous dysplasia](#), pleuropulmonary blastoma, proliferative myositis, nasopharyngeal carcinoma and proliferative fasciitis. As the sole abnormality, it has been associated with post-transplant lymphoproliferative disorders (PTLD).

Isolated trisomy 2 has been reported in 4 cases of MDS and in two patients with MDS transforming to AML. These cases account for the following stages of MDS [refractory anaemia](#) (RA), [RA with excess blasts](#) (RAEB), [RA with excess blasts in transformation](#) (RAEB-t) and [chronic myelomonocytic leukaemia](#) (CMML).

It has been suggested that the presence of trisomy 2 in MDS is an early genetic event that, in combination with other chromosomal changes, may give rise to AML. All of the reported cases appear to be mosaic in nature, and thus its true incidence may be higher. Further case reports are needed to ascertain the effect of trisomy 2 at clinical presentation in both MDS and AML, its association with progression of MDS to AML and prognostic significance.

It has also been suggested that the presence of trisomy 2 may be age-related. Trisomy 2 has been observed in in vitro senescent lymphocytes in elderly patients ranging in age from 70-100 years. All the published cases of trisomy 2 as a sole abnormality in MDS fall within this age range and thus the possibility that the presence of trisomy 2 may be an age-related phenomenon cannot be excluded.

**Prognosis** Trisomy 2 may define a distinct subtype of MDS, which in combination with further clonal chromosomal changes gives rise to AML. Further cases need to be collated to substantiate this.

### Cytogenetics

Note The patient is a 73-year-old Asian male with rheumatoid arthritis, ischaemic heart disease, benign prostate hyperplasia and b-thalassaemia trait who presented with severe anaemia. Patient's blood film showed erythrocyte anisocytosis and poikilocytosis, platelet anisocytosis and dysplastic neutrophils. Blood counts showed: haemoglobin 9.4 g/dL, white cell count  $6.8 \times 10^9/L$  and platelet count  $98 \times 10^9/L$ . Bone marrow aspirate was hypercellular with trilineage dysplasia and 18% myeloblasts, consistent with MDS, WHO category <sup>3</sup>refractory anaemia with excess blasts (RAEB-II)<sup>2</sup>. Chromosome analysis on bone marrow cells showed: 47,XY,+2[5]/46,XY[13].

### To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:  
**you are welcome to submit a paper to our new [Case Report section](#).**

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