

Part 2 – Case Management



The patient was referred to our local fetal treatment center, where she underwent consultation via our fetal surgeon, adult endocrinology, as well as pediatric endocrinology. She was advised to immediately discontinue her GTA-Forte and iodine and she was converted to levothyroxine.

She underwent percutaneous umbilical sampling and amniocentesis which demonstrated findings consistent with fetal hypothyroidism with secondary goiter from excessive maternal iodine intake (of note, fetal TSH was 94). Amniotic fluid sampling demonstrated normal single nucleotide polymorphism (**SNP**)-microarray and karyotype.



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Pros and cons of fetal therapy were then discussed with the patient and she decided to proceed with treatment. Intraamniotic levothyroxine 500mcg was injected at 22 weeks, 24 weeks, and 26 weeks. (Next slide – Image 3 shows Fetal thyroid gland size at 23 weeks) Percutaneous umbilical sampling was performed at the 24-week mark to assess fetal thyroid function; by then, the thyroid function tests had normalized. Subsequent ultrasounds at 27 and 30 weeks demonstrated a significant relative decrease in the fetal goiter size. (Image 4)

Another percutaneous umbilical sampling was done at 30 weeks and fetal thyroid function remained within normal limits (her prior levothyroxine injection had been given at 26 weeks). Good fetal growth and normalization of amniotic fluid was noted at subsequent ultrasound as well. At the time of this case writing, the patient was considering evaluation via fetal MRI to investigate the fetal CNS system.



Image 3 – Fetal thyroid gland size at 23 weeks (1-week post first injection of Levothyroxine treatment)





Ultrasound images courtesy of:

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Image 4 – Fetal thyroid gland size at 27 weeks (5 weeks post initiation of Levothyroxine treatment)





Ultrasound images courtesy of:

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Summary & Review of Literature



Fetal goiter is a rare condition, affecting only 1 in every 30 to 50,000 fetuses (Daneman & Howard, 1980). It can often be identified on ultrasound via a bilobed mass in the fetal mid anterior neck that is homogenous in nature. An enlarged size can lead to hyperextension of the neck and polyhydramnios may be noted as well. Sometimes there can be increased vascularity to the mass as well. A large goiter in the fetus can be often related to hypo or hyperthyroidism, or even in some cases, euthyroidism. Various etiologies include tumors, transplacental passage of maternal antibodies, intrinsic thyroid dysfunction, mutations on the thyroid stimulating hormone receptor, or iodine excess or deficiency. (Lazarus, 2000).

Early onset congenital hypothyroidism may be associated with various neurological and developmental sequelae, including mental retardation, deaf-mutism, spasticity, and ataxia. Treatment in either the fetal period or postpartum period can help curtail these effects, however, in utero effects may have already occurred prior to the time of diagnosis, especially if the hypothyroidism occurred in early gestation (de Escobar, Obregon, & del Rey, 2004; Zoeller & Rovet, 2004).



Summary & Review of Literature



In utero diagnosis via percutaneous umbilical sampling (PUBS) and injection of intra-amniotic levothyroxine has been successfully reported in the literature as a treatment for congenital hypothyroidism (Davidson, Richards, Schatz, & Fisher, 1991). Dose recommendations include a goal of 10ucg/kg /day of levothyroxine (Abuhamad et al., 1995)

lodine crosses the placenta via active transport; the recommended daily allowance of iodine intake is 200 ug daily. (Institute of Medicine, 2001) In this particular case, the patient was taking 12.5mg of iodine daily – this equates to more than 60 times higher than the recommended daily allowance. Excess iodine intake can actually inhibit thyroid hormone secretion and thyroid synthesis; this is known as the Wolf-Chaikoff effect. As in this case, careful review of nutritional supplementation is very important: in cases where excess iodine is suspected to be contributory to the fetal goiter, it should be stopped immediately.

Fetal goiters that are large in size can be potentially dangerous. They may potentially cause hyperextension of the neck and may compress the trachea, leading to airway compromise at delivery. If a large mass such as a goiter is noted, it may be advisable to assemble a multidisciplinary team including anesthesiology, perinatology, neonatology, and pediatric otolaryngology. Ex utero intrapartum treatment (EXIT) procedure may be considered in cases of suspected airway compromise (Dighe, Peterson, Dubinsky, Perkins, & Cheng, 2011).

