

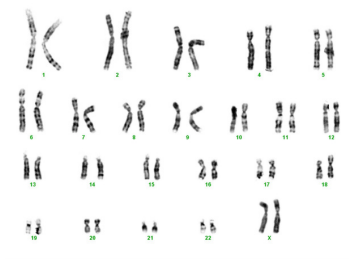
# An Unusual Finding in the Mother of a Prenatally Diagnosed Spina Bifida – A case report

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## Introduction

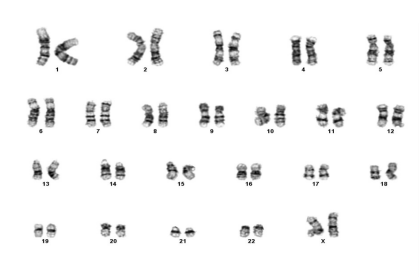
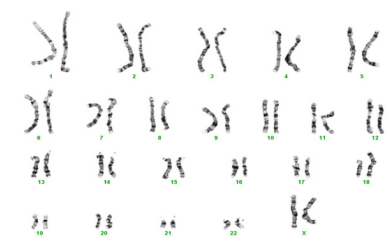
We report on a 30 year old female gravida 2, para 1 who was referred during her second pregnancy following an abnormal ultrasound scan. The pregnancy, affected with spina bifida, was terminated at 22 wks gestation and the cord stump and a maternal blood sample were received for cytogenetic follow-up

**Fetal karyotype**  
46,XX,der(18)



**Maternal karyotype** (confirmed on second sample)

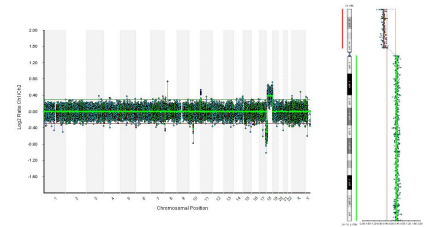
46,XX,i(18)(q11)[27]/46,XX,der(18)[3].ish i(18)(subtel(18)(p-q++),18cen(D18Z1x1))[54]  
/der(18)(subtel(18)(p-),18cen(D18Z1x2))[6]



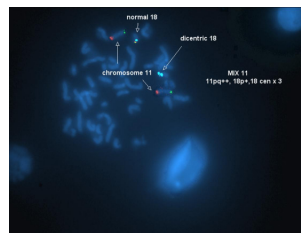
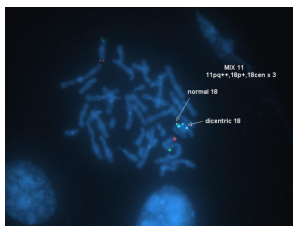
**Tissue abnormal 18 composite**



**Blood abnormal 18 composite**



**Targeted FISH on tissue**



## Literature Review

- i(18q) syndrome:** Holoprosencephaly; mild trisomy 18 features; IUGR; severe MR
- 18p- syndrome:** Brachy-microcephaly; holoprosencephaly; cyclopia; facial and skeletal anomalies; moderate/severe MR; autoimmune disorders; irregular teeth; short broad neck; growth hormone deficiency; (uncommonly) borderline normal intelligence
- Dup 18p syndrome:** (under diagnosed due to lack of severity of phenotype) mild, unspecific dysmorphism; intelligence average to mildly subnormal
- Dup 18p/ i(18q):** (our patient) No family history; normal vaginal delivery; soft dysmorphic features (overcrowding of teeth, mild neck webbing, some hair on chest); development parameters at the low end of normal: attended main stream school and subsequently worked; height 110cm

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