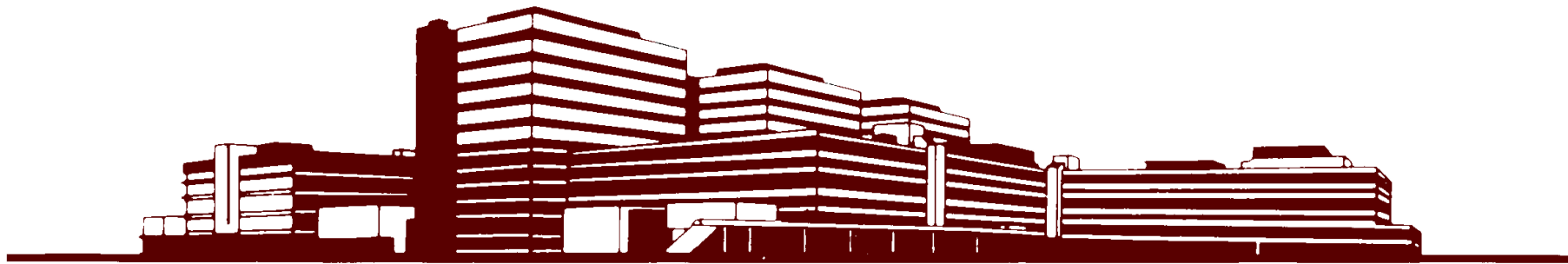


**Review of ten years of cytogenetic
analysis of IUFD products
towards a molecular approach**



Intra uterine fetal death

- in utero fetal death from 16 weeks of gestation till the moment of delivery.
- Rate of IUFD: 0,7% of all gestations
- Causes: infection, severe congenital anomalies, placental lesions, trauma, maternal diseases (diabetes and blood coagulation disorders), **genetic abnormalities.**

