Greenwood Genetic Center

CYTOGENETIC LABORATORY

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Patient Sample

Study #: **14-04296FWMS**

Date of birth: **4/30/1971**

Type of Specimen: **Peripheral blood**

Requested by: **Edwin Francisco Herrera Paz, M.D.**

Test Requested: **Fluorescence in situ hybridization**

Reason for Request: **Rule out Williams syndrome, Male with distinctive facial features during infancy, Broad forehead, Medial eyebrow flare, Periorbital**

Collection Date: **3/12/2014**

Referral ID#: **N/A 0501 1971 02759**

Sample Received: **3/13/2014**

Test Request Received: **3/13/2014**

Date Reported: **3/19/2014**

Results & Interpretation

ISCN Nomenclature: ish 7q11.23(ELNx2)

No evidence of a microdeletion in the Williams syndrome critical region was detected by FISH.

Comment: Fluorescence in situ hybridization (FISH) was performed using a probe specific for the elastin gene [ELN] in the Williams Syndrome Critical Region on chromosome 7 (Vysis #32-190041). In an individual that does not have the deletion, four signals will be detected- two signals on each chromosome 7. The signal at 7q11.23 is specific for the elastin gene, while the signal at 7q36 is the D7S427 Chromosome 7 control probe which facilitates identification of the chromosome 7 homologs.

In the present study, 25 of 25 metaphase spreads and 65 of 66 interphase nuclei examined produced four signals indicating that this region is not deleted.

Reviewed and electronically signed by
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cc: Hector Miguel Ramos Zaldivar, M.S.