Next Generation Sequencing (NGS) in the prenatal diagnosis of recurrent Steel syndrome

Marta Larroya¹, Virginia Borobio¹, Antoni Borrell³, Olga Gómez¹, Montse Pauta², Maria Segura³, Alfons Nadal⁴, Eduard Gratacós¹.
¹BCNatal, Hospital Clinic, University of Barcelona, Barcelona, Spain; ²BCNatal, IDIBAPS, Barcelona, Spain; ³qGenomics. Esplugues de Llobregat, Spain; ⁴Pathology Department, Hospital Clinic, Barcelona, Spain.

Introduction

Steel syndrome (STLS) is characterized by dislocated hips and radial heads, carpal coalition, characteristic facies, short stature, scoliosis and cervical spine anomalies. It is caused by mutations in the COL27A1 gene (9q32) and it is inherited in an autosomal recessive manner.

Case report

We describe two STLS cases diagnosed in consecutive pregnancies in 2018 and 2019.

Ultrasound findings: horseshoe kidney and distal arthrogryposis. Array-CGH analysis: normal.
Termination of pregnancy at 22 weeks.

Second pregnancy while awaiting the exome results
Ultrasound findings: malposition of lower limbs. Chorionic villous sample: same previous mutations.

Conclusion

AS far as we know, these are the first prenatally diagnosed STLS cases. None of the 2 heterozygous mutations was previously described in affected cases.