

Sélection des spermatozoïdes chez les hommes porteurs de réarrangements chromosomiques équilibrés

Marine DOMART

Docteur junior en gynécologie-obstétrique 2ème année

Hôpital Pierre Rouquès Les Bluets-Paris

GYNAZUR

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Caryotype



1



2



3



4



5



6



7



8



9



10



11



12



13



14



15



16



17



18



19



20



21



22



X Y

or

X X

Shown here are human chromosomes

Indications du caryotype masculin en PMA

1. Oligozoospermie sévère

- < 5 millions de spermatozoïdes/mL
- Plus la concentration est basse, plus la probabilité d'anomalie chromosomique est élevée (jusqu'à 10% dans les azoospermies)

2. Azoospermie (absence totale de spermatozoïdes)

- Obstructive ou non obstructive

3. Fausses couches à répétition

Recherche de translocation équilibrée ou d'anomalies constitutionnelles chez l'homme

4. Antécédents personnels ou familiaux évocateurs

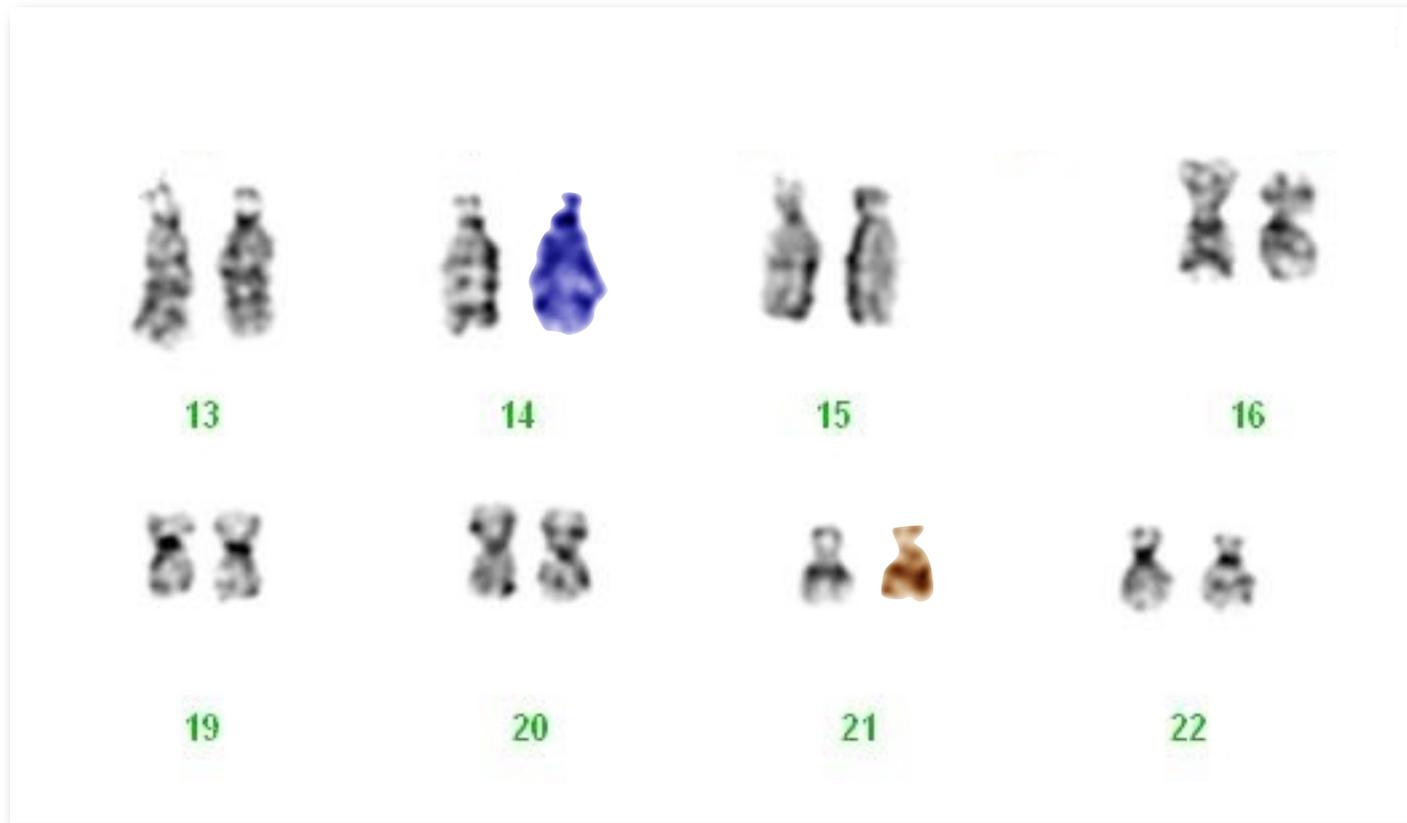
- Retard pubertaire, micropénis, gynécomastie
- Antécédents de stérilité familiale ou de malformations génitales

6. Syndrome clinique évocateur (suspicion de syndrome de Klinefelter (47,XXY))

Réarrangements chromosomiques équilibrés

- **Translocations Robertsoniennes**
- **Translocations réciproques**
- **Inversions péricentriques**

