Y Chromosome Deletion Test by PCR

Patient ID:	Gender:		Sample Collected:
Patient Name:	Clinician Name:	NA	Sample Received:
Age :	Hospital Name :	NA	Report Released :

CLINICAL HISTORY

Advised Y Chromosome Microdeletion.

SAMPLE DESCRIPTION

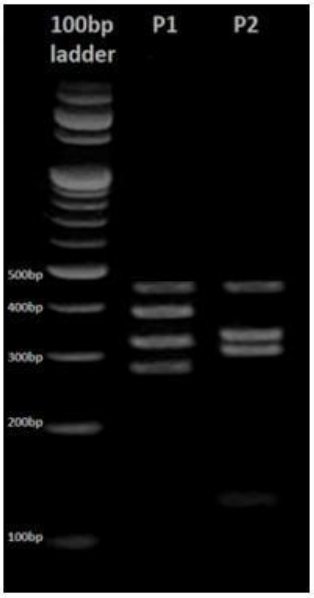
Sample quality is optimum for the test.

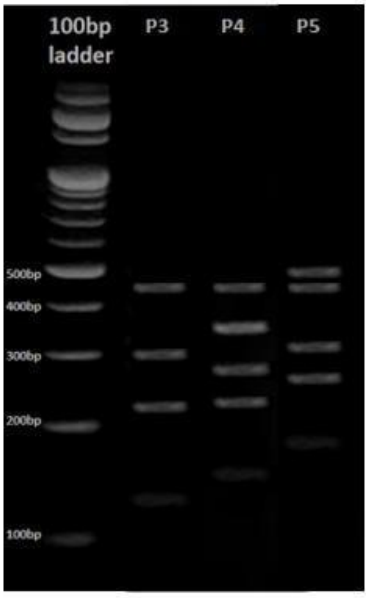
RESULT

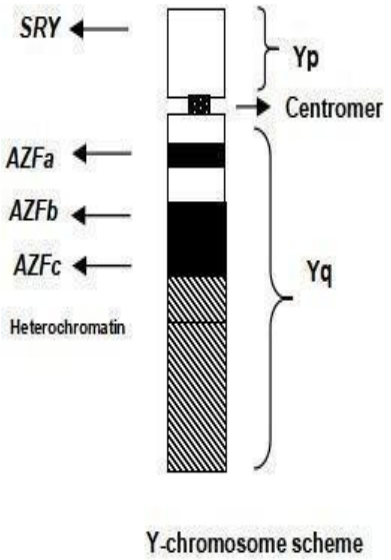
Negative

INTERPRETATION

- The patient does not have deletions in the loci tested in this module in the Y Chromosome that is attributed for Azoospermia and severe oligospermia. Therefore Y chromosome is mutation free for these loci which are mentioned in table.







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Patient Name : Patient ID: Date:

LANE	STS	Locus	Region	Results	LANE	STS	Locus	Region	Results
Marker	100 bp Ladder				Marker	100 bp Ladder			
LANE P1	sY14	SRY	Control	472 bp band detected	LANE P4	sY14	SRY	Control	472 bp band detected
	sY254	DAZ	AZFc	380 bp band detected		sY153	DYS237	AZFb	139 bp band detected
	sY86	DYS148	AZFa	318 bp band detected		sY160	DYZ2	Hetero-chromatin	236 bp band detected
	sY127	DYS218	AZFb	274 bp band detected		sY83	DYS11	AZFa	277 bp band detected
LANE P2	sY14	SRY	Control	472 bp band detected		sY1191	G73809	b2/b3	355 bp band detected
	sY84	DYS273	AZFa	326 bp band detected	LANE P5	sY14	SRY	Control	472 bp band detected
	sY134	DYS224	AZFb	301 bp band detected		sY121	DYS212	AZFb	190 bp band detected
	sY255	DAZ	AZFc	123 bp band detected		sY143	DYS231	AZFb	311 bp band detected
LANE P3	sY14	SRY	Control	472 bp band detected		sY1291	G72340	gr/gr	527 bp band detected
	sY88	DYS276	AZFa	123 bp band detected		sY82	DYS272	AZFa	264 bp band detected
	sY1065	G64724	AZFa	239 bp band detected	LANE P3	sY105	DYS201	AZFb	301 bp band detected

