The Neck

Normal Anatomy of the Neck

Current ultrasound equipment allows the visualization of the main anatomic structures of the neck: upper airways and esophagus,[1,2] vessels,[2] and spine.

A detailed investigation of the cervical structures is usually possible as early as the 18th week of gestation. However, flexion of the head and an unfavorable position of the fetus, can make the examination difficult. A midsagittal scan of the fetal neck at 25 weeks of gestation is provided in Figure 3-1. Most anatomic components of this area are demonstrated in this view. The tongue is seen filling the oral cavity. Shadowing from the facial bones

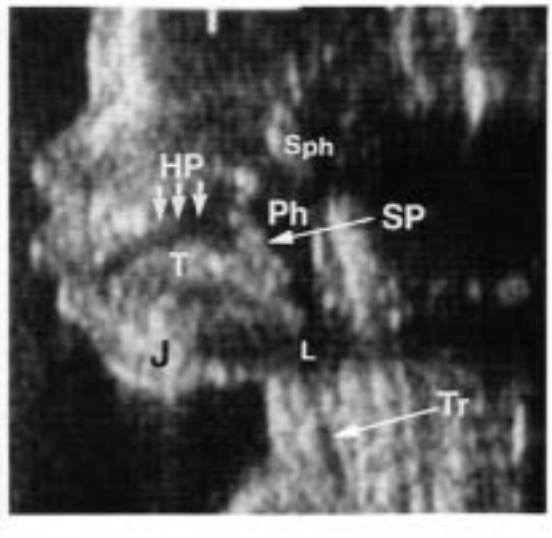

Figure 3-1. Midsagittal scan of the head of a 25-week fetus. The tongue (T) is seen filling the oral cavity. The pharynx (Ph) is delimited anteriorly and superiorly by the soft palate (SP) and the sphenoid (Sph), respectively, and it can be traced inferiorly to the larynx (L), which is partly obscured by acoustic shadowing arising from the jaw (J). The trachea (Tr) is seen in the midportion of the neck. HP, hard palate.

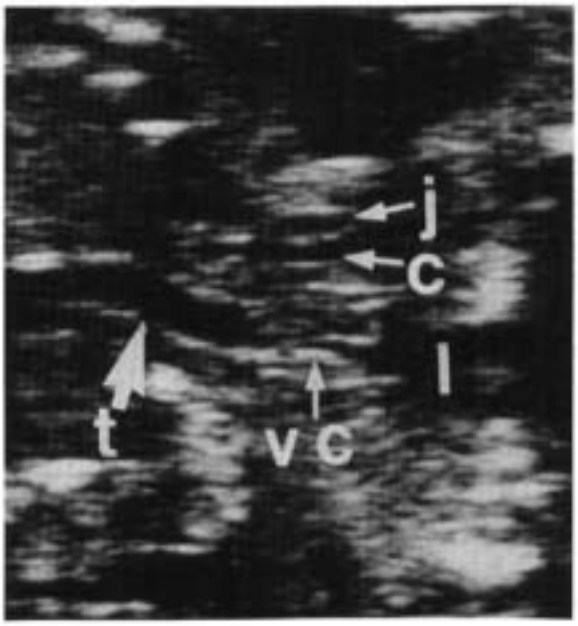

Figure 3-2. A slightly angled sagittal scan of the fetal neck, revealing the continuity between the larynx (l) and trachea (t). The common carotid artery (c) and jugular vein (j) are also demonstrated. At the level of the larynx, the vocal cords (vc) are presumably seen.