Translocation Robertsonienne



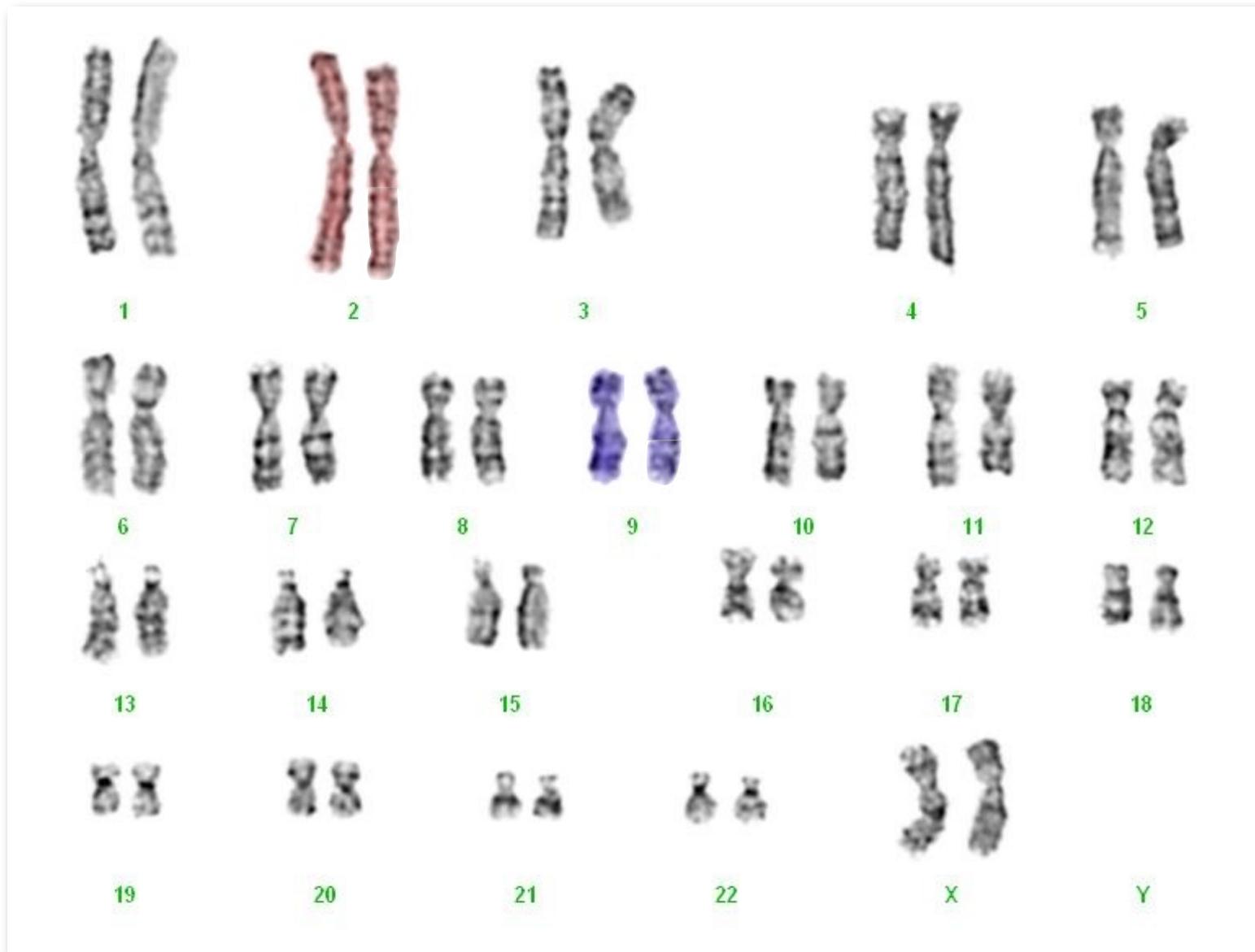
Translocation Robertsonienne



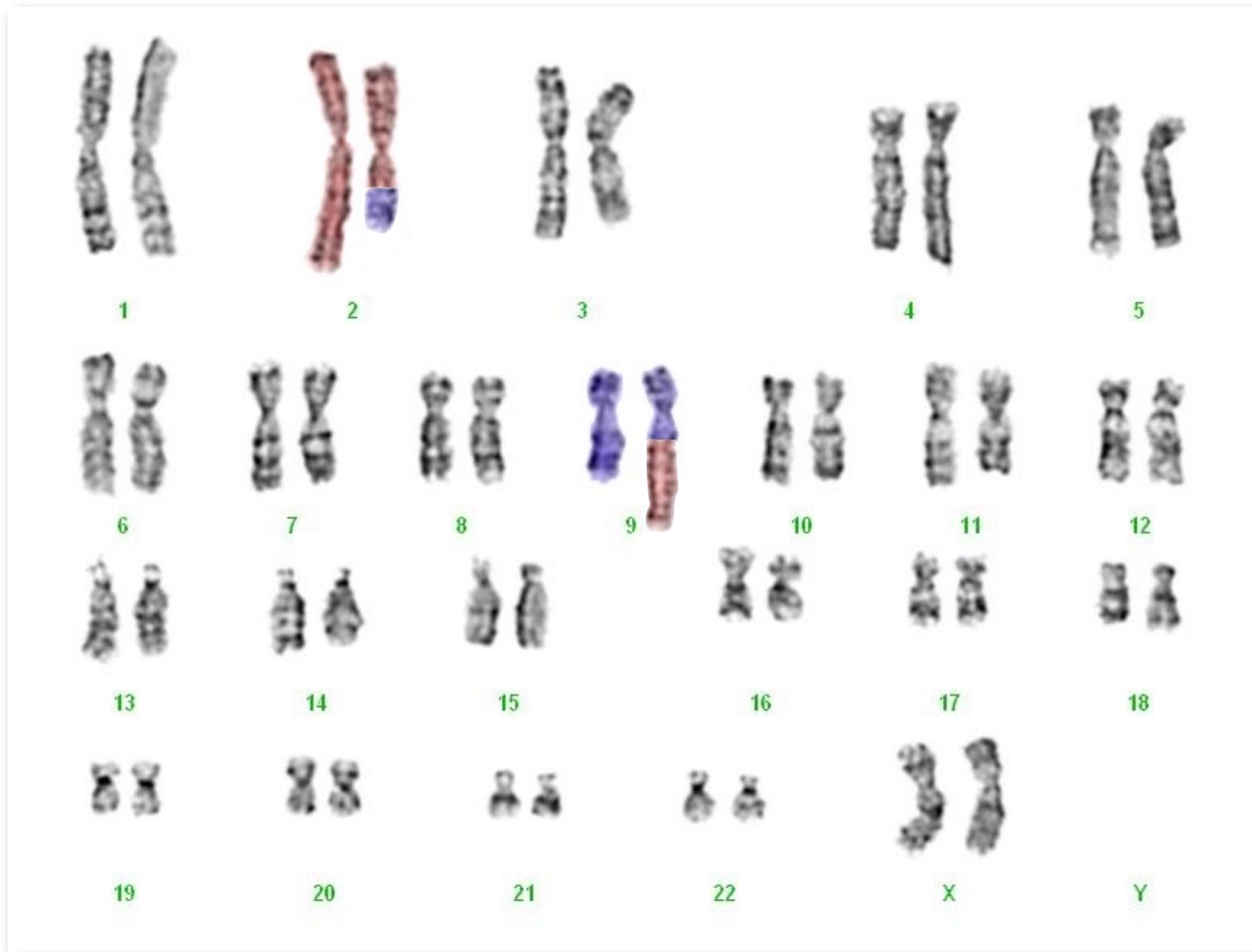
Translocation Robertsonienne



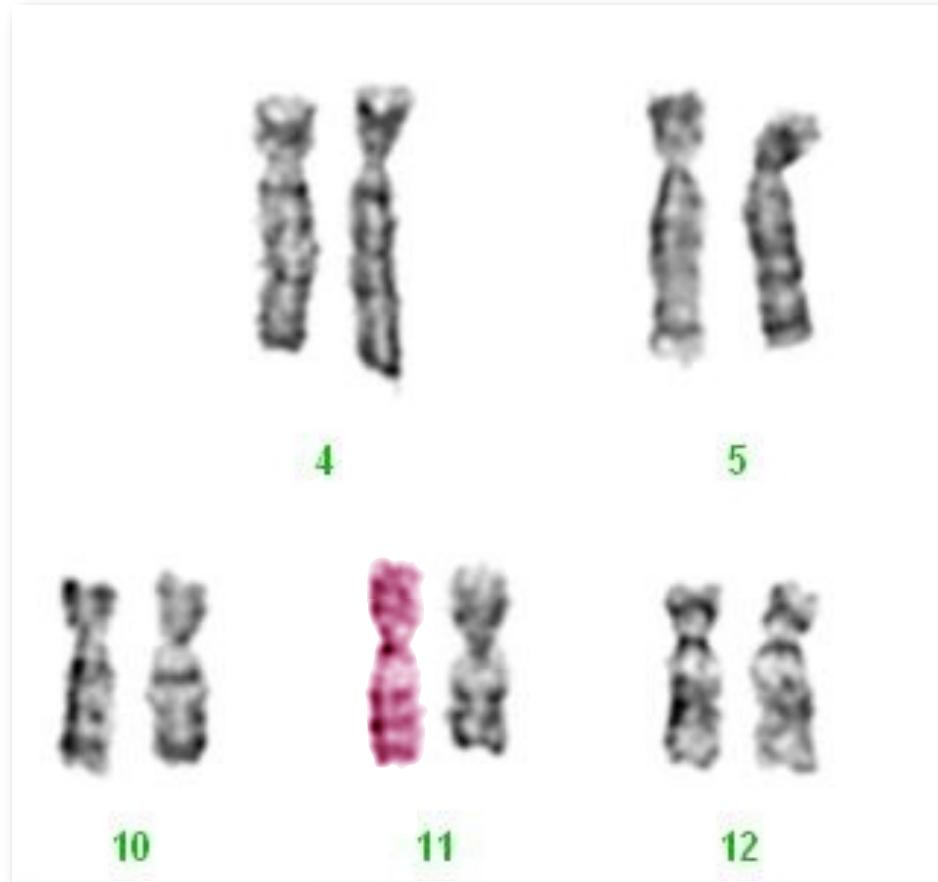
Translocation réciproque



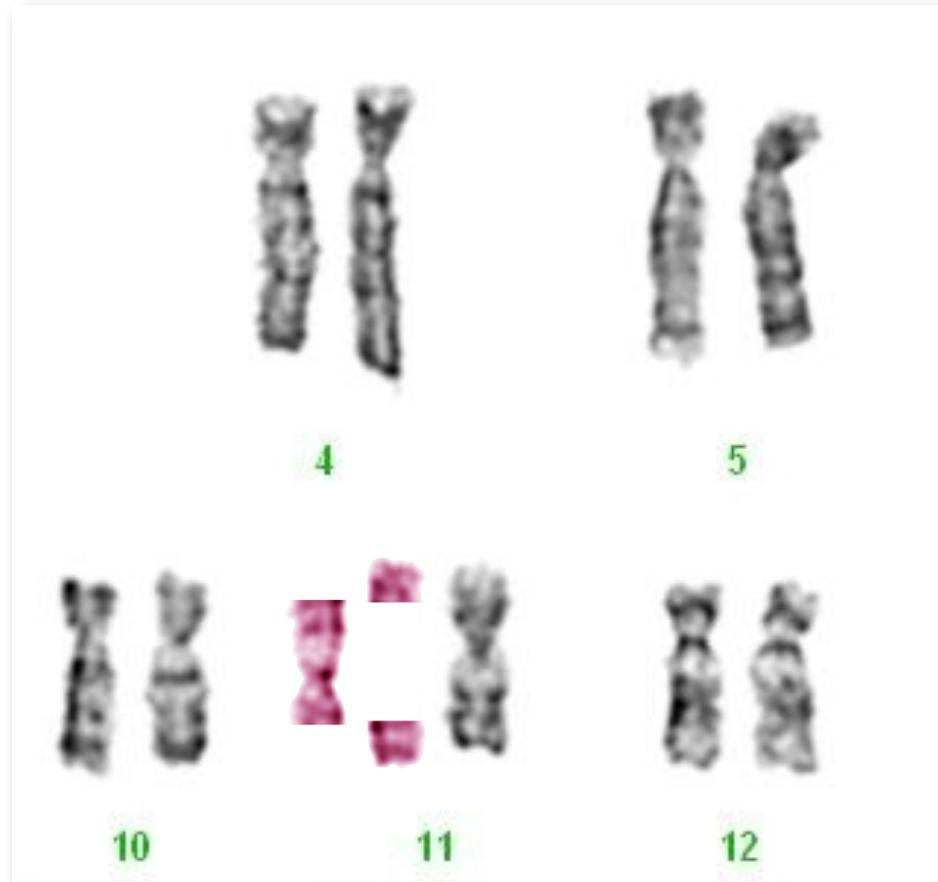
Translocation réciproque



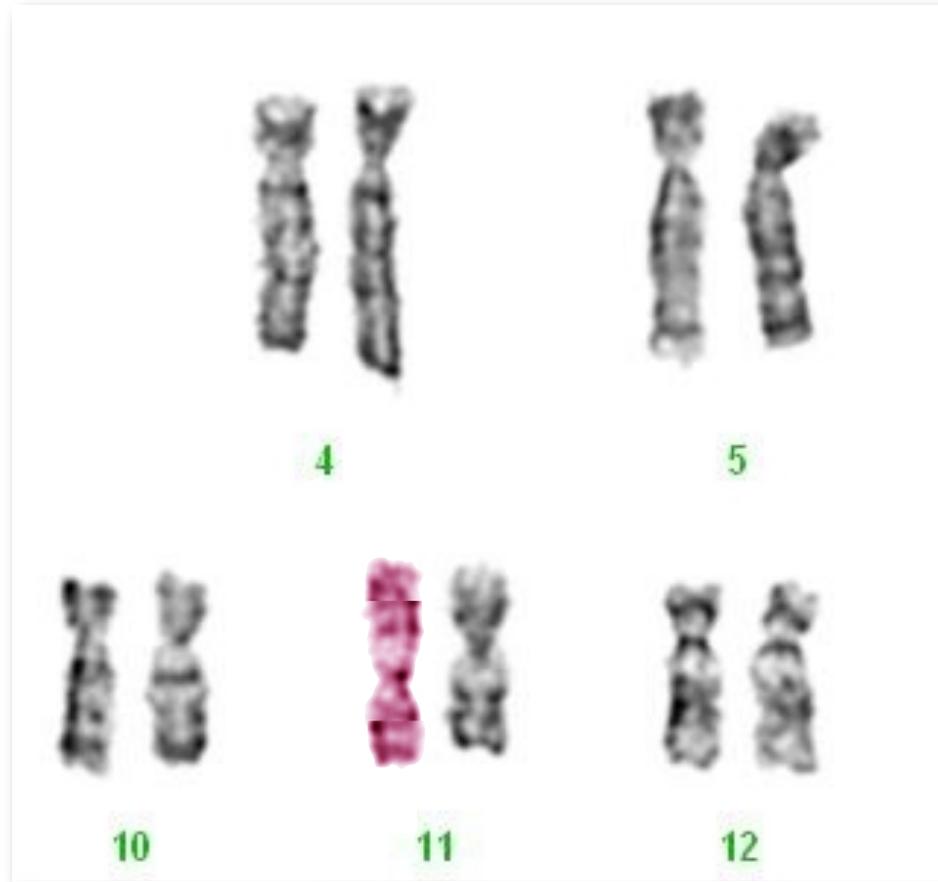
Inversion péricentrique



Inversion péricentrique



Inversion péricentrique



Phénotype normal

Car anomalie équilibrée

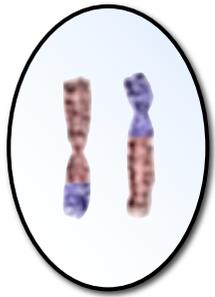
Reproduction

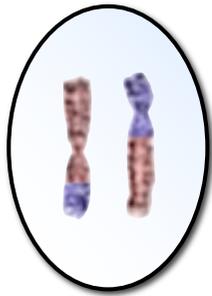
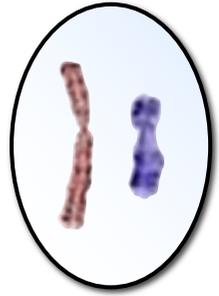
- Parfois, OATS/azoospermie ou IOP
- Toujours, malségrégation chromosomique

Ségrégation chromosomique : translocation réciproque

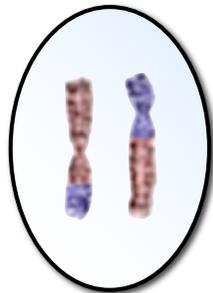




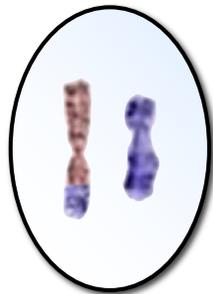
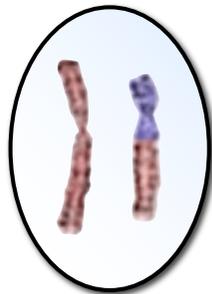




