

POPULATION INCIDENCE OF THE PERICENTRIC INVERSION OF CHROMOSOME 9 IN THE CZECH POPULATION

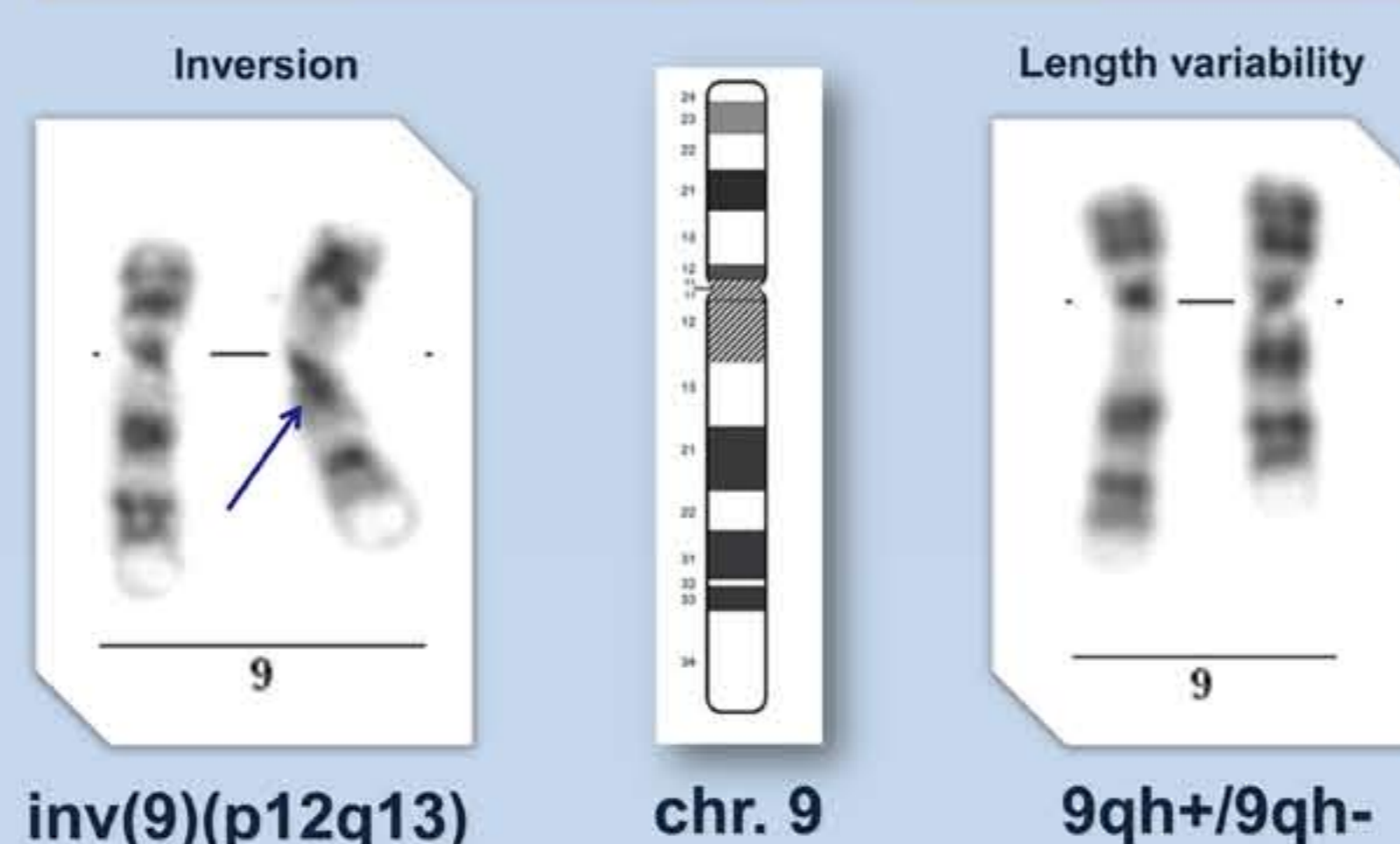
**Antonín Šípek Jr.^{1,2}, Aleš Panczak¹, Romana Mihalová¹, Lenka Hřčková¹,
Vladimír Sobotka³, Petr Lonský³, Nikola Kaspříková⁴, Antonín Šípek^{2,3}, Vladimír Gregor^{2,3}**

- 1) Institute of Biology and Medical Genetics, First Faculty of Medicine, Charles University and General University Hospital, Prague, Czech Republic
2) Department of Medical Genetics, Thomayer Hospital, Prague, Czech Republic
3) Department of Medical Genetics, Pronatal Sanatory, Prague, Czech Republic
4) Institute of Biophysics and Informatics, First Faculty of Medicine, Charles University, Prague, Czech Republic

Introduction:

Pericentric inversion of the human chromosome 9 [inv(9)] is the most common heterochromatin inversion in the human karyotype. Previous studies estimated the general incidence of this rearrangement to be 1-4% in the general population. Although widely believed to be an insignificant variant of human karyotype, various authors reported possible association of inv(9) and reproductive failure. Since inv(9) isn't specifically reported/registered in the Czech Republic, we have created 3 special study groups in order to estimate the population incidence of inv(9) in the Czech Republic.

