Molecular diagnosis of Male Infertility

Identification of ITPR1 gene as a diagnostic marker in non-obstructive azoospermia

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Application of Molecular diagnosis in male infertility

Male fertility tests

Health Check

Semen Analysis

Hormone testing

Ultrasound

Infection Screening

Genetic Testing

Testicular Biopsy

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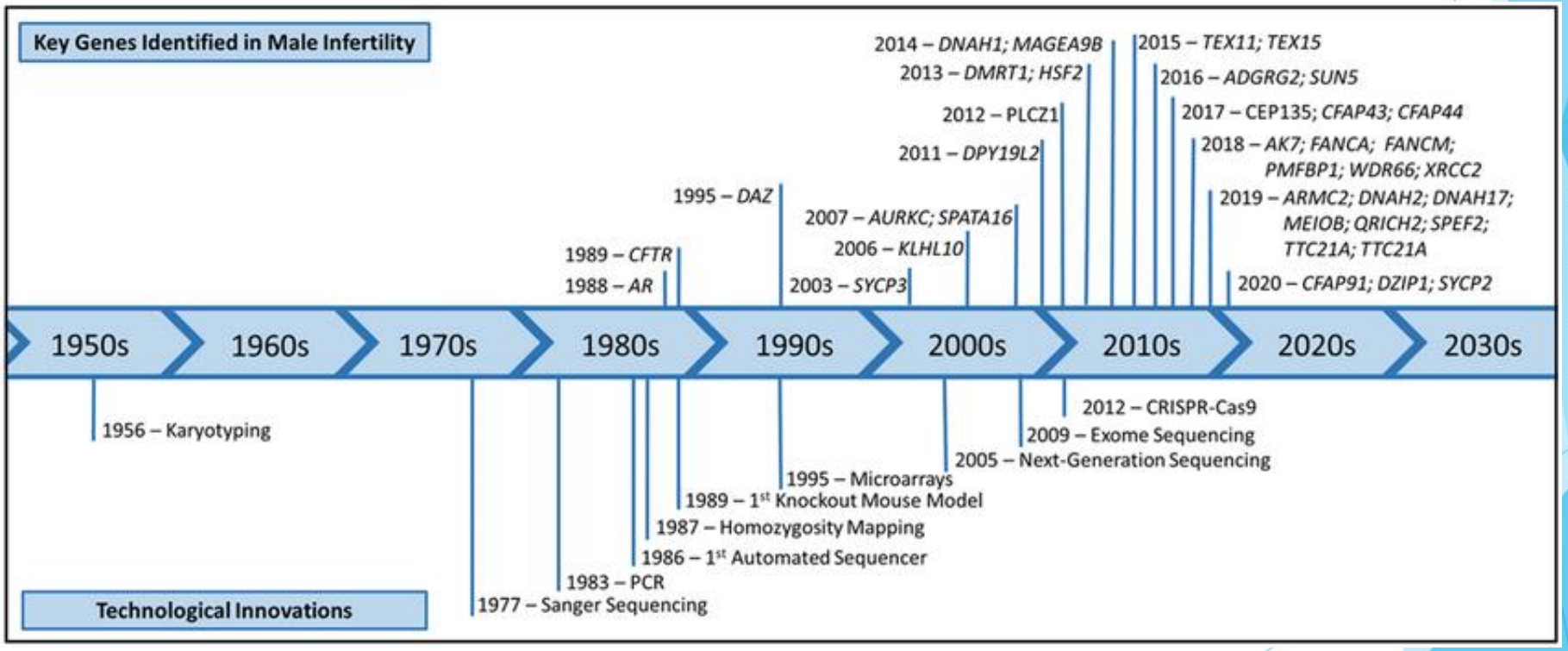
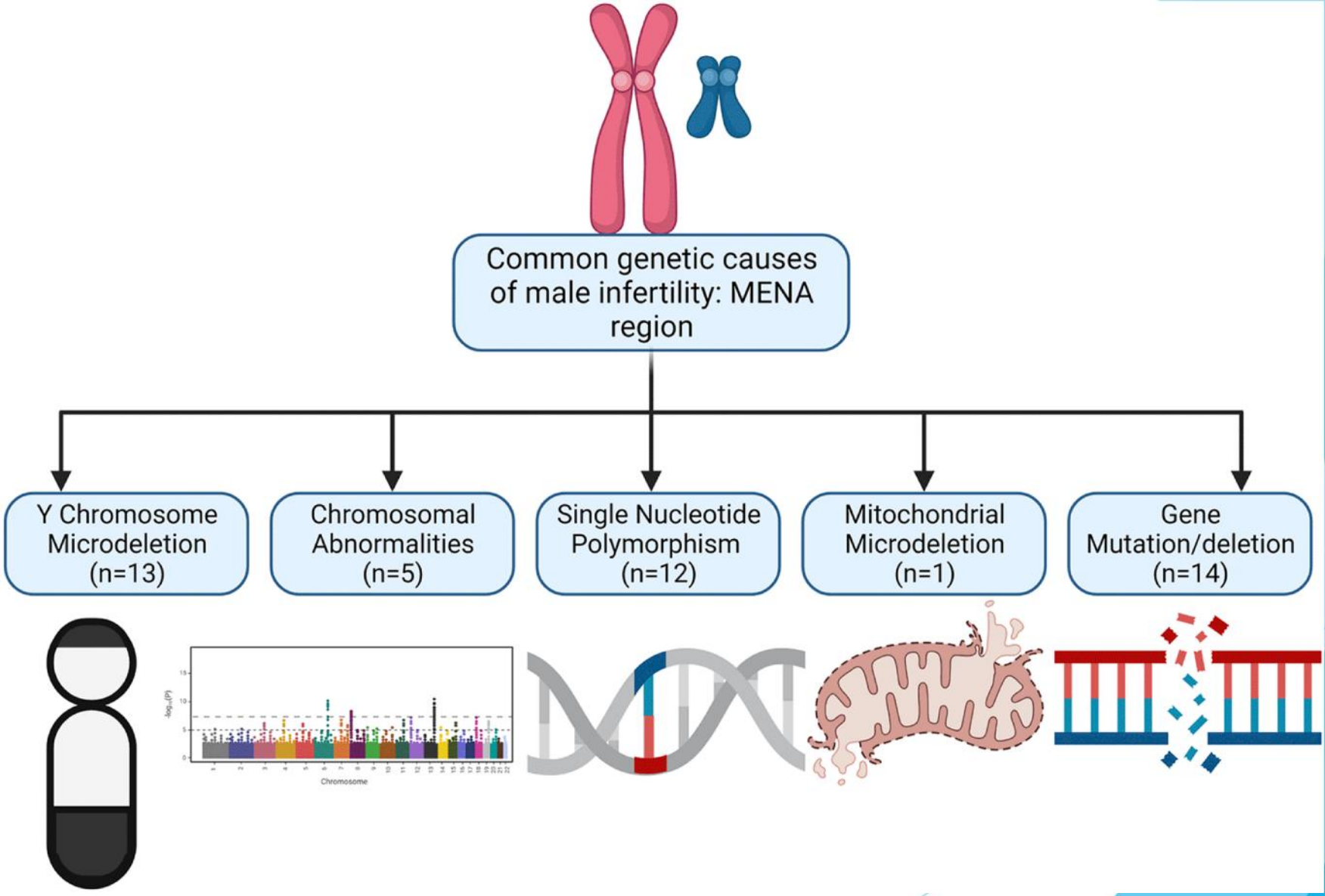
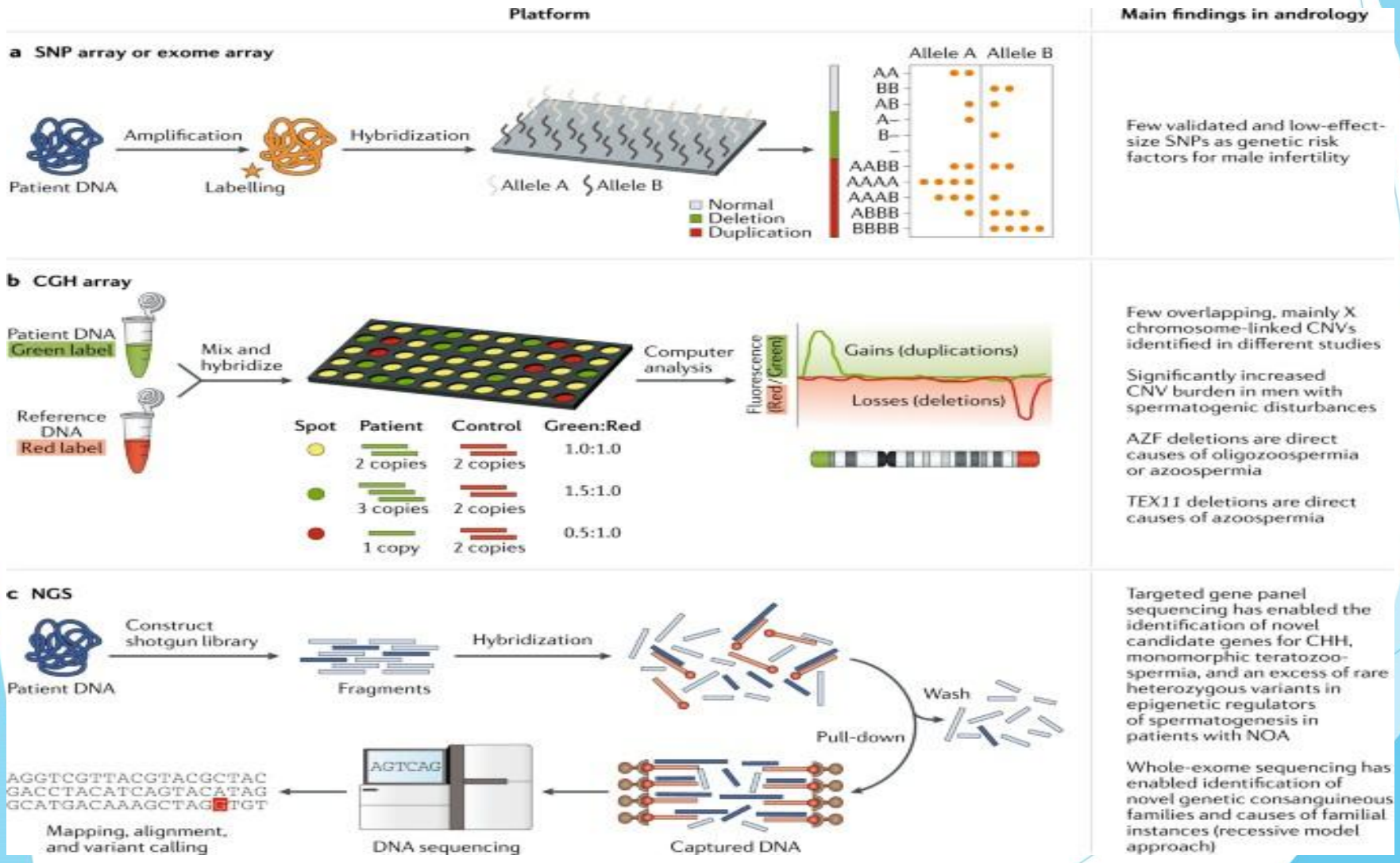
Timeline of the discovery of key genes involved in male infertility

