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Quality & Innovation in Clinical Genetics

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External Quality Assessments
United Kingdom National External Quality Assessment Scheme in Clinical Cytogenetics (UK NEQAS)
Cytogenetic European Quality Assessment (CEQA)
The European Molecular Genetics Quality Network (EMQN)
Cystic Fibrosis Network (CF Network)
Control de Calidad de la Asociación Española de Diagnóstico Pre-Natal (AEDP)
Quality Control for Molecular Diagnostics (QCMD)
Fetal Medicine Foundation (FMF)
PORTUGAL
SPAIN
USA

Mr(s).
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Nucleotide Complex Ground Flr.,Lab
PO BOX 500767 Dubai

REPORT NO. 1678051 (Page 1/1)
(Version 1. Published on 18-07-2016.)

Name : Rania Ham Hammoud Baby of

Date of birth: 03-07-2016
Your record number: UHS000088968
Sample: Peripheral Blood (heparin)
Gender: Female
Collection date : 10-07-2016
Reception date : 12-07-2016
Sample ID : 416475

Clinical information : Not provided.

Chromosome analysis of stimulated cultures (blood)
Study no. 739405

Study started on 12-07-2016

Method
The analysis was performed on 25 metaphases with GTG banding. Chromosome resolution was 600 bands (ISCN 2013).

Result
47,XX,+21

Interpretation
The cytogenetics analysis revealed an abnormal female karyotype with trisomy of chromosome 21. This result is compatible with Down Syndrome.

Comment
This result is consistent with clinical diagnosis of Down syndrome. The great majority (about 95%) of Down syndrome is due to simple trisomy of chromosome 21, resulting from meiotic non disjunction errors. Clinical review by a multidisciplinary team is recommended in order to implement early clinical intervention in the patient.
As this is a sporadic event the empirical recurrence risk of trisomy 21 is estimated at 1% for future pregnancies. Prenatal diagnosis should be considered in a future gestation.
Genetic counseling for patient/parents is recommended.

Tavares

Purificação Tavares, MD, PhD
Medical Genetics Specialist
CGC Genetics Director

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Dr. Paula Rendeiro

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Dr. Paula Rendeiro

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Dr. Rosario Nogueira

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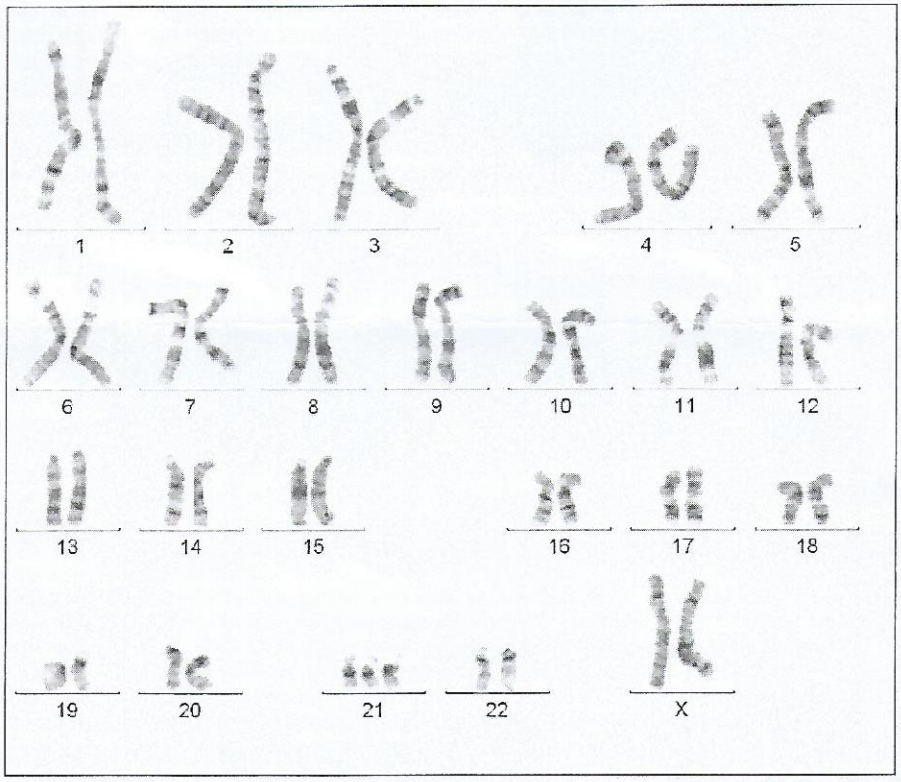

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