Mode Alterne



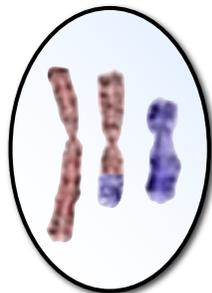
Mode Alterne



Mode Adjacent I

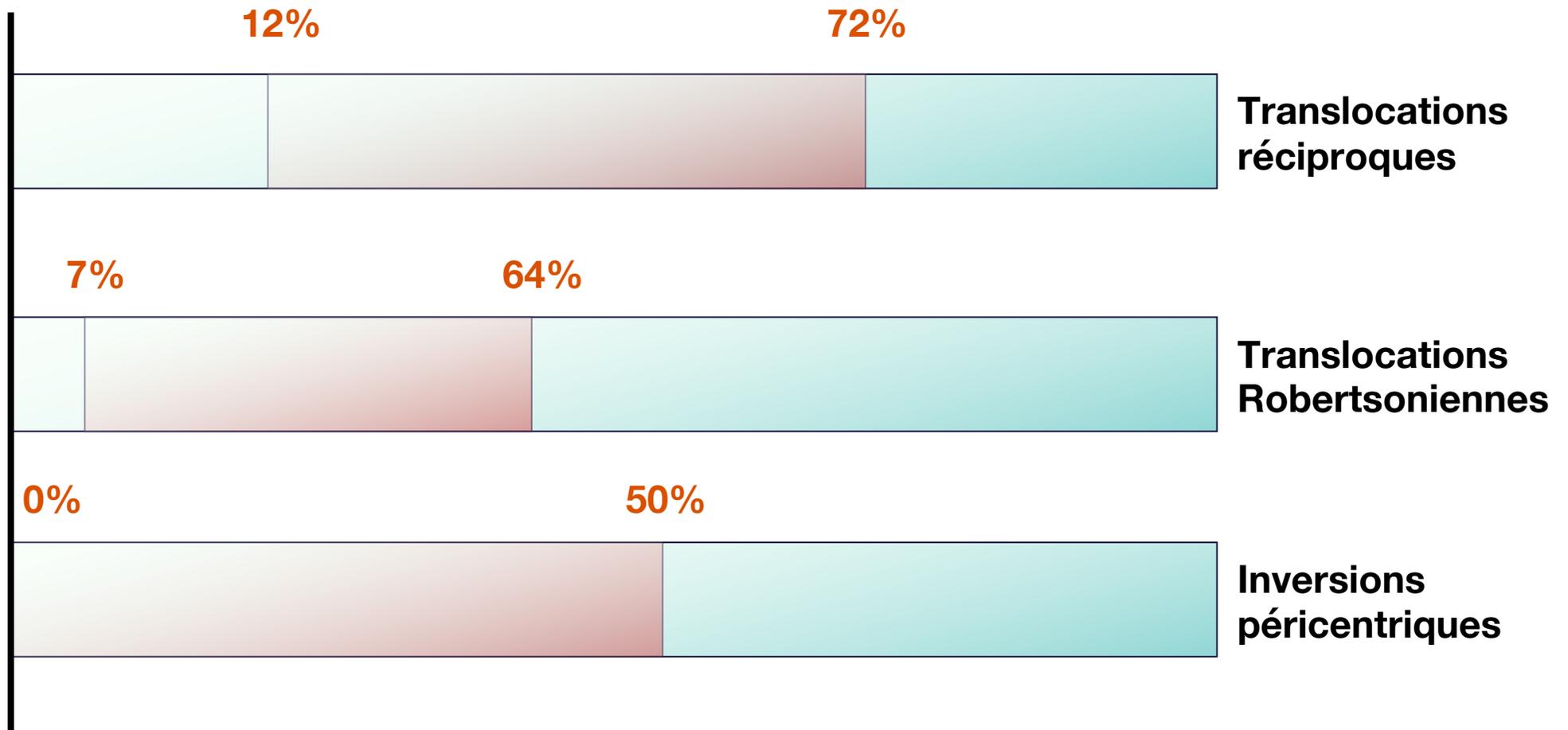


Mode Adjacent II

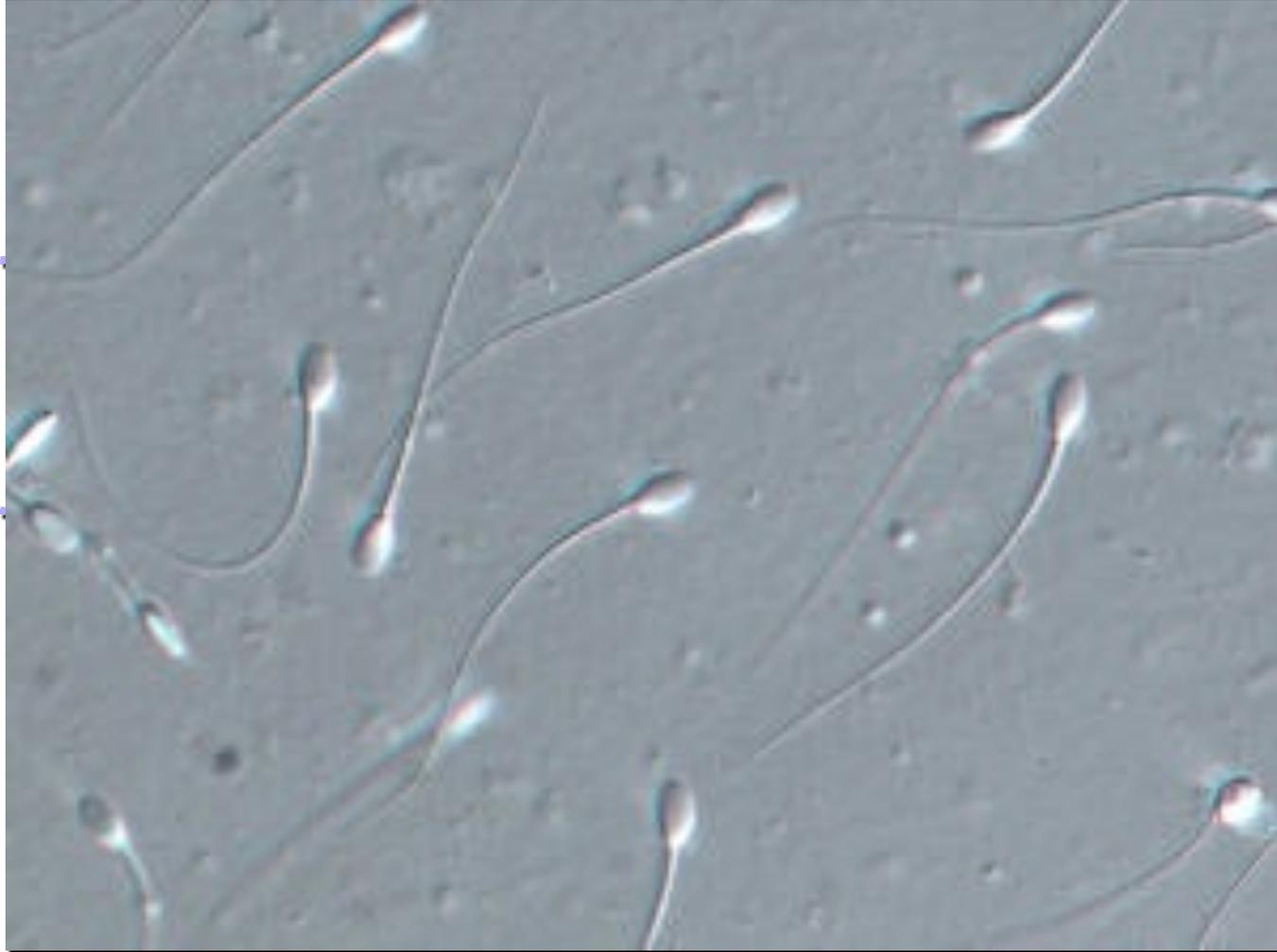
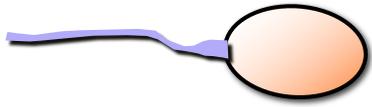


Mode 3:1

Proportions de spermatozoïdes déséquilibrés



(Coutton *et al.*, 2023; Morel *et al.*, 2004, Rouen *et al.*, 2017)



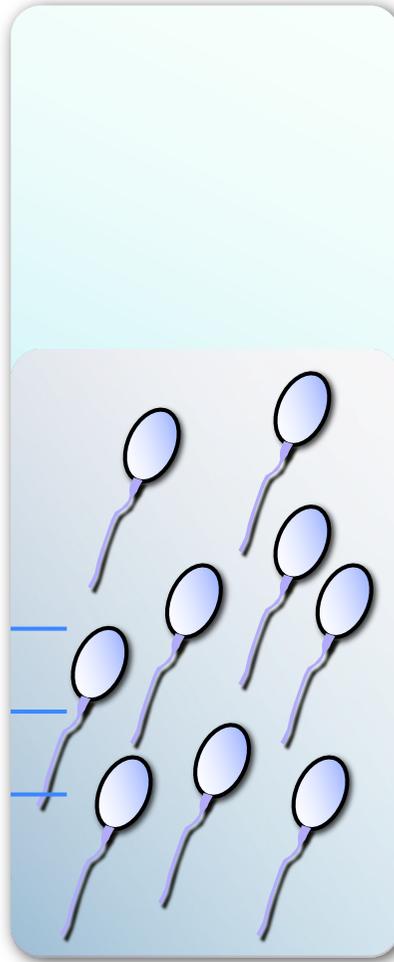
Prise en charge

- Conception naturelle avec diagnostic prénatal (DPN)
- Diagnostic préimplantatoire (DPI)

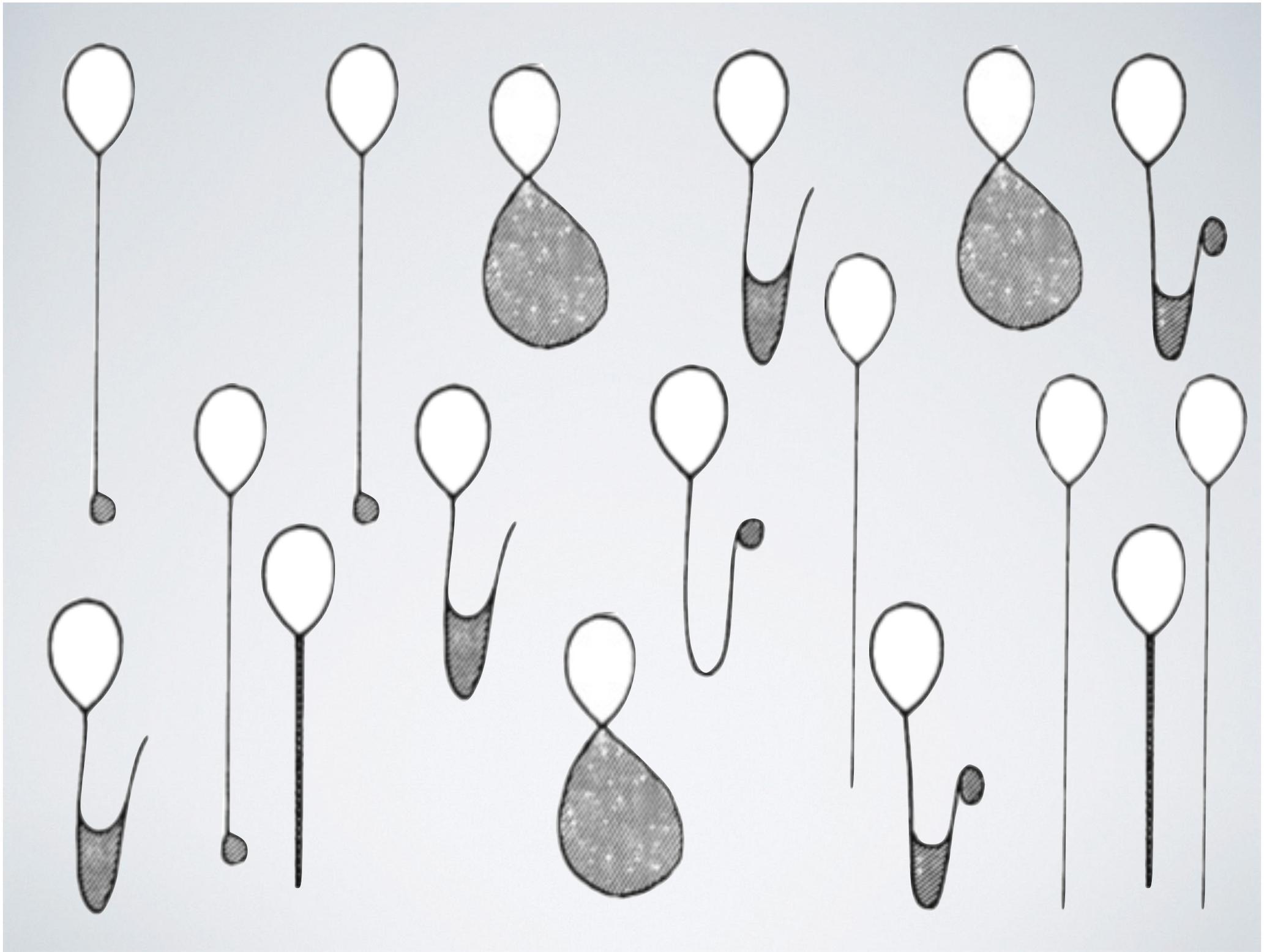
Hypo-Osmotic Swelling Test (HOST)