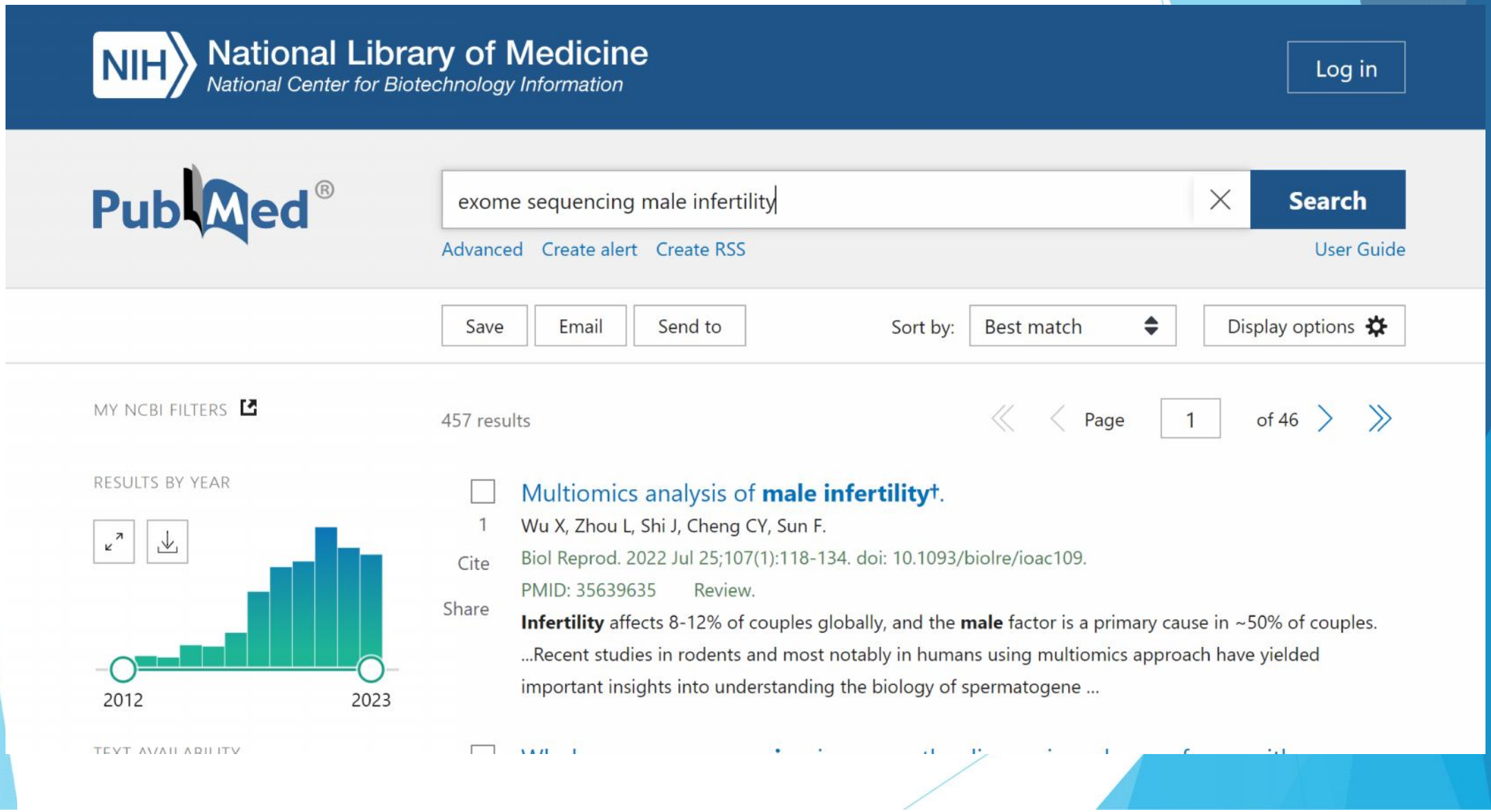



Common genetic causes of male infertility: MENA region
Y Chromosome Microdeletion (n=13)
Chromosomal Abnormalities (n=5)
Single Nucleotide Polymorphism (n=12)
Mitochondrial Microdeletion (n=1)
Gene Mutation/deletion (n=14)
Chromosome

Advanced molecular techniques in diagnosis of male infertility



Nature Reviews Urology **volume 15**, 369–384 (2018)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RESULTS BY YEAR
2012
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Multiomics analysis of male infertility†.
1
Wu X, Zhou L, Shi J, Cheng CY, Sun F.
Cite
Biol Reprod. 2022 Jul 25;107(1):118-134. doi: 10.1093/biolre/ioac109.
PMID: 35639635
Review.
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Infertility affects 8-12% of couples globally, and the male factor is a primary cause in ~50% of couples.
...Recent studies in rodents and most notably in humans using multiomics approach have yielded
important insights into understanding the biology of spermatogene ...