Chromosome 9 variants



Methods:

The incidence of inv(9) was studied in 3 specific groups of healthy individuals: a) a group of gamete donors (n = 2288; taken from the cytogenetic laboratory at Pronatal Sanatory), b) a group of children awaiting adoption (n=814; from the database of Cytogenetic laboratory at Thomayer Hospital) and c) a cohort created from fetuses karyotyped solely because of the advanced maternal age of their mothers (n = 1064; from the database of Cytogenetic laboratory at the General University Hospital). All karyotype examinations were performed using the standard G-banding visualization method.

Study Group	Total			Females			Males			F/M difference
	inv(9) cases	All cases	Population incidence (%)	inv(9) cases	All cases	Population incidence (%)	inv(9) cases	All cases	Population incidence (%)	P value (95% CI)
Gamete Donors	42	2288	1.84%	41	2092	1.96	1	196	0.51%	0.257 (0.654 - 158.39)
Children awaiting adoption	14	814	1.72%	7	380	1.84	7	434	1.61%	1.0 (0.339 - 3.862)
Fetuses of mothers with advanced maternal age	12	1064	1.13%	8	551	1.45	4	513	0.78%	0.389 (0.498 - 8.555)
Total	68	4166	1.63%	56	3023	1.85	12	1143	1.05%	0.075 (0.938 - 3.660)

Table 1 - Numbers and estimate of population incidence of cases with inv(9) – data from three specific population samples, Prague, Czech Republic

Laboratory	Total			Females			Males			F/M difference
	inv(9) cases	All records	Laboratory Incidence (%)	inv(9) cases	All records	Laboratory Incidence (%)	inv(9) cases	All records	Laboratory Incidence (%)	P value (95% CI)
General University Hospital	170	10 933	1.55%	105	5943	1.77%	65	4990	1.30%	0.052 (0.988 - 1.891)
Thomayer University Hospital	131	8 611	1.52%	66	4553	1.45%	65	4058	1.60%	0.597 (0.630 - 1.297)
Pronatal® Sanatorium	120	7 053	1.70%	81	4562	1.78%	39	2491	1.57%	0.564 (0.768 - 1.716)
Total	421	26597	1.58%	252	15058	1.67%	169	11539	1.46%	0.181 (0.937 - 1.400).

Table 2 - Numbers and laboratory incidence of cases with inv(9) – data from three cytogenetic laboratories, Prague, Czech Republic

Results:

The total incidence of inv(9) in all groups was 1.63%, S.D. = 0.3%. If evaluated separately by gender, the incidence of inv(9) was higher in individuals of female gender in all three groups (but the difference was not statistically significant).

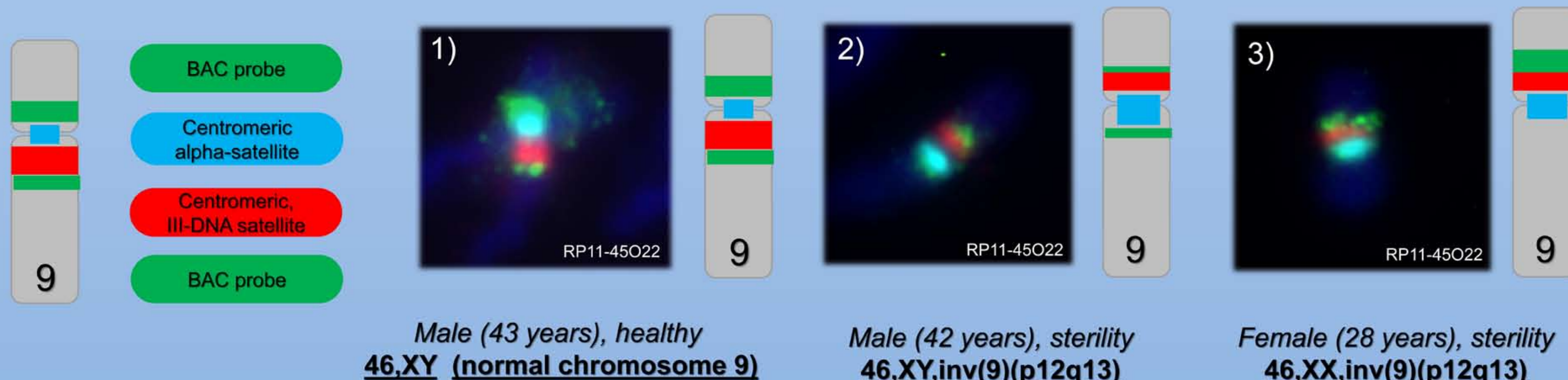
Discussion:

The estimated incidence of inv(9) (range 1.13%–1.84% or 1.63% overall) doesn't differ from values reported in other population samples. The higher proportion of females among inv(9) carriers has also been reported before, however no explanation has been given so far.

Conclusion:

The inv(9) polymorphism is quite common finding in the Czech Population. No significant difference to other studies/populations has been found.

Advanced visualization of inv(9) using our multiprobe-FISH method



- References:
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