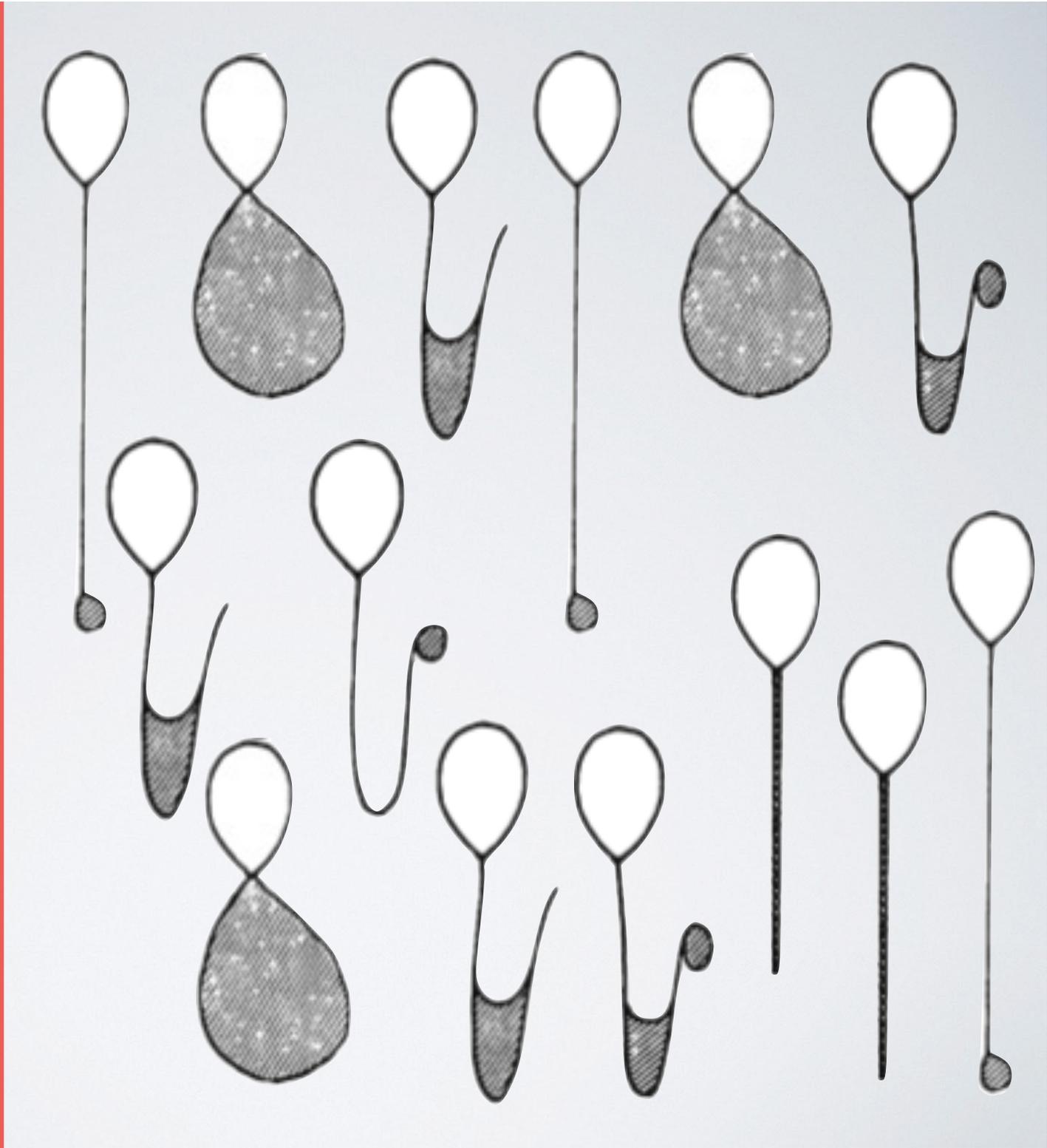
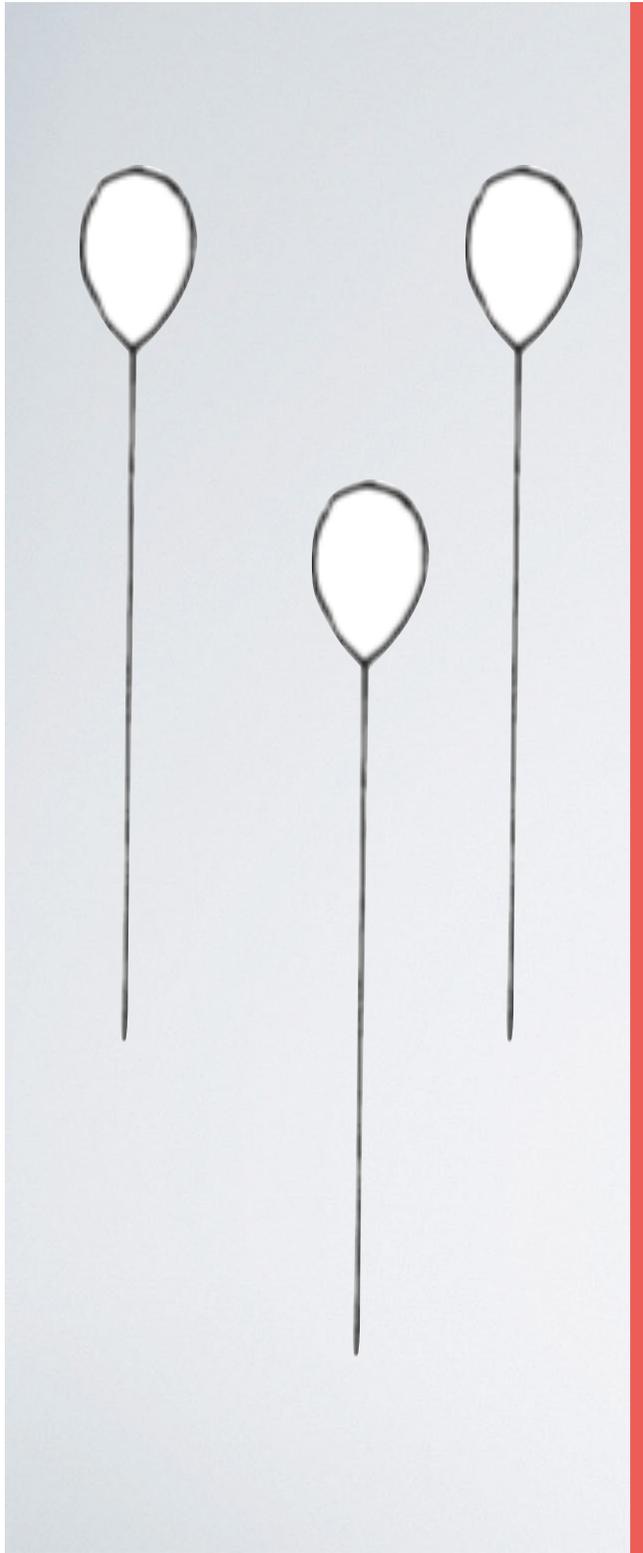


Hypo-Osmotic Swelling Test (HOST)

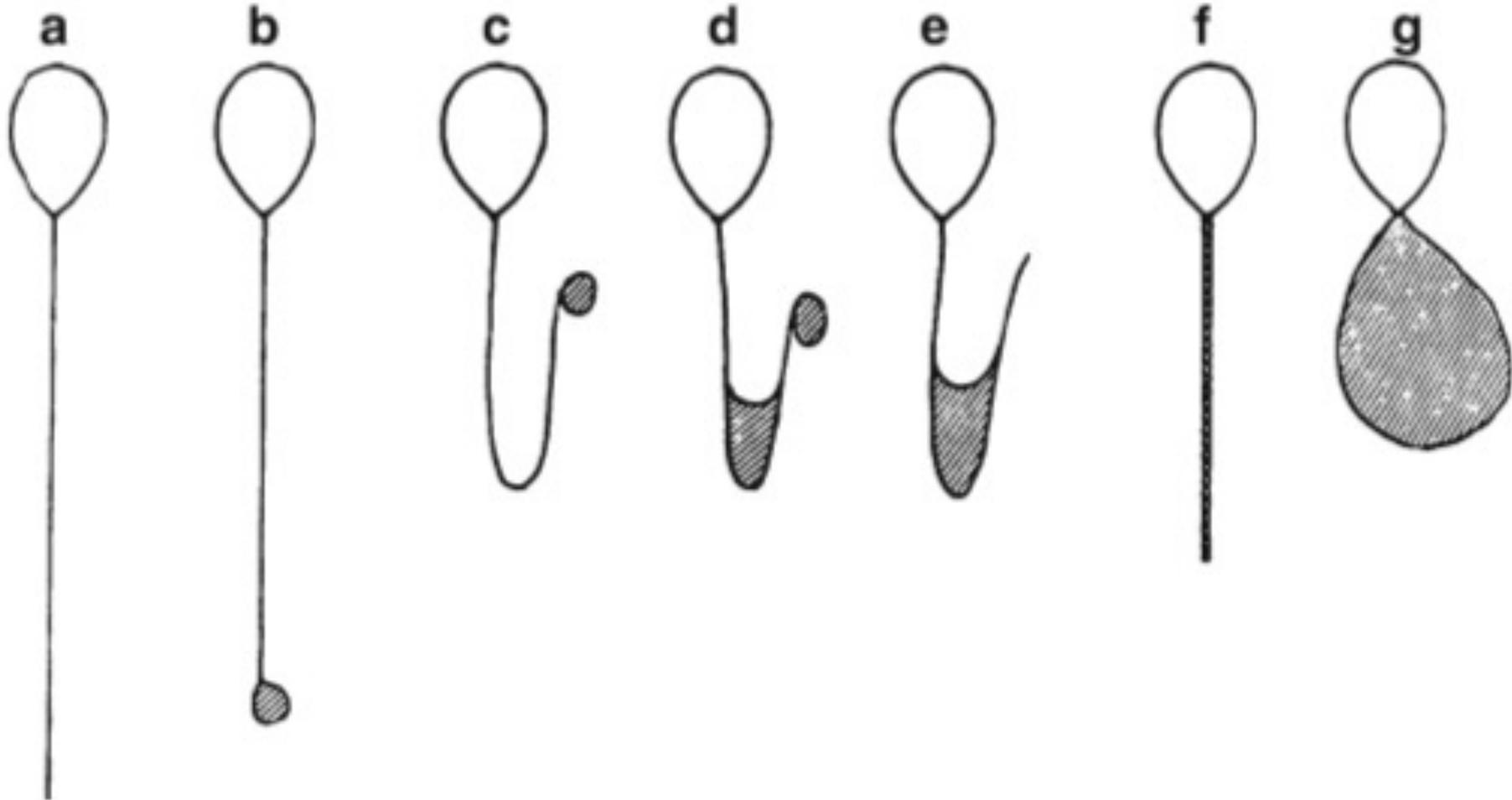


**Solution hypo-osmotique
(NaCl dilué au demi)**



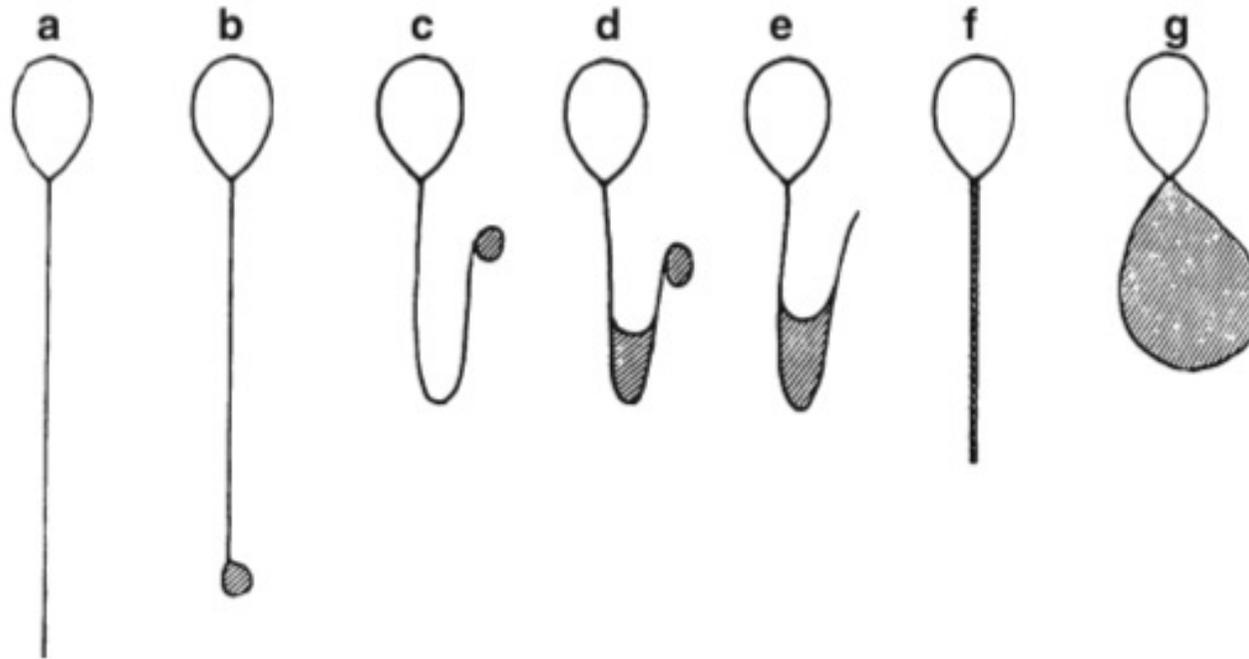


Hypo-Osmotic Swelling Test (HOST)