exome sequencing

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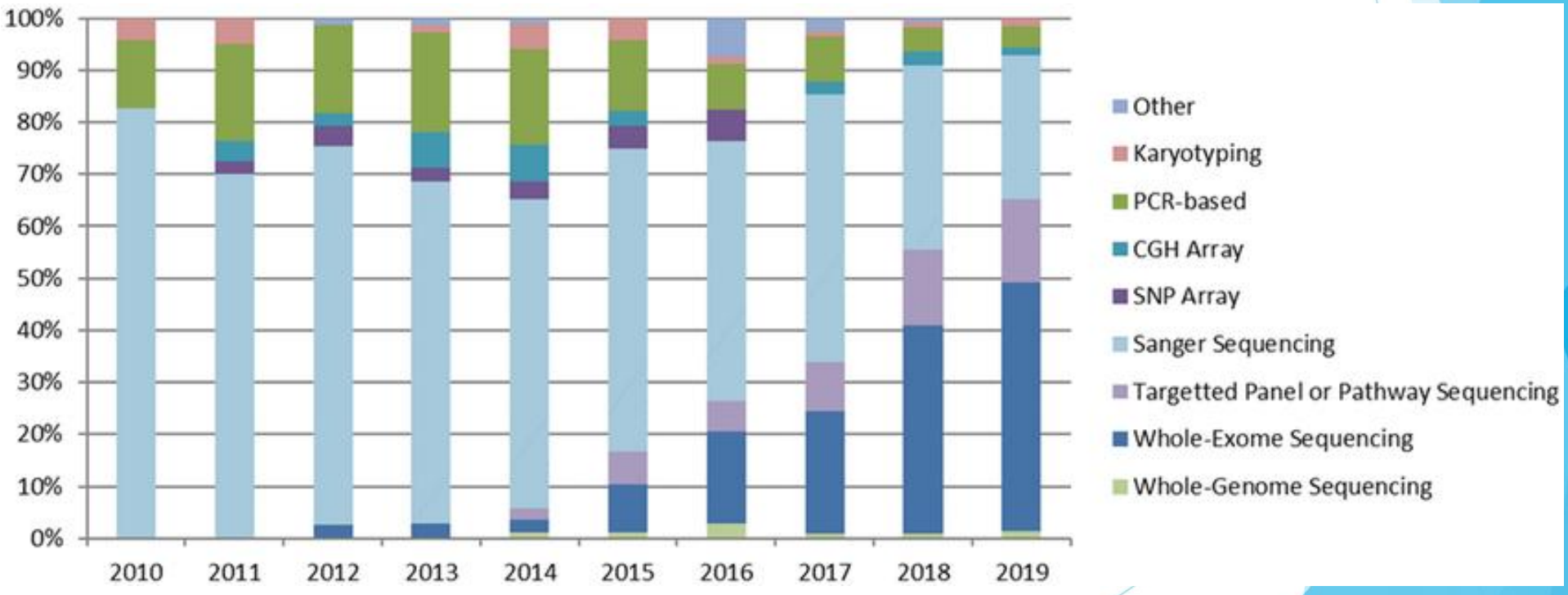
Breakdown of genetic tools used in published male infertility studies



Biology of Reproduction

SSR Society for the Study of Reproduction

Volume 107, Issue 1
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JOURNAL ARTICLE

Multiomics analysis of male infertility† FREE

Xiaolong Wu, Liwei Zhou, Jie Shi, C Yan Cheng, Fei Sun ✉

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Article Contents

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Introduction

Abstract

Infertility affects 8–12% of couples globally, and the male factor is a primary cause in ~50% of couples. Male infertility is a multifactorial reproductive disorder, which can be caused by

Table 3 Summary on the use of WES results to study male infertility in humans

Gene	Location	Phenotype	References	Gene	Location	Phenotype
PLCZ1	12p12.3	Abnormal Ca^{2+} transients and early embryonic arrest	[188]	*DNAH9*	17p12	Severe asthenozoos
CEP112	17q24.1	Acephalic spermatozoa	[121]	*DPY19L2*	12q14.2	Globozoospe
PMFBP1, TSGA10	16q22.2, 2q11.2	Acephalic spermatozoa	[115]	*SEMA5A, NEURL4, BRD2, CD1D, and CD63*	5p15.31, 17p13.1, 6p21.32, 1q23.1, 12q13.2	Idiopathic
SUN5	20q11.21	Acephalic spermatozoa syndrome	[113–117]	*CATSPERE*	1q44	IVF failure
SPATC1L	21q22.3	Acephalic spermatozoa	[118]	*HAUS7*	Xq28	IVF failure
PMFBP1	16q22.2	Acephalic spermatozoa syndrome	[119]	*ANOS1*	Xp22.31	KS
TSGA10	2q11.2	Acephalic spermatozoa	[191]	*SEMA3A*	7q21.11	KS

ZSWIM7	17p12	MA	[107]	*CFAP251*	12q24.31	Sperm immc
SYCP3	12q23.2	MA	[201]	*CFAP65*	2q35	Sperm immc
SYCE1	10q26.3	MA	[204]	*CFAP69*	7q21.13	Sperm immc
EMI1	22q13.2	MA	[105]	*CFAP47*	Xp21.1	Sperm immc
TEX15	8p12	MA	[206]	*CFAP58*	10q25.1	Sperm immc
PSMC3IP	17q21.2	MA	[207]	*CFAP70*	10q22.2	Sperm immc
STX2	12q24.33	MA	[208]	*CFAP91*	3q13.33	Sperm immc
RNF212	4p16.3	MA	[209]	*DNAAF6*	Xq22.3	Sperm immc
FKBP6	7q11.23	MA	[211]	*DNAH1*	7p15.3	Sperm immc
ATG4D	19p13.2	NOA	[212]	*DNAH2*	17p13.1	Sperm immc
FANCM	14q21.2	NOA/SCOS	[213]	*DNAH8*	6p21.2	Sperm immc

