

Rare & Inherited Disease Genomic Laboratory

Great Ormond Street Hospital for Children NHS Foundation Trust Levels 4-6 Barclay House, 37 Queen Square, London WC1N 3BH



Operational Lead & Head of Service (Cytogenetics): Deborah Morrogh FRCPath Head of Service (Molecular Genetics): Sam Loughlin DipRCPath Telephone: 020 7829 8870; Email: gos-tr.norththamesgenomics@nhs.net

External Requesting

CLINICIAN Ms. Vogt

Womens Health and Diagnostics

Whittington Health Magdala Avenue

London N19 5NF Patient Name: KINDLER, Fetus Of Zoe

Patient DOB: 20/11/1988
Patient Gender: Unknown
NHS Number: Missing
GOSH MRN: 3479640

Family Number: **Z2187037**

External Pat ID: 11474767 / NHS: 434 070 9255

REFERRAL REASON:

Fetus with a likely chromosomal abnormality

Clinical Details (As stated on request form): 3 early miscarriages. Missed miscarriage at 7 weeks.

MICROARRAY RESULT - NO SIGNIFICANT ABNORMALITY DETECTED

Chromosomal microarray analysis using DNA extracted from uncultured chorionic villi indicated that there was no evidence of any chromosomal imbalance likely to be causal for the loss of this pregnancy.

The array analysis result obtained was consistent with an XY (male) chromosome complement.

Reported by: Robert Howie, Pre-registration Scientist

Verified by: Sarah Chu, Clinical Scientist

Array used: Illumina Beadchip CytoSNP850K:The resolution of the microarray varies according to the clinical relevance of the chromosomal region. It is expected that the microarray analysis will detect copy number variation of 1Mb or larger.

expected that the microarray analysis will detect copy number variation of TMb or large Analysis Software used: infoQuant Fusion v7

Limitations: Microarray analysis does not detect balanced rearrangements or variations of the genetic sequence, may not detect mosaicism and may be complicated by the presence of maternal tissue.

Copy number changes identified by microarray are compared to databases of known genetic variation and to reports of known pathogenic changes. Variants which are not known to have a pathogenic effect, or do not have a high risk of pathogenic effect will not be reported.

Products of Conception specimen ID 25RG-002G0030 from Product of Conception Collected 30/12/2024 10:00 Received 2/1/2025 10:57 Authorised 29/1/2025 10:03 by Sarah CHU, BMS Priority Routine

RQ653097 Page: 1 of 1 Printed: 29/1/2025 12:01