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PRINCIPLE AND METHODOLOGY

The second most frequent genetic factor contributing to male sterility is Y chromosome microdeletions (YCMDs). Microdeletions occur as a consequence of meiotic recombination errors in the Y chromosome's highly repetitive sequences. The AZFa, AZFb, and AZFc regions of the Y chromosome are three regions that seem especially prone to deletion. Men with YCMD usually exhibit normal phenotypes because the majority of the genes on the Y chromosome are unique to spermatogenesis. Depending on the level of spermatogenic impairment, the development of testicles may vary. The basis of PCR is the DNA polymerase's potential to create new DNA strands that are similar to the provided template strand. DNA polymerase requires a template to which it can add the first nucleotide because it can only add a nucleotide onto an already-existing 3'-OH group.This prerequisite allows the researcher to identify a particular area of the template sequence that they want to enhance. The specific sequence will accumulate in billions of copies at the end of the PCR reaction. (amplicons). Microdeletions are too small to be detected on a karyotype, so YCMD testing is usually done by polymerase chain reaction amplification of several short segments within each AZF region. PCR multiplex assays that show genomic microdeletions in the significant repetitive genomic sequence blocks on the long arm of the human Y chromosome rule out Y chromosome microdeletion. To assess AZF microdeletion, STS deletion markers within the three AZF sections of the Y gene that are known to be expressed in male germ cells are used.

LIMITATIONS

- Diagnostic errors can occur due to rare sequence variations or could be due to the genetic differences between the target and primer sequence. Mutations within individual genes included in the AZF regions will not be detected.
- This assay will not detect all of the causes of infertility or azoospermia. Therefore, the absence of a detectable microdeletion does not rule out the presence of other genetic or nongenetic factors that may be the cause of clinical findings.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.
- Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- A genetic consultation is recommended for all patients undergoing this testing. Additional consultation with a reproductive endocrinologist/urologist to discuss reproductive options is recommended when a deletion is detected.

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RECOMMENDATION

Genetic counselling is advised.

REFERENCES

- Krausz, C., et al. "EAA/EMQN best practice guidelines for molecular diagnosis of Y-chromosomal microdeletions: state-of-the-art 2013." *Andrology* 2.1 (2014): 5-19.
- Pan, Yuan, et al. "Molecular microdeletion analysis of infertile men with karyotypic Y chromosome abnormalities." *Journal of International Medical Research* 46.1 (2018): 307-315.
- Okutman, Ozlem, et al. "Genetic evaluation of patients with non-syndromic male infertility." *Journal of Assisted Reproduction and Genetics* 35 (2018): 1939-1951.
- Blanco, Patricia, et al. "Divergent outcomes of intrachromosomal recombination on the human Y chromosome: male infertility and recurrent polymorphism." *Journal of medical genetics* 37.10 (2000): 752-758.
- Chandrapal, Jason C., and James M. Hotaling. "Genetic Testing of Y-Chromosome Microdeletion." *A Practical Guide to Sperm Analysis*. CRC Press, 2017. 37-52.
- Dutta, Saurav, et al. "Prevalence of Y chromosome microdeletion in azoospermia factor subregions among infertile men from West Bengal, India." *Molecular Genetics & Genomic Medicine* 9.10 (2021): e1769.
- Al-Janabi, Anwar M., et al. "Prevalence of Y chromosome microdeletion in azoospermic infertile males of Iraqi population." *Journal of genetics* 99 (2020): 1-5.

.....End of Report.....

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