SPAG17	1p12	Asthenozoospermia	[192]	*ADAD2, TERB1, SHOC1, MSH4, and RAD21L1*	16q24.1, 16q22.1, 9q31.3, 1p31.1, 20p13	MA
ADCY10	1q24.2	Asthenozoospermia	[160]	*DMC1*	22q13.1	MA
DNAH10	12q24.31	Asthenoteratozoospermia	[144]	*M1AP*	2p13.1	MA
MEIOB, TEX14, DNAH6	16p13.3, 17q22, 2p11.2	Azoospermia	[101]	*MEIOB*	16p13.3	Meiotic arres
FBXO43	8q22.2	Azoospermia	[196, 197]	*Stag3*	7q22.1	MA (mice)
TDRD7	9q22.33	Azoospermia and congenital cataract	[98]	*TEX11*	Xq13.1	MA
CFTR	7q31.2	CAVD	[108]	*TDRD9*	14q32.33	MA
ADGRG2	Xp22.13	CBAVD	[109, 110, 112, 198, 199]	*XRCC2*	7q36.1	MA

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Male Infertility Diagnosis: Improvement of Genetic Analysis Performance by the Introduction of Pre-Diagnostic Genes in a Next-Generation Sequencing Custom-Made Panel

Vincenza Precone[1], *Rossella Cannarella*[2], *Stefano Paolacci*[3*], *Gian Maria Busetto*[4], *Tommaso Beccari*[5], *Liborio Stuppia*[6], *Gerolamo Tonini*[7], *Alessandra Zulian*[3], *Giuseppe Marceddu*[1], *Aldo E. Calogero*[2] *and Matteo Bertelli*[1,3,8]

Association of mutations with primary defects of spermatogenesis

	Gene(s)	Clinical suspect	Sperm parameters[1]	FSH serum levels (IU/ml)	Testicular volume (right and left)[2]	Testicular histology	FSH responsivenes
Subject 1	*DNAH11, DNAI1, GALNTL5*	Primary defects of spermatogenesis	Mild OAT	6.6	9.6 ml and 14.9 ml	—	No
Subject 2	*DNAH5, AMELY*	Primary defects of spermatogenesis	Azoospermia	8.0	19.5 ml and 19.9 ml	NA	NA
Subject 3	*CCDC40*	Primary defects of spermatogenesis	OAT	5.4	9.8 ml and 11.2 ml	—	NA
Subject 4	*DNAH10*	Primary defects of spermatogenesis	?	?	?	?	?
Subject 5	*KLK4*	Primary defects of spermatogenesis	Mild OAT	3.6	15.1 ml and 11.7 ml	—	NA
Subject 6	*DNAH10*	Primary defects of spermatogenesis	Mild OAT	7.3	7.5 ml and 12.6 ml	—	No
Subject 7*	*DNAH11*	Primary defects of spermatogenesis	Normozoospermia	5.7	10.9 ml and 10.7 ml	—	Yes
Subject 8	*CFTR*	Primary defects of spermatogenesis	OAT	16.3	6.3 ml and 9.8 ml	—	—
Subject 9	*CATSPER2, KLK14*	Primary defects of spermatogenesis	Azoospermia	32.7	6.7 ml and 8.7 ml	Sertoli cell only syndrome	—
Subject 10	*ADCY10*	Primary defects of spermatogenesis	OAT	3.6	10.1 ml and 12.5 ml	—	No

[1]Assessed using WHO 2010 guidelines.
[2]Evaluated by ultrasound (ml).
[3]FSH responsiveness was defined by the doubling of sperm concentration or total sperm count vs. pre-treatment values.
**The patient was diagnosed for reversal central hypogonadism. The values shown have been measured following 5 months from treatment withdrawal.*
Severe oligozoospermia was defined for total sperm count <1.0 million; mild oligozoospermia was defined for total sperm count enclosed between 1.0 and 5.0 million; oligozoospermia for total sperm count enclosed between 5.0 and 3 million (21).

Exome sequencing identifies n
Identification of ITPR1 gene as
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Identification of ITPR1 gene as a novel target for hsa-miR-34b-5p in non-obstructive azoospermia: a Ca2+/apoptosis pathway cross-talk
Bahareh Maleki, Parastoo Modarres, Peyman Salehi & Sadeq Vallian
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Questions and discussion

- **How could help WES/NGS in genetic diagnosis of male infertility?**