An unusual hyperdiploid karyotype in a case of paediatric AML

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Patient details

- 4 year old boy
- presented October 2010
- ?acute leukemia
- hyperdiploid (53 chromosomes)
- +4, +8, +10, +15, +20, +21, +22
Final Karyotype

- 46,XY,inv(16)(p13q22)[4]/
- 53,idem,+4,+8,+10,+15,+20,+21,+22[5]/
- 46,XY[2]
How to report?

- inv(16) usually has favourable prognosis. Additional changes (especially +22) in younger adults (16-59 years) better
- ?child
- ?multiple chromosomal gains (acquired in a single step)
- Hyperdiploidy in AML mainly seen in adults and not favourable
Similar cases

- Corral et al (March 2008)
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- “Complex Chromosome t(8;21) with associated hyperdiploidy in Acute Myeloid Leukemia (FAB M2)”
Patient

- 11 year old male
- 46,XY,t(8;21)[3]/
  48,XY,idem,+4,+6[3]/
  52,XY,idem,+4,+6,+8,+17,+22,+mar[13]/
  46,XY[1]
- Cytogenetics, FISH, SKY, CGH
- Still in remission, +11months from diagnosis
Glasgow case
Follow up

- Sample received 2\textsuperscript{nd} November
- ?remission
- FISH for CBFB
- Normal signal pattern
- Consistent with cytogenetic remission
- Further samples 15th November 2010/Dec2010/Feb 2011
- Continued cytogenetic remission
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