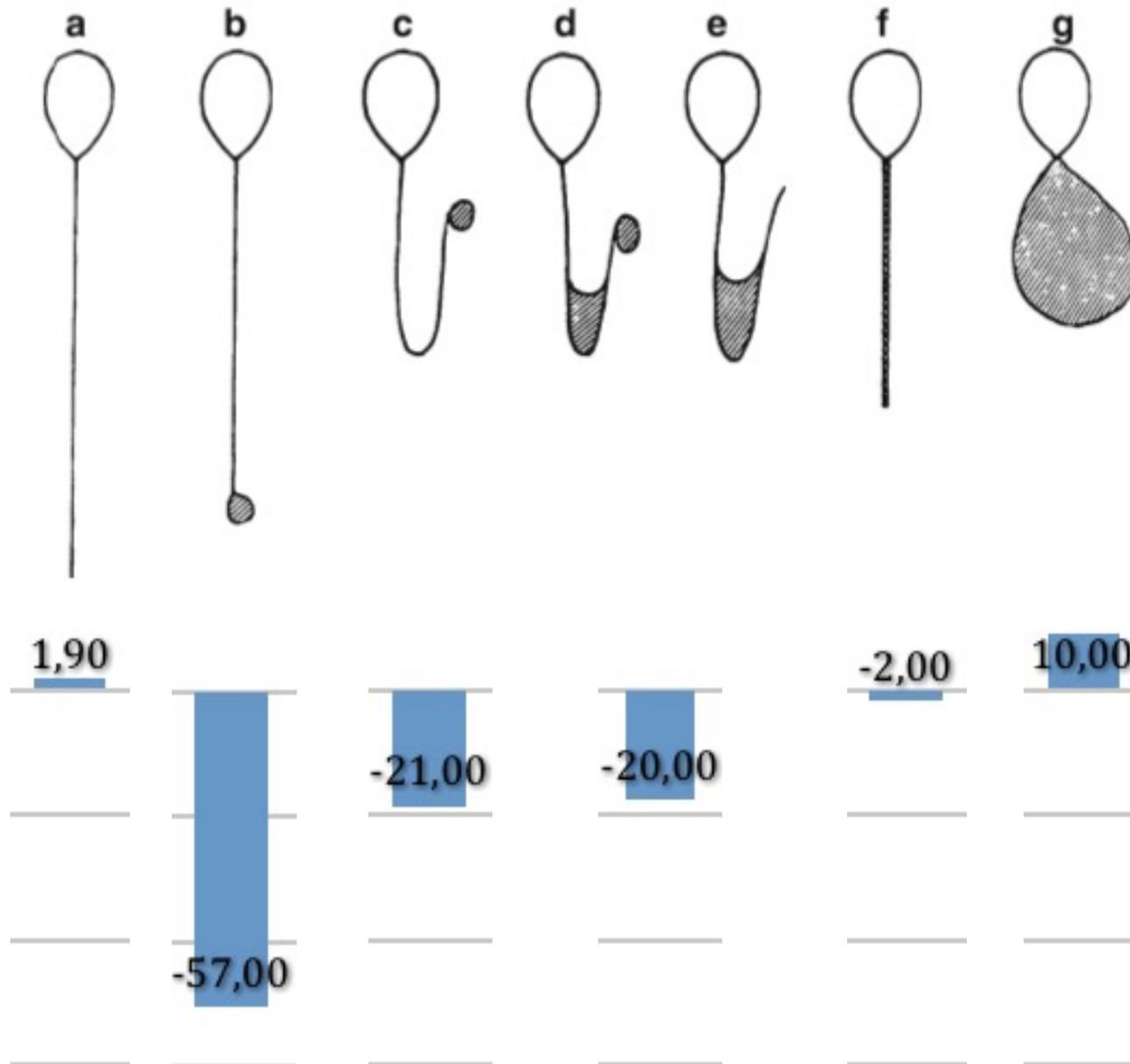


Hypo-Osmotic Swelling Test (HOST)

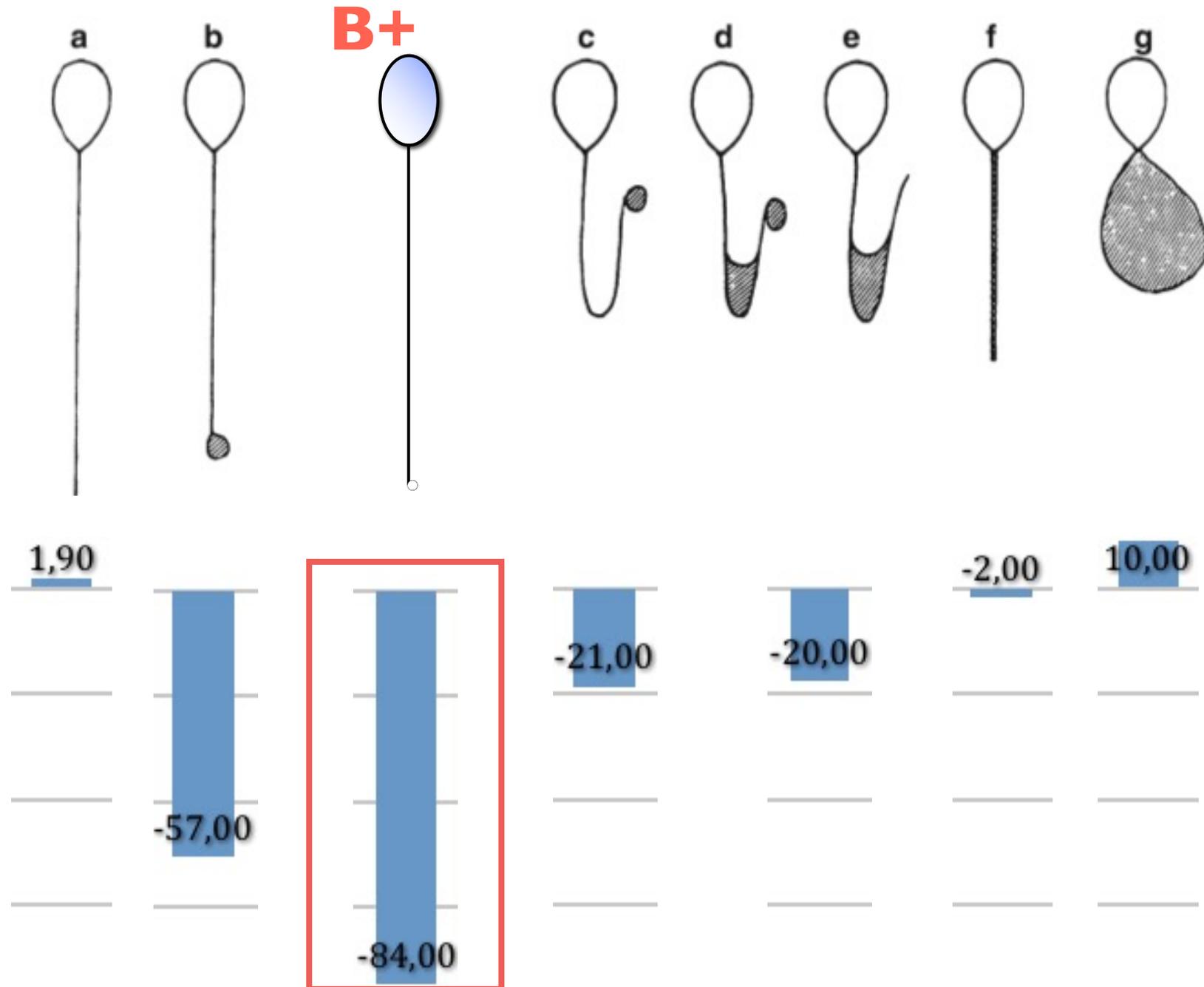
% global de spermatozoïdes déséquilibrés
VS.

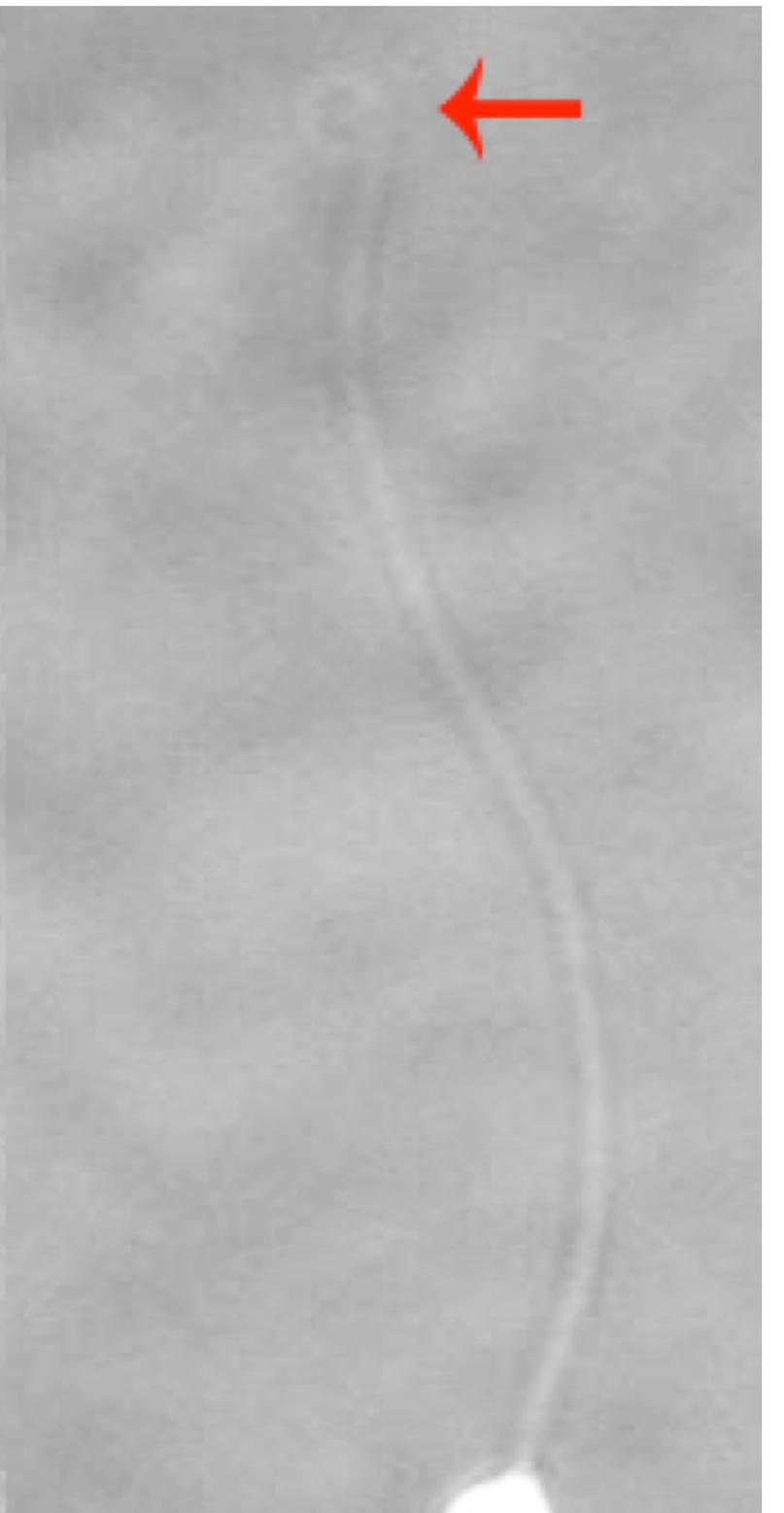
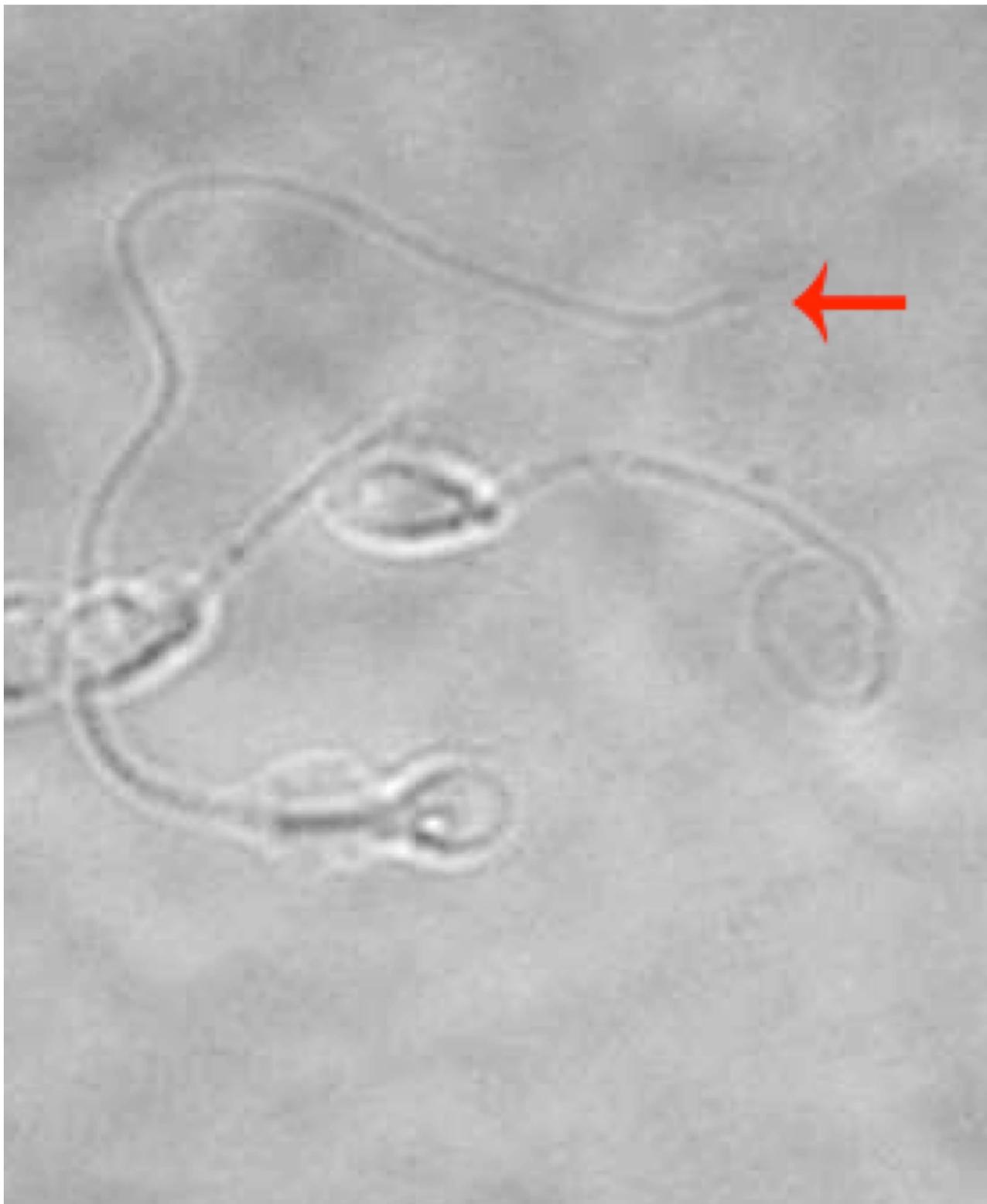