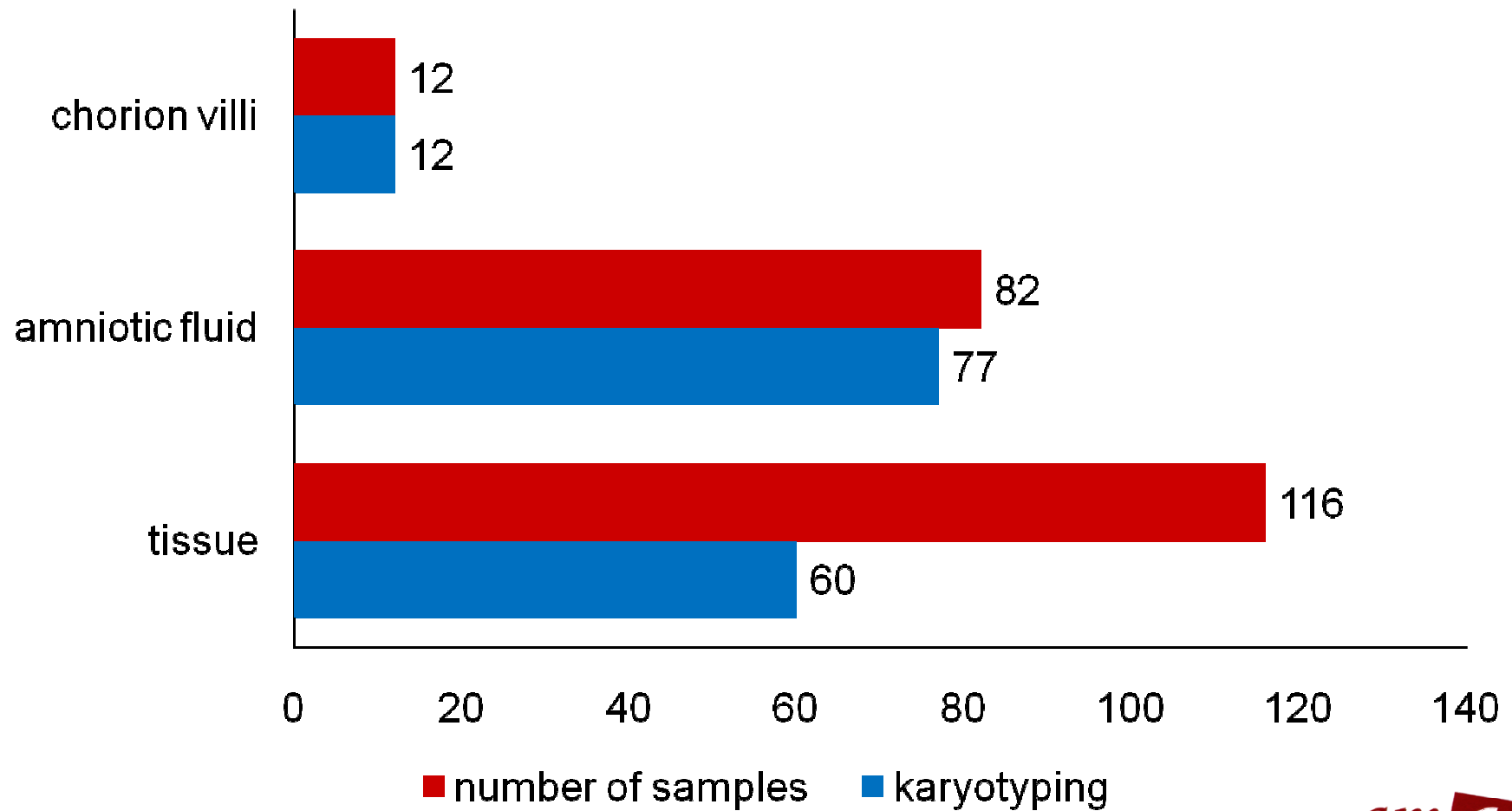
Genetic abnormalities

- Genetic etiology: 25% of the IUFD
- Cytogenetic visible: 6%-17%
 - Trisomies
 - Monosomies
 - Confined placental mosaicism
- Other genetic etiology:
 - Single gene mutations
 - X-linked disorders
 - Micro deletions/duplications

Current protocol

- Amniotic fluid (AF): karyotyping
- Chorionic villi (CV): karyotyping
- Tissue of IUFD:
 - multiple congenital anomalies: karyotyping
 - no observed abnormalities: Direct FISH for chromosomes 13, 18, 21, X and Y
 - culture failure: FISH

Success rate karyotyping 2000-2010



Results 2000-2010

	AF/CV	tissue	tissue
	karyotyping	karyotyping	Nuc ish
Normal	71	43	27
+13	1	0	5
+18	4	2	2
+21	6*	4	15
+22	1	1	-
45,X	3	3	5
47,XXY	1		1
Triploidy	3	1	1
Translocations	1 t(1;16)	1 t(5;16)	-
No growth	3	56	-
*1 combined with t(4;16), 1 combined with inv(20)			

Conclusion/question

- Many cultures done, low success rate (52%).
- Can karyotyping and FISH be replaced by a another method with a higher success rate?

QF-PCR

- No culture needed
- High success rate

- Specific QF-PCR for IUFD tissues
- Chromosomes: 13, 15, 16, 18, 21, 22, X and Y

Results 2000-2010

	AF/CV	tissue	tissue
	karyotyping	karyotyping	Nuc ish
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Validation IUFD QF-PCR

- Aim: replace FISH by IUFD QF-PCR
- Study design: 50 samples done with IUFD QF-PCR compared with earlier results
- Criteria: IUFD QF-PCR must show the same result as karyotyping/FISH

Near future protocol

- Amniotic fluid and chorionic villi of a IUFD: culture and karyotyping, success rate is high
- Tissue of IUFD:
 - DNA isolation from tissue
 - Specific QF-PCR for IUFD
 - When congenital anomalies are present and a normal QF-PCR result is found: array CGH
- Karyotyping of the parents