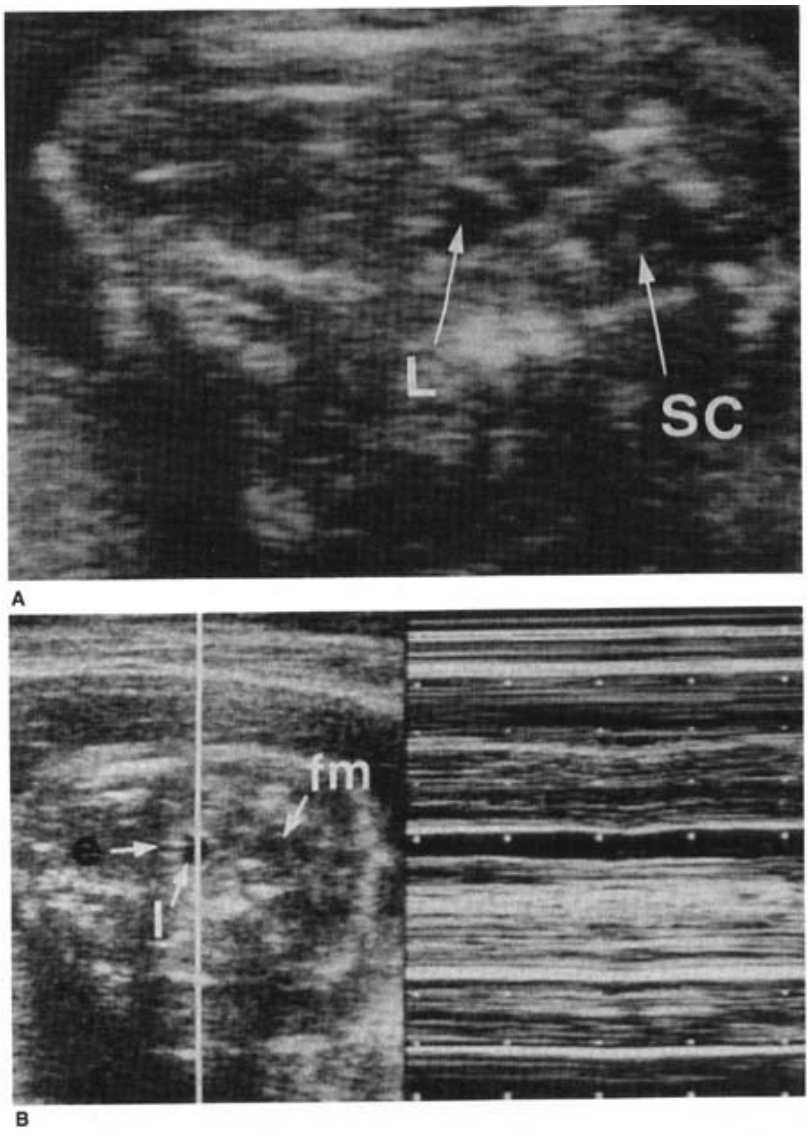

Figure 3-3. A. In this axial scan slightly caudal to the skull base, the larynx (L) is seen in the midportion of the neck, anterior to the cervical spine. SC, spinal cord. **B** Simultaneous real-time, M-mode representation of a contraction movement of the vestibulum of the larynx (l). The epiglottis (e) is seen anteriorly to the larynx. fm,foramen magnum.

obscures the hard palate, but its position can be inferred by the curvature of the tongue and by the soft palate. Real-time examination greatly enhances the understanding of these structures, since movement of both the tongue and the soft palate is frequently seen. Behind the soft palate and below a high-level echo complex presumably arising from the sphenoid, the fluid-filled pharyngeal cavity is seen and can be followed until it bifurcates into the larynx and hypopharynx. The esophagus is not usually seen.

With slight angulation of the transducer, the main neck vessels, the common carotid artery, and the jugular veins are readily demonstrated. At closer inspection, minute details, such the vocal cords, may be appreciated (Fig. 3-2).

Sagittal scans directed from posterior to anterior allow one to evaluate the soft tissues overlying the spine (see Fig. 1-23).

In axial planes, the previously described structures can be recognized as well. On real-time ultrasound examination, it is frequently possible to demonstrate the contractions of the vestibulum of the larynx (Fig. 3-3).

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Cystic Hygroma

Synonyms
Lymphangioma and jugular lymphatic obstructive sequence.

Definition
The term "hygroma" means moist tumor. Cystic hygromas are anomalies of the lymphatic system characterized by single or multiple cysts within the soft tissues, usually involving the neck.

Incidence
Cystic hygromas were found in 1 in 200 spontaneously aborted fetuses with crown-rump length greater than 3 cm.[2]

Etiopathogenesis
Cystic hygromas are frequently found in association with chromosomal aberrations (mainly Turner's syndrome). Table 3-1 shows other syndromes or conditions reported in association with cystic hygromas. When isolated, this anomaly can be inherited as an autosomal recessive trait.[5] Webbed necks or redundant skin are found in genetic and nongenetic syndromes, such as Noonan's syndrome, familiar pterygium colli, and fetal alcohol syndrome.
In the embryo, the lymphatic system drains into the jugular lymphatic sac. A communication between this primitive structure and the jugular vein is formed at 40 days of gestation (conceptional age). Failure of development of this communication results in lymphatic stasis. Dilatation of the lymphatic channels leads to the clinical manifestations of the jugular lymphatic obstructive sequence[l7] (Figs. 3-4, 3-5).
Dilatation of the jugular lymphatic sac leads to the formation of cystic structures in the cervical region. If a connection between the lymphatic and the venous system does not occur at this point, a progressive peripheral lymphedema and nonimmune hydrops will develop, leading to early intrauterine death (Figs. 3-6, 3-7).[3] If the connection is formed, the sequence is interrupted, and the fluid collections are resorbed. The redundant skin will give rise to webbed neck (pterygium colli), which is a typical manifestation of Turner's syndrome and of manv other genetic and nongenetic conditions. Uplifting and anterior rotation of the ears and an abnormal hair pattern are other consequences of overdistention of the jugular lymphatic sac. Distention of the tributary lymphatics may result in peripheral lymphedema, which in turn may give rise to redundancy of the skin of the face and puffy hands and feet, with deep-set narrow nails (Fig. 3-5). It has also been suggested that transitory ascites may result in laxity of the anterior abdominal wall and prune-belly syndrome.