Hypo-Osmotic Swelling Test (HOST)

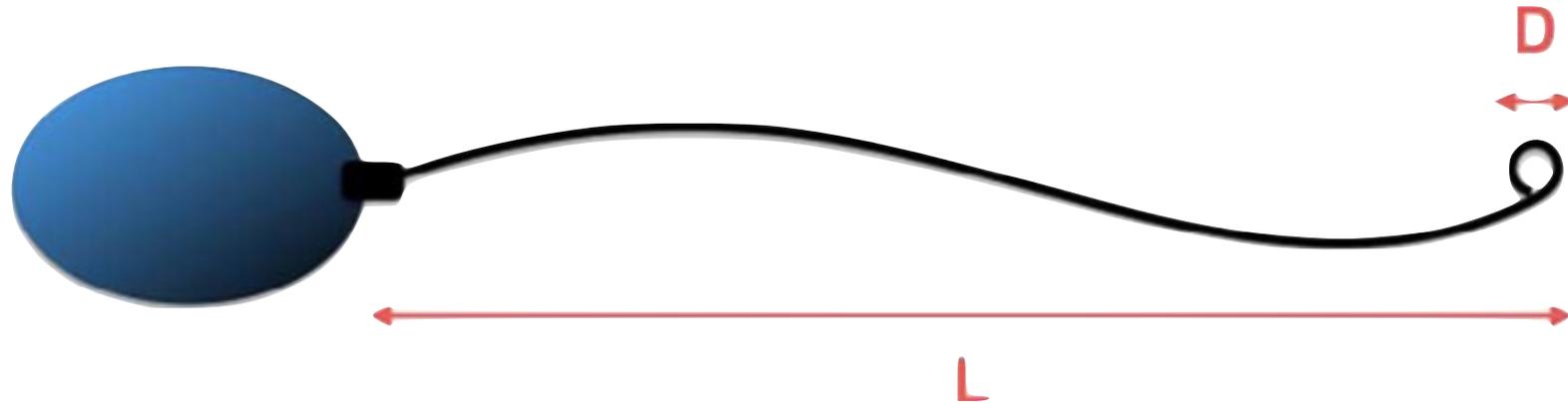


Hypo-Osmotic Swelling Test (HOST)



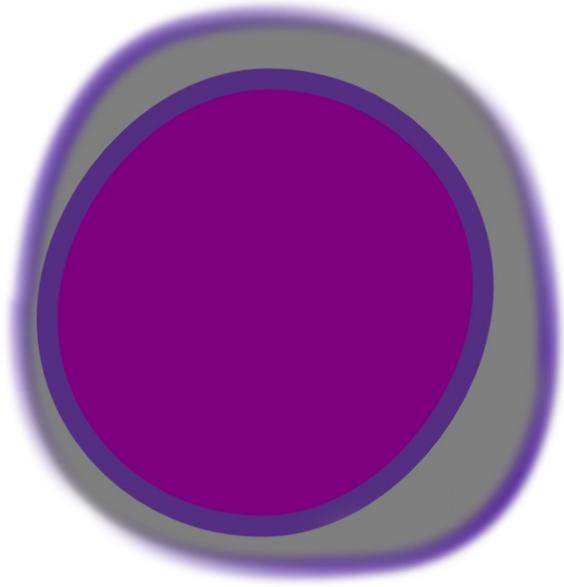


Hypo-Osmotic Swelling Test (HOST)

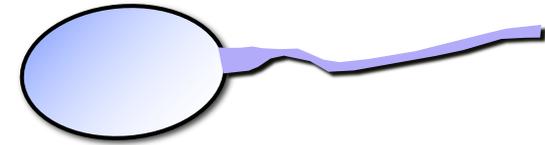


$$L/D > 20$$

Physiologie spermatique

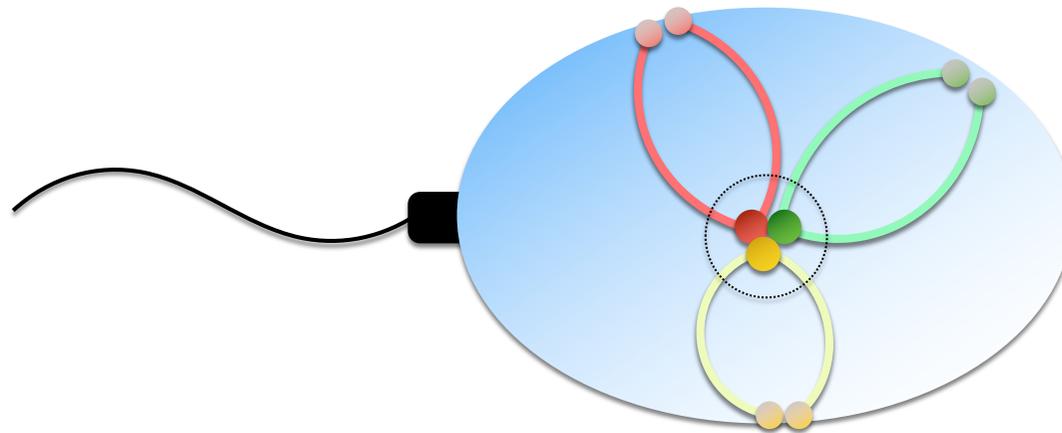
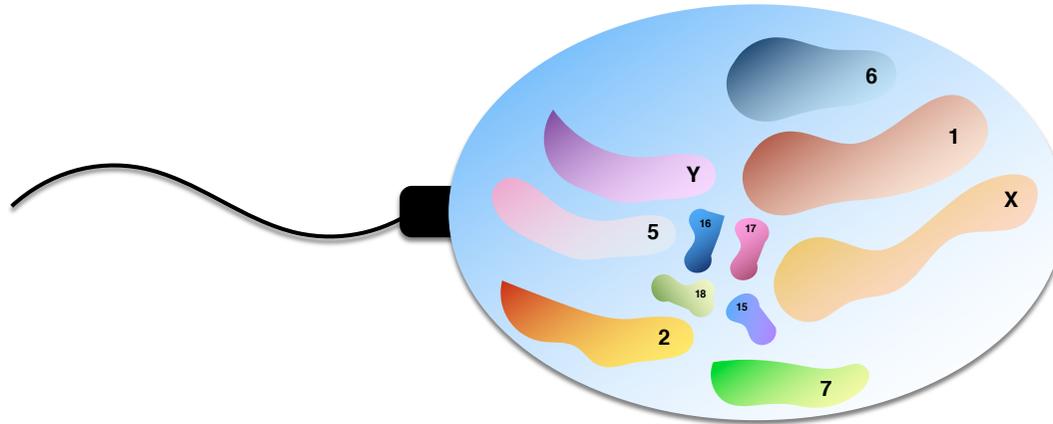


