Nance-Horan syndrome – Cataract 40, x-linked in a male fetus

Trivli A1, Matalliotakis Ch2, Krithinakis K2, Matalliotakis M2, Demosthenous E2

1Department of Ophthalmology, General Hospital of Agios Nikolaos, Lasithi, Crete, Greece
2Department of Obstetrics & Gynecology, Venizeleio General Hospital of Heraklion, Crete, Greece

Objective
Nance-Horan syndrome (NHS) is an extremely rare condition characterized by bilateral congenital cataract, with microphthalmia, microcornea, various forms of intellectual disability and typical dental and facial anomalies. It is inherited in a co-dominant manner with heterozygous females manifesting milder characteristics than affected males. We report a rare case of a male fetus with x linked cataract disease, caused by mutation in the NHS gene on chromosome Xp22.13, diagnosed at 22 weeks of gestation.

Methods
A 29-year-old multigravida with a spontaneous singleton pregnancy complicated with left sided congenital cystic adenomatoid malformation (CCAM) and fetal x-linked cataract disease diagnosed at the anomaly scan. No previous medical and family history. First trimester screening test: low risk for aneuploidies.

Results
Well of note, the frequency of chromosomal abnormalities and genetic syndromes is not increased in CCAM cases. There is a small risk for cardiac, renal, GI defects and fetal hydrops. In our case, fetal echocardiography indicated normal cardiac structures with right sided deviation. TORCH serum tests did not reveal any abnormality. PCR analysis of amniotic fluid did not detect the common aneuploidies, ΔF508 mutation and SMN1 gene mutation. High resolution chromosomal microarray analysis detected a male fetus with the presence of a duplication of -162 thousand bases in chromosome Xp22.33 (chr: 17,417,102_17,579,976). Parental karyotyping test indicated that the mother carried the aforesaid mutation. Genetic counseling was performed and the couple opted for termination of the pregnancy.

Conclusion
Proper genetic counseling and a collaboration between specialties such as obstetricians, ophthalmologists and other health-care professionals play a vital role in the approach of such and future pregnancies. Visual prognosis remains poor. Affected infants with cataract require early surgery with a high risk of postsurgical complications such as retinal detachment, glaucoma, corneal lesion and optic atrophy.