mesenchymal core cells. After specific enzyme digestion of the placental villi (treatment with trypsin then collagenase), the cells from the mesenchymal core were isolated and collected on glass slides using a CytospinTM technique. The Q-FISH technique was applied to these preparations, and normal telomere length distribution was observed. CONCLUSIONS: Our results showed that cell culture of placental villi produced a bimodal telomere length distribution. It is probable that this is related to cell culture conditions (oxidative stress) and could indicate the rapid onset of senescence in some of the cultured placental cells. These results encourage us to estimate telomere length in the future from uncultured placental cells, in order to avoid the variability inherent in cell culture.

## P1-8

### A 32 year retrospective prenatal study of 47,XXX and 47,XYY dysgonosomies

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OBJECTIVES: 47,XXX and 47,XYY syndromes are two of the most common chromosomal abnormalities with a 1/1000 frequency. Their prenatal diagnosis (PD) is often fortuitous. We aimed to evaluate the different prenatal diagnosis parameters which enabled these PD and pregnancies' outcomes to improve genetic counseling. METHOD: A retrospective collaborative study using data from 21 French laboratories over a period of 31 years from 1981 to 2011 was conducted by collecting the following data for each case: indication of the collection, nature of sample, maternal age, fetal karyotype and pregnancy outcome. All parameters were analyzed according to the implementation of multidisciplinary centers for prenatal diagnosis (MCPD) created since 1997. RESULTS: 291 cases of 47,XXX and 167 cases of 47,XYY syndromes were collected. Most were done after amniotic fluid retrieval (90% and 88%). The main indication were advanced maternal age (AMA) (53.3%) and ultrasound findings (UF) (22.4%) for 47, XXX. For 47,XYY, the main indication were similar UF (35.9%) and AMA (31.7%). Mean maternal age was different ($p < 0.0001$) at 37.3 and 34.5 years old respectively for 47,XXX and 47,XYY. Karyotype was homogenous in 87.6% of cases. Outcome was similar regardless of the karyotype: 78.9% of delivery, 17.9% of termination of pregnancy (TOP), 3.2% pregnancy loss. There is a statistical difference ($p < 0.0001$) for outcomes of pregnancies without UF (isolated advanced maternal age, maternal serum markers, personal convenience) in both groups before and after 1997, with a decrease of TOP from 28.6% to 5.4%. CONCLUSIONS: As supposed, due to both maternal and paternal inheritance, 47,XXX dysgonosomy is twice more frequent than 47,XYY one. Difference in maternal age was mainly due to PD indication, with an increase incidence of maternal age for 47,XXX. Furthermore, we clearly showed that establishment of MCDP permitted to progressively change outcomes of these pregnancies allowing to improve the genetic counseling provided to couples.

## P1-9

### Prenatal diagnosis of mosaic tetrasomy 18p

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OBJECTIVES: To report the case of a prenatally detected de novo mosaic tetrasomy 18p where a combination of different methodologies was used,



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including conventional cytogenetics and molecular genetics techniques. METHOD: Molecular rapid aneuploidy diagnosis, by quantitative fluorescent polymerase chain reaction (QF-PCR), conventional cytogenetics and fluorescence in situ hybridization (FISH). RESULTS: Amniocentesis was performed at 22 + 5 gestational weeks, on a 37-year-old woman, due to ultrasound detection of fetal interventricular communication, overlapped fingers and unilateral club foot. Molecular rapid aneuploidy testing, by QF-PCR, showed highly increased ratios for markers on the short arm of chromosome 18. Cytogenetic analysis revealed a male karyotype with mosaicism involving two cell lines: one with a supernumerary isochromosome 18p and another with two extra derivative chromosomes 18. Parental karyotypes were normal and QF-PCR analysis indicated that the extra chromosome 18p material was of maternal origin. After counseling the couple opted for pregnancy termination. Anatomopathological studies, as well as further characterization of the derivative chromosomes, are underway in order to provide more accurate genotype-correlation. CONCLUSIONS: The majority of tetrasomy 18p reported cases were ascertained postnatally, had a de novo occurrence and an isochromosome 18p in all cells examined. Prenatal diagnoses of mosaic tetrasomy 18p are, on the other hand, rarely described in the literature. The case herein reported, where a mosaic is present and none of the cell lines is chromosomally normal, may provide further insight on this rare syndrome and help in the knowledge of the associated phenotype. The application of a combined strategy, using QF-PCR, conventional cytogenetics and FISH analysis allowed not only for the identification of the extra chromosome 18p material, in the form of a mosaicism involving isochromosome 18p, but also for determining its parental origin. This information is of particular importance in recurrence risk assessment and therefore crucial for genetic counseling.

## P1-10

### Prenatal diagnosis of a del(22)(q11.2q11.2) inherited from a mother with a mosaic del(22)(q11.2q11.2)/dup (22)(q11.2q11.2) karyotype

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OBJECTIVES: Screening for 22q11.2 deletion in a fetus with a right sided aortic arch, growth restriction and retrognathia. The mother was affected by a cardiopathy and cleft lip and palate. METHOD: Primigravida, 31 years old woman with cleft lip and palate, valvular atresia and hypoplastic pulmonary artery. An amniocentesis was performed at 30 weeks and 6 days of gestation, due to fetal right sided aortic arch, growth restriction and retrognathia (suspicion of DiGeorge Syndrome). Cytogenetic analysis was performed on cultured amniocytes from amniotic fluid and on the mother′s peripheral blood lymphocytes using GTL-banding technique. MLPA technique (SALSA P250 Kit) and FISH analysis using the TUPLE1 probe localized to the DiGeorge/velocardiofacial syndrome critical region were done. RESULTS: Karyotype performed on amniocytes revealed a 22q11.2 deletion. Maternal karyotype revealed the same deletion. In both cases the deletion was confirmed by MLPA. Further FISH analysis performed on mother peripheral blood sample revealed an additional 22q11.2 duplication in the homologous chromosome as a mosaic line: ish del (22)(q11.2q11.2)(TUPLE1-)[100/100]/dup(22)(q11.2q11.2)(TUPLE1 + +)[13/100]. CONCLUSIONS: The 22q11.2 deletion syndrome is the most common microdeletion syndrome and most often occurs as a de novo event. Only about 10% of the 22q11.2 deletions are inherited. However, cases of rare deletions and duplications with different sizes and locations have also been reported. To the best of our knowledge, this is the first case with a 22q11.2 deletion in all cells, plus duplication (same region) on the other homologous in about 13% of the cells. It is difficult to propose a mechanism to this deletion/duplication mosaicism, since a single meiotic event cannot origin two abnormal cell lines. In the absence of a detectable normal cell line, an error during the first mitotic division or in a very early post-zygotic division seems the most plausible event. The mechanism of mosaic cell line formation in our patient appears to be consistent with a mitotic event involving inter-chromosomal unequal recombination enhanced by low copy number repeats. This case shows the importance of an accurate analysis of phenotypes and highlights the relevancy of choosing the most suitable techniques for each case, once duplications 22q11.2 may exhibit phenotypes more benign and therefore easily overseen.

## P1-11

### Fetal arrhythmias-a case report

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OBJECTIVES: Fetal arrhythmias complicate 1%–2% of all pregnancies. The most common are