600 - 1400 μm^3

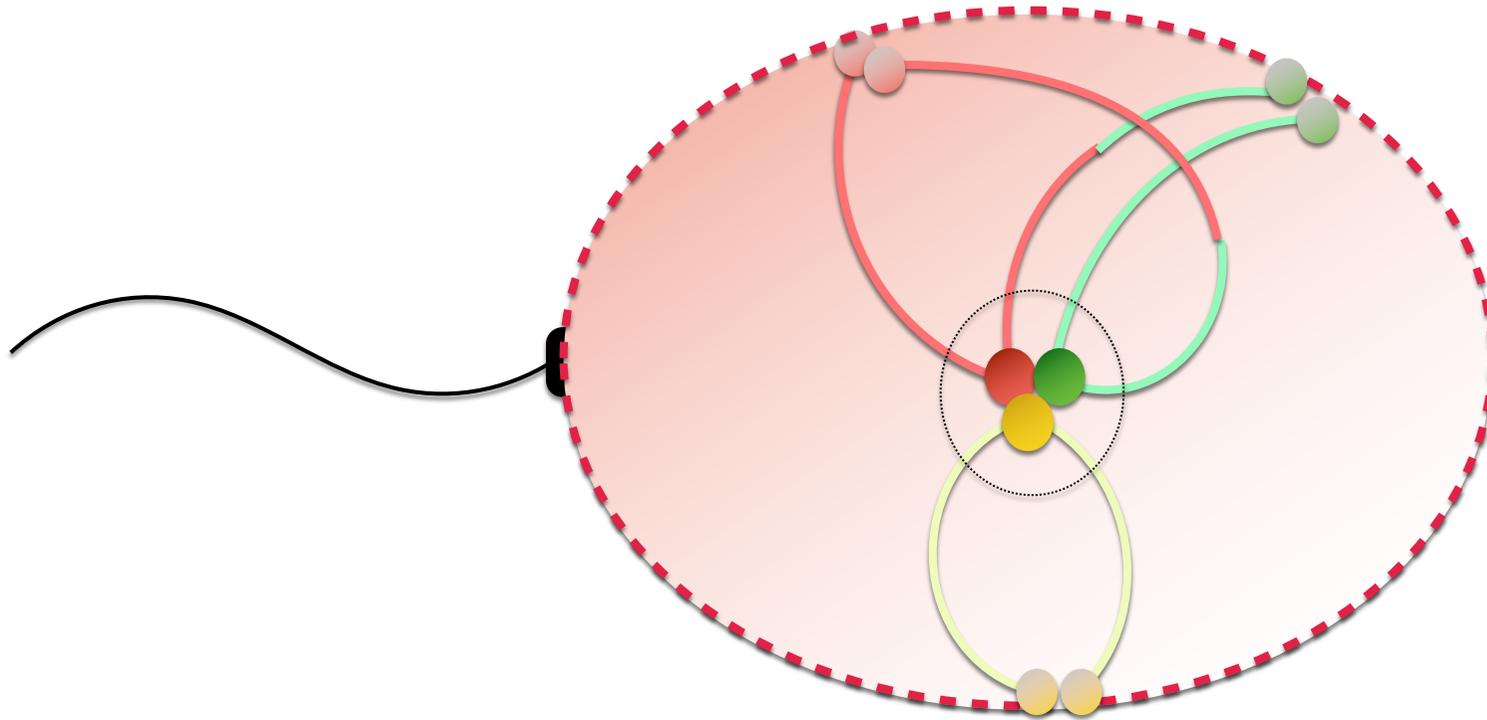


60 μm^3

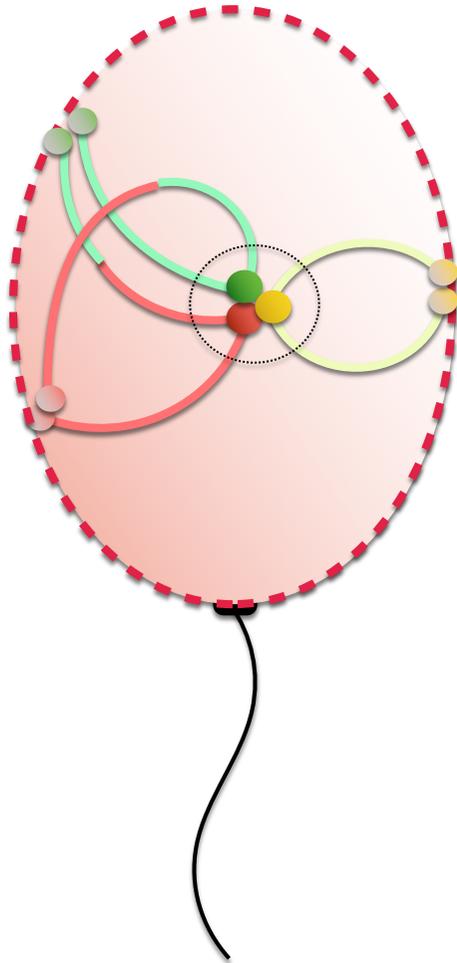
Physiologie spermatique



Physiologie spermatique



Physiologie spermatique

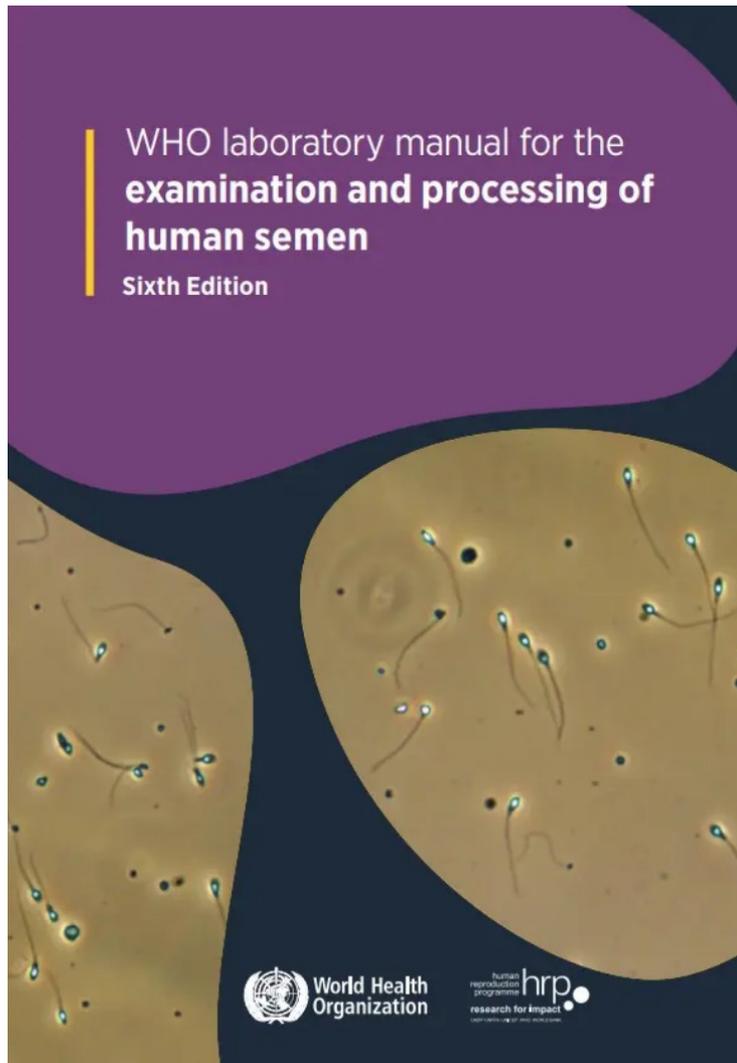


- **Volume nucléaire augmenté en microscopie confocale**

- **Éliminés partiellement par le test de migration survie (TMS) : 23% (6-66)**

- **Fragmentation de l'ADN augmentée**

Inocuité du HOST

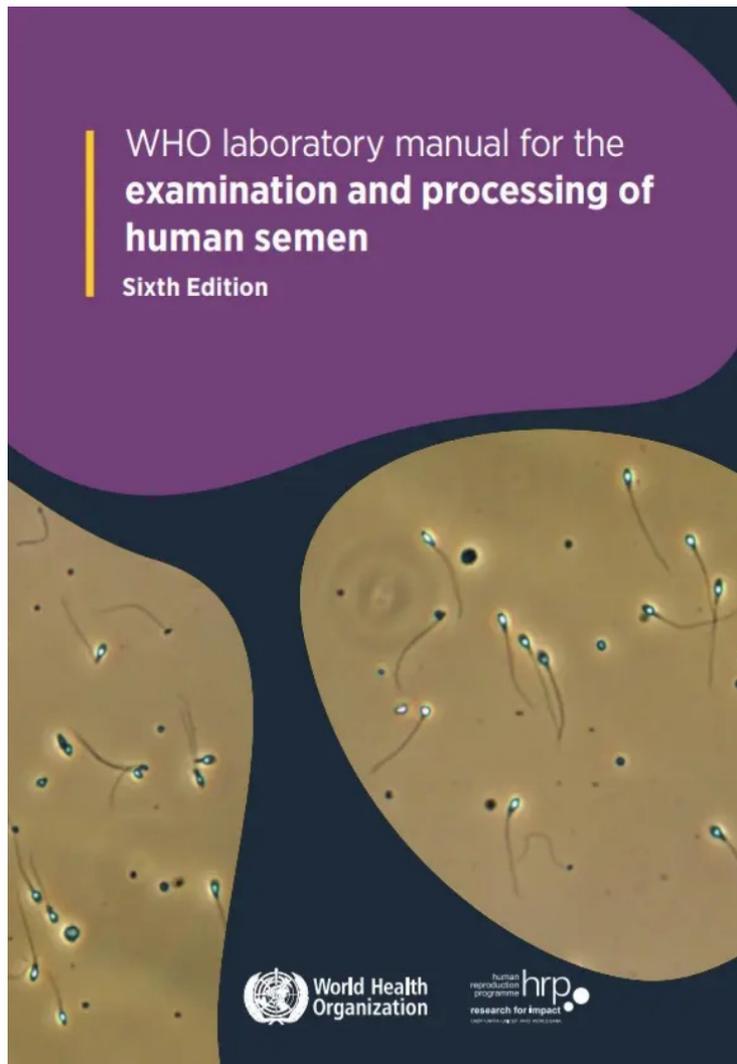


2.5.13.2 Vitality test using hypo-osmotic swelling

As an alternative to dye exclusion, the hypo-osmotic swelling test may be used to assess vitality (145). This is useful when staining of spermatozoa must be avoided, e.g. when choosing spermatozoa for intracytoplasmic sperm injection (ICSI). The hypo-osmotic swelling test presumes that only cells with intact membranes (live cells) can swell in hypotonic solutions. Spermatozoa with intact membranes swell within 5 minutes in hypo-osmotic medium, and all flagellar shapes are stabilized by 30 minutes (146).

- Use 30 minutes incubation for routine diagnostics.
- Use 5 minutes incubation when spermatozoa are to be processed for therapeutic use.

Innocuité du HOST



(2021)

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- Use 5 minutes incubation when spermatozoa are to be processed for therapeutic use.

Hypo-Osmotic Swelling Test (HOST)

Potential selection of genetically balanced spermatozoa based on the hypo-osmotic swelling test in chromosomal rearrangement carriers

**Alexandre Rouen ^{a,*}, Léa Carlier ^a, Solveig Heide ^a, Matthieu Egloff ^a,
Pauline Marzin ^a, Flavie Ader ^a, Mathias Schwartz ^a, Eli Rogers ^a,
Nicole Joyé ^a, Richard Balet ^b, Nathalie Lédée ^b, Laura Prat-Ellenberg ^b,
Nino Guy Cassuto ^c, Jean-Pierre Siffroi ^a**

(Rouen *et al.*, 2017)

B-SCOPE

B+ Selection after Conditioned Osmotic Processing and Evaluation

Balanced sperm selection in chromosomal rearrangement carriers: meiotic segregation results from 34 balanced chromosomal carriers in our center over 7 years.

Marine Domart, MD,^{1,2} Capucine Rossi, BS,¹ Eli J Rogers, MD,¹ Nino Guy Cassuto, MD,³ Léa Ruoso, PharmD,³ Laura Prat-Ellenberg, MD,² Jean-Pierre Siffroi, MD, PhD,¹ Pierre Lévy, MD,⁴ Alexandre Rouen*, MD, PhD^{1,2,5}

(Domart *et al.*, en cours de revue)

B-SCOPE

Subject	Chromosomal rearrangement	Unbalanced spermatozoa (% native ejaculate)	Unbalanced spermatozoa (% DGC)	% Decrease with DGC	Unbalanced spermatozoa (%, B- SCOPE)	% Decrease with B-SCOPE
Reciprocal translocation						
1	t(1;2)(q43;q31)	52	47	10	13	75
2	t(1;4)(p31.1;p35.1)	54	38	30	25	54
3	t(1;7)(p34;p11.2)	66	42	36	10	85
4	t(1;10)(p23.3;q22.3)	56	40	29	16	71
5	t(1;12)(p35;q14)	60	57	5	28	53
6	t(2;4)(p23.1;p14)	42	38	10	17	60
7	t(2;6)(p15;p25.3)	57	53	7	16	72
8	t(3;7)(p11;p21)	53	31	42	9	83
9	t(3;11)(q29;q24.1)	71	53	25	27	62
10	t(5;7)(p14;q21)	39	39	0	0	100
11	t(5;8)(p13;q13)	71	67	6	13	82
12	t(5;10)(q34;p12.1)	65	45	31	9	86
13	t(5;18)(q23.1;q21.1)	39	33	15	11	72
14	t(6;8)(q15;q22)	56	51	9	30	46
15	t(6;11)(q23;q32)	40	40	0	13	68
16	t(6;14)(q16.1;q22)	61	60	2	40	34
17	t(7;10)(q31.3;q25.3)	32	15	53	10	69
18	t(7;15)(p12;q26)	52	42	19	0	100
19	t(7;15)(p15.1;q21.1)	65	50	23	11	83
20	t(8;12)(p23.1;p13.31)	51	43	16	7	86
21	t(9;10)(p10;p10)	46	30	35	17	63
22	t(9;13)(q32;q12.3)	53	49	8	10	81
23	t(11;22)(q23;q11)	72	72	0	26	64
24	t(13;17)(q33;q22)	51	45	12	10	80
Total		54	45	18	7	72

B-SCOPE

Subject	Chromosomal rearrangement	Unbalanced spermatozoa (% native ejaculate)	Unbalanced spermatozoa (% DGC)	% Decrease with DGC	Unbalanced spermatozoa (%, B- SCOPE)	% Decrease with B-SCOPE
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4	t(1;10)(p23.3;q22.3)	56	40	29	16	71
5	t(1;12)(p35;q14)	60	57	5	28	53
6	t(2;4)(p23.1;p14)	42	38	10	17	60
7	t(2;6)(p15;p25.3)	57	53	7	16	72
8	t(3;7)(p11;p21)	53	31	42	9	83
9	t(3;11)(q29;q24.1)	71	53	25	27	62
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16	t(6;14)(q16.1;q22)	61	60	2	40	34
17	t(7;10)(q31.3;q25.3)	32	15	53	10	69
18	t(7;15)(p12;q26)	52	42	19	0	100
19	t(7;15)(p15.1;q21.1)	65	50	23	11	83
20	t(8;12)(p23.1;p13.31)	51	43	16	7	86
21	t(9;10)(p10;p10)	46	30	35	17	63
22	t(9;13)(q32;q12.3)	53	49	8	10	81
23	t(11;22)(q23;q11)	72	72	0	26	64
24	t(13;17)(q33;q22)	51	45	12	10	80
Total		54	45	18	7	72

B-SCOPE

Subject	Chromosomal rearrangement	Unbalanced spermatozoa (% native ejaculate)	Unbalanced spermatozoa (% DGC)	% Decrease with DGC	Unbalanced spermatozoa (%, B- SCOPE)	% Decrease with B-SCOPE
Robertsonian translocation						
25	rob(13;14)	50	43	14	10	80
26	rob(13;14)	52	35	33	17	67
27	rob(13;14)	31	22	29	0	100
28	rob(13;14)	27	10	63	0	100
29	rob(13;14)(q10;q10)	36	31	14	9	75
30	rob(13;14)(q10;q10)	28	23	18	10	64
31	rob(14;15)(q10;q10)	19	14	26	0	100
32	rob(14;21)	38	36	5	7	82
Total		35	27	25	7	84
Inversion						
33	inv(2)(p23q21)	16	11	31	0	100
34	inv(6)(p21q22.3)	41	41	0	10	76
Total		28	26	16	5	88

