

## -21 or monosomy 21 (solely)

### Identity

**Note** True monosomy 21 mosaicism can appear as the sole cytogenetic abnormality but it is rare. Many of the reported cases in the literature probably represent technical artifacts due to random loss of chromosome 21 in multiple cells by chance alone. Suspected monosomy 21 mosaicism should be confirmed by analysis of additional metaphase cells and by FISH analysis of interphase cells. Of the reported cases that are most likely to represent true clonal disease, Acute myeloid leukemia (AML) and [chronic myelogenous leukemia \(CML\)](#) were the most common diagnoses, with one case each of myelodysplasia (MDS), MDS/MPD (myelodysplastic syndrome), acute lymphocytic leukemia (ALL), and [multiple myeloma](#) .

### Clinics and Pathology

**Disease** See [Acute myeloid leukemia](#)  
Nine cases of AML have been reported, with 14-81% of metaphase cells exhibiting monosomy 21 as a sole cytogenetic abnormality. These include four with AML-M2, two with M4, and one with M5.

**Epidemiology** Most of the AML patients were male and most over age 40 at diagnosis.

**Prognosis** Some of the patients achieved complete remission, with survival ranging from 30+ months to 54+ months. Other patients succumbed to their disease in 2-13 months.

**Disease** See [Chronic lymphocytic leukemia \(CLL\)](#)  
Four cases of CLL have been reported, with 15-41% of metaphase cells exhibiting monosomy 21 as a sole cytogenetic abnormality. Most cases were staged A/O in the Binet/Rai classification.

**Epidemiology** All five cases were males aged 45-77 years

**Cytogenetics** One CLL patient exhibited a 13q deletion by FISH that was not observed in the metaphase analysis.

**Prognosis** Survival was 77 months and 90+ months in the two cases where information was provided.

### External links

Other database [-21 or monosomy 21 \(solely\)](#) [Mitelman database \(CGAP - NCBI\)](#)

### 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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**Contributor(s)**

**Written** 08-2002 Daniel L. Van Dyke

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