Isolated microgastria as a single sonographic finding representing CHARGE syndrome.

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Case Description

- 30 year old patient G2P1 AT 27 weeks
- Referred for genetic consultation due to an impression of a “small stomach” on routine anatomy scan.
- Couple:
  - Caucasian
  - Non consanguineous
  - Healthy
- Family history positive for 2 first degree relatives with non investigated developmental delay
- Pregnancy testing otherwise unremarkable.

Genetic testing

- Fetal DNA obtained from cultured amniotic fluid.
- Sequence analysis using the Blueprint Genetics (BpG) Comprehensive Hearing Loss (HL) and Deafness Panel identified a heterozygous CHD7 c. 4644+2T>A variant affecting the consensus donor splice site in intron 20 and a heterozygous nonsense variant OTOGL c.2773C>T, p.(Arg925*).

Discussion

- OTOGL is an autosomal recessive gene and in an heterozygous state probably is not the cause of the patient Hearing loss.
- CHD7 is the disease causing gene of autosomal dominant CHARGE syndrome in which Hearing loss is a major feature.

Conclusion

- Isolated Microgastria or Tracheo-esophageal fistulas are relatively common US findings.
- Rarely, these findings may represent an uncommon presentation of a genetic syndrome.
- We conclude that even in isolated cases of abnormal US findings a thorough genetic, as directed mutation panels or whole exam sequencing is beneficial for diagnosis.