B-SCOPE

Subject	Chromosomal rearrangement	Unbalanced spermatozoa (% native ejaculate)	Unbalanced spermatozoa (% DGC)	% Decrease with DGC	Unbalanced spermatozoa (%, B- SCOPE)	% Decrease with B-SCOPE
Robertsonian translocation						
25	rob(13;14)	50	43	14	10	80
26	rob(13;14)	52	35	33	17	67
27	rob(13;14)	31	22	29	0	100
28	rob(13;14)	27	10	63	0	100
29	rob(13;14)(q10;q10)	36	31	14	9	75
30	rob(13;14)(q10;q10)	28	23	18	10	64
31	rob(14;15)(q10;q10)	19	14	26	0	100
32	rob(14;21)	38	36	5	7	82
Total		35	27	25	7	84
Inversion						
33	inv(2)(p23q21)	16	11	31	0	100
34	inv(6)(p21q22.3)	41	41	0	10	76
Total		28	26	16	5	88

B-SCOPE

Translocations réciproques - 72%

Translocations Robertsoniennes - 84%

Inversions chromosomiques - 88%

Prise en charge

**Réarrangement
chromosomique
chez l'homme**



**Etude de la
ségrégation**

**Réarrangement
chromosomique
chez l'homme**

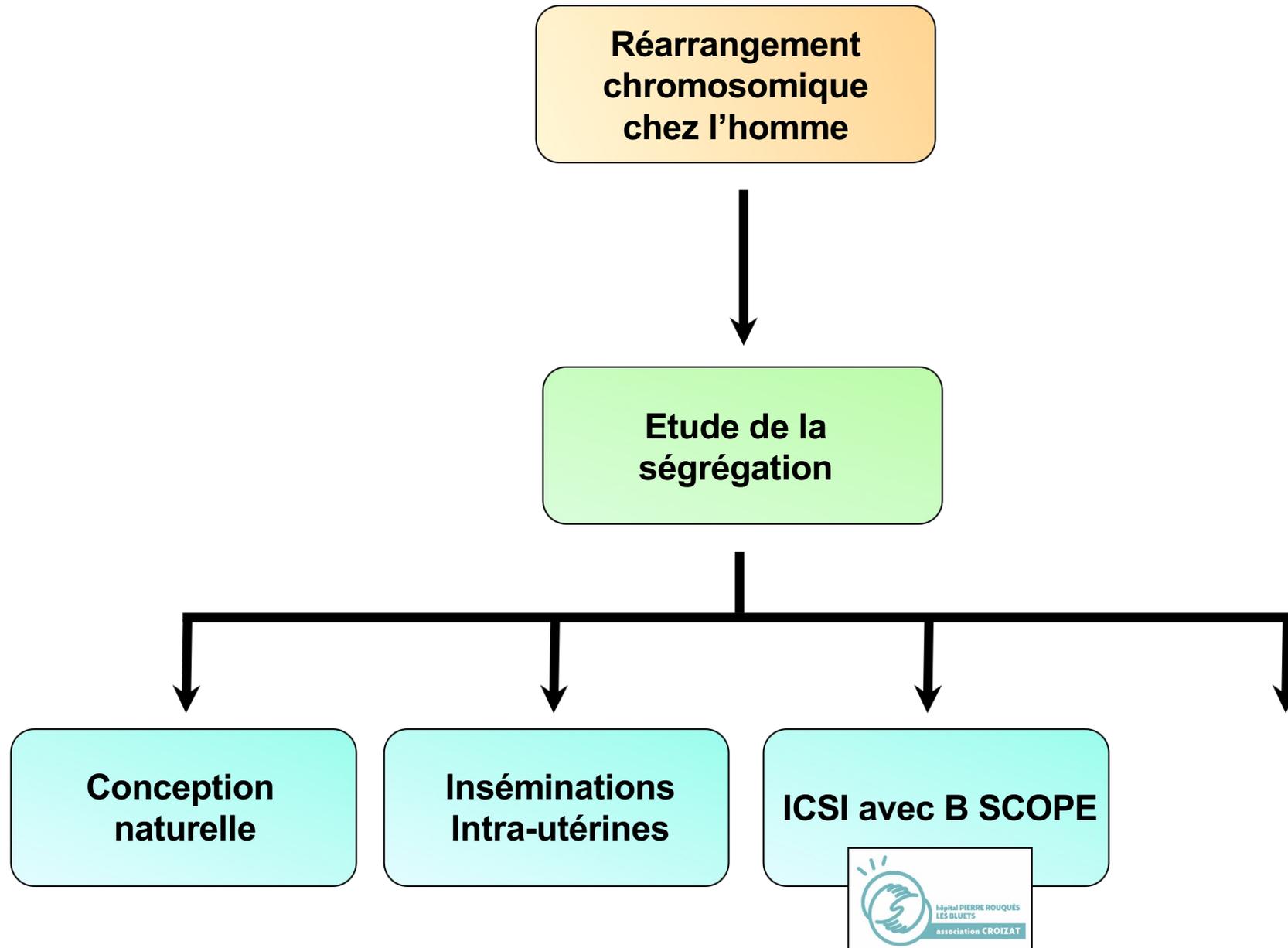


**Etude de la
ségrégation**

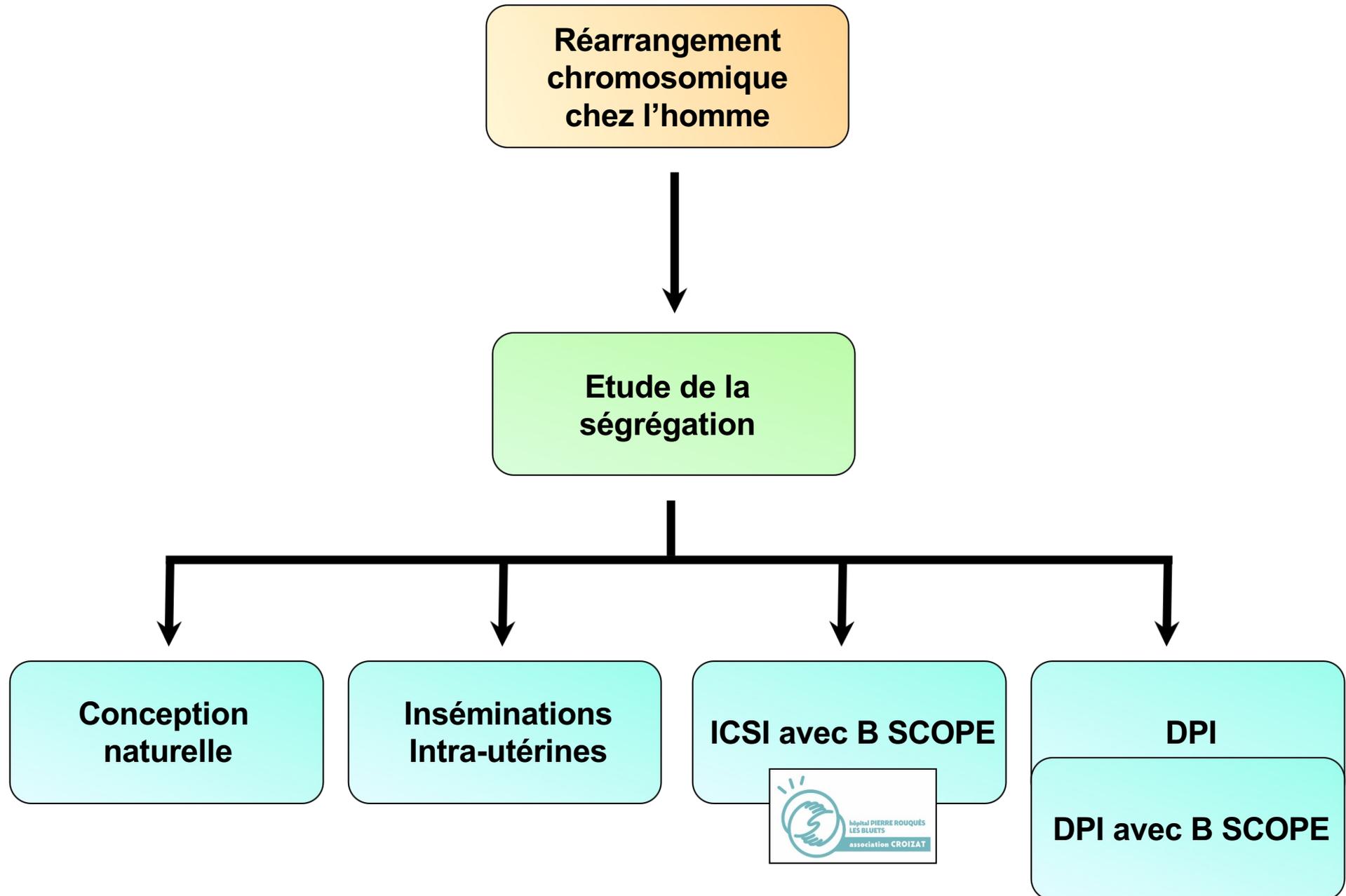
FISH sur spermatozoïdes

- % initial
- % après TMS
- % après B-SCOPE

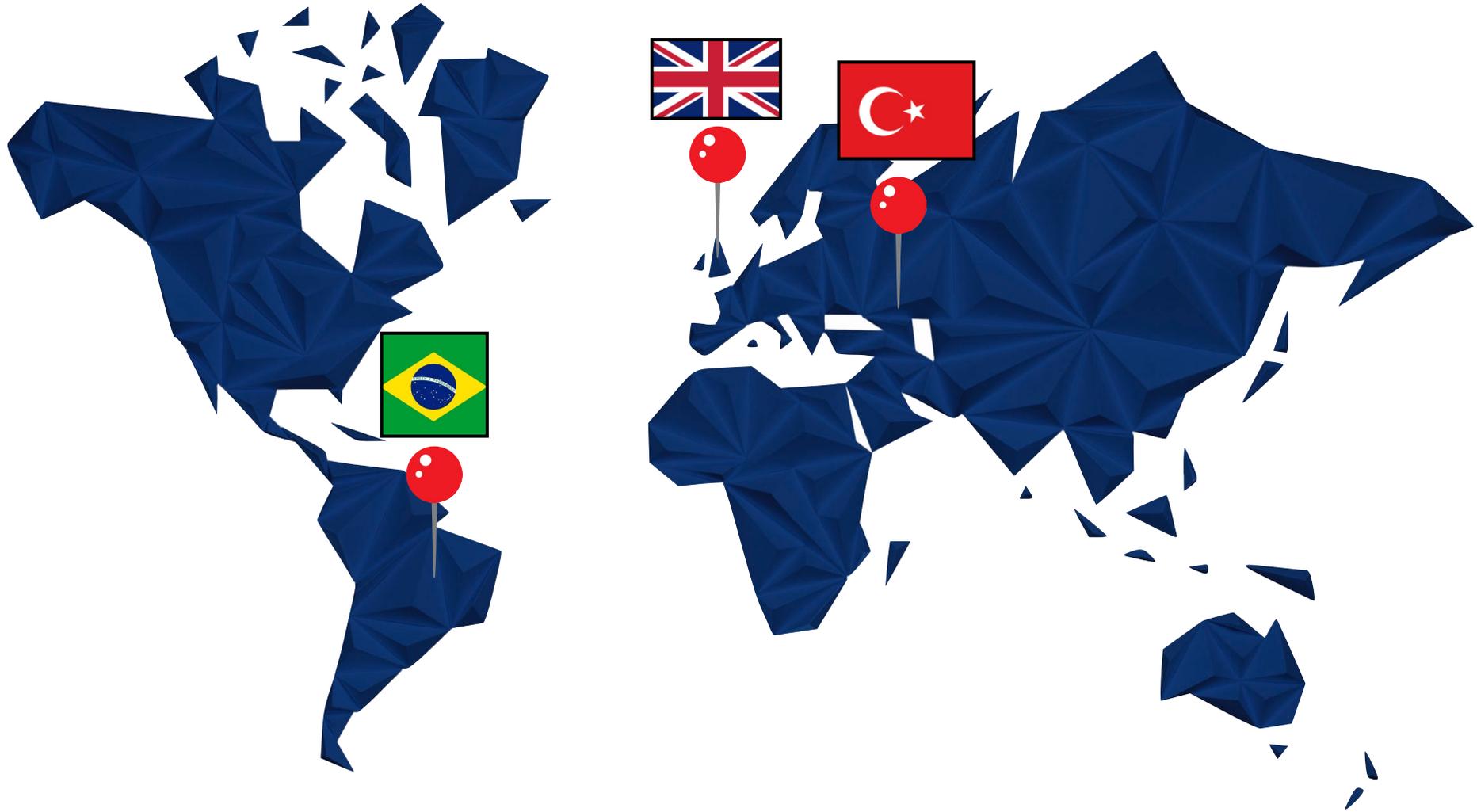
Prise en charge



Prise en charge



B SCOPE + DPI





Merci pour votre attention

Avis, prise en charge, question : alexandre.rouen@aphp.fr