

Test Report

Name of Patient : -. SP LVP
Pt. No :
Age : -
Sex : Notspecified
Reason for Referral : N/A
Referred By : DR L V PRASAD

Collection center : OSMA0002
Date of Collection : 28-10-2022 03:05:00
Date of Receipt : 29-10-2022 12:21:03
Date of Reporting : 21-11-2022 19:13:30

Name of Test : Karyotyping
Specimen : Human Induced Pluripotent Stem Cells

Test Result

No. of Metaphases:

Counted : 20
Analysed : 20
Karyotyped : 5

Karyotype : 46,XY

Test Report

Chromosomal analysis revealed apparently normal male karyotype, the banding resolution [ISCN (2020)] of the chromosomes is 525.

Interpretation

The karyotypes are designated as per International System of Human Cytogenetic Nomenclature (ISCN, 2020) whereby the chromosomes are classified into 7 groups (A - G) consisting of 22 pairs of autosomes and one pair of sex chromosomes. Thus, a human diploid cell has 46 chromosomes. The basic steps involved in chromosome preparation from Cultured cells include long term growth of cells in tissue culture flasks in Nutrient medium and arresting of the spindle formation in cell division by colcemid. The chromosomes are in the condensed form in this phase (metaphase) of the cell cycle. The cells are then given hypotonic treatment so that they swell and the chromosomes are released. The slides are prepared and stained using G - banding method for the identification and analysis of chromosomes. The metaphases are analyzed using Cytovision software.

Note:

- 1.The report should be correlated with the history.
- 2.The results are not to be used as sole means for clinical diagnosis or cell line management decisions.
- 3.Presence of low grade mosaicism cannot be ruled out.
- 4.The microdeletion syndrome and cryptic chromosomal anomalies cannot be detected. Also the cytogenetic study cannot rule out the molecular basis of any inherited genetic disorder.

Test Indications:

Karyotyping of Cultured cells in indicated to evaluate its stability along with to detect both numeric and structural alterations, including



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balanced rearrangements and mosaicism.

Test Specifications:

Cultured cell karyotyping is performed on metaphase spreads obtained from long term cultures in tissue culture flasks in Nutrient medium and arresting of the spindle formation in cell division by colcemid. The chromosomes are in the condensed form in this phase (metaphase) of the cell cycle. The cells are then given hypotonic treatment so that they swell and the chromosomes are released. The slides are prepared and stained using G - banding method. The metaphases are analyzed on bright field microscope and karyotyped using the appropriate software.

Karyotype has the advantage of detecting numerical and structural chromosomal aberrations at a resolution of approximately 10 million DNA base-pairs, i.e. >10 mb level.

Clinical Significance:

Karyotyping of Cultured cells is done to ascertain the stability of cell line.

Limitation of the Assay:

Culture failure may be seen in cases of infected samples, samples with very low cell count and poor cell viability.

References:

1. An International System for Human Cytogenetic Nomenclature, Ed: Jean McGowan-Jordan, Ros J. Hastings, Sarah Moore - Karger Publications, USA, 2020.
2. The AGT Cytogenetics Laboratory Manual, 3rd, Ed: Barch M J, Knutsen T, and Spurbeck J L, Lippincott-Raven Publishers, New York, 1997, 19-76, 173-320, 481-526.



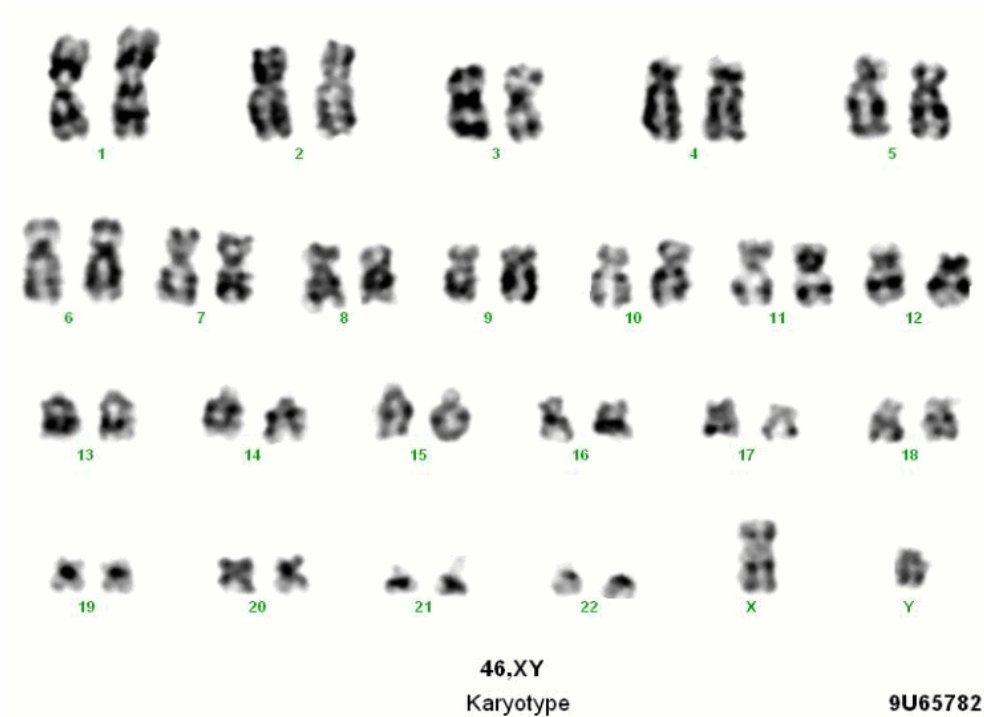
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Metaphase

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End of Report



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4. The test results are interpreted based on the quality and source of the specimen tested. Any query regarding the same needs to be addressed to the concerned technical head.
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14. PCR is a technique used to amplify the number of copies of a specific region of DNA, in order to produce enough DNA to be adequately tested. Using specific primers, mutations in β -globin gene are detected by ARMS PCR. Currently available data indicate that the technical error rate for all types of DNA analysis is approximately 1-2%. The PCR technology platform is prone to errors due to the processivity of the DNA Polymerase being used. This may lead to an occasional false positive for which Reliance Life Sciences will not be held responsible.
15. Karyotyping is a standard cytogenetic technique that analyses metaphase chromosome spreads obtained from cultured cells. All genetic abnormalities like single gene / polygenic disorders, microdeletions, subtle rearrangements and low grade mosaicism cannot be ruled out by karyotyping.
16. FISH is a molecular diagnostic tool for a rapid and precise identification of chromosomal aberrations (aneuploidies, microdeletions, translocations, etc.) using specific commercially available DNA probes. However, only probe-specific defects get identified. Very small microdeletions, point mutations or other genetic etiologies cannot be detected by FISH.
17. All clinicians involved in the care of families requesting prenatal diagnosis, and the families themselves should be aware of the data in Point No. 14, 15 and 16 before decisions about termination of pregnancy are taken.
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