TABLE 3–1. KARYOTYPE IN 60 CASES OF FETAL CYSTIC HYGROMA

Karyotype	No. of Cases (%)
Abnormal karyotype	
Turner's syndrome	
45 XO	30 (50)
Mosaic	1 (1.6)
Trisomy 21	4 (6.6)
Trisomy 18	3 (5)
Trisomy 13	2 (3.3)
47 XXY	1 (1.6)
Total	41 (68)
Normal karyotype	11 (18)
Karyotype not available	8 (13)

Data derived from Bluth et al.: South Med J 77:1335, 1984; Chervenak et al.: N Engl J Med 309:822, 1983; Garden et al.: Am J Obstet Gynecol 154:221, 1986; Greenberg et al.: Clin Genet 24:389, 1983; Pearce et al.: Prenat Diagn 4:371, 1984; Redford et al.: Prenat Diagn 4:327, 1984; and from cases collected by the authors.

Pathology
Overdistention of the jugular lymphatic sacs that are located in both sides of the neck results in the formation of a cystic structure that is usuallv partitioned by a thick fibrous band corresponding to the nuchal ligament. Within the cystic structure, thinner septa are seen and are thought to derive from either fibrous structures of the neck or deposits of fibrin.[4] The size of the lesions may vary greatly from small collections of fluid to enormous cysts that may be larger than the fetus. In cases of generalized hydrops, pleural effusions, ascites, and severe skin edema are present.

Associated Anomalies
Cystic hvgromas are very frequently associated with chromosomal aberrations (Table 3-1) and, consequently, with a wide variety of anatomic defects.

Diagnosis
The diagnosis of cystic hygroma relies on the demonstration of cystic structures usually located in the occipitocervical region.[1,3,4,7-1-5] These lesions have a typical honeycomb appearance due to the presence of multiple septa. Large lesions are usually characterized by a thick septum dividing the cyst along the

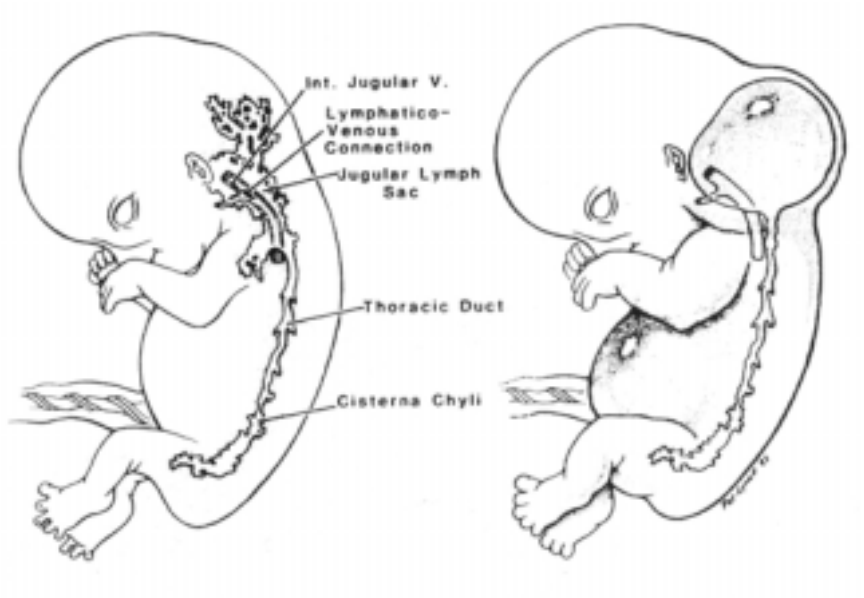

Figure 3-4. Left. Normal lymphatic system in the fetus. Right. Cystic hygroma has developed as a consequence of lymphatic obstruction. (Re-*produced with permission from Chervenak et al.: N Engl J Med 309.-822, 1983.)*

anteroposterior axis, the monographic counterpart of the nuchal ligament[4] (Figs. 3-8, 3-9).

A careful evaluation of the fetal anatomy is indicated for identification of other anatomic abnormalities, as well as signs of nonimmune hydrops. Most patients have decreased amounts of amniotic fluid, and a few have either normal fluid or polyhydramnios.

The diagnosis of cystic hygroma has been made in the first trimester.[9] The differential diagnosis includes cervical meningocele (Fig. 3-10), cephaloceles,. neck tumors, and subcutaneous edema. The distinction is often difficult.[7,11] A useful diagnostic hint is the typical multiseptate appearance of cystic hygromas. With a cephalocele, it is usually possible to demonstrate a bony defect in the skull.[13] The presence of hydrocephalus increases the index of suspicion for a cephalocele. Hydrops and generalized edema are more frequently associated with cystic hygromas. Amniotic fluid alpha-fetoprotein may be either elevated or normal in both conditions.[13] Neck tumors usually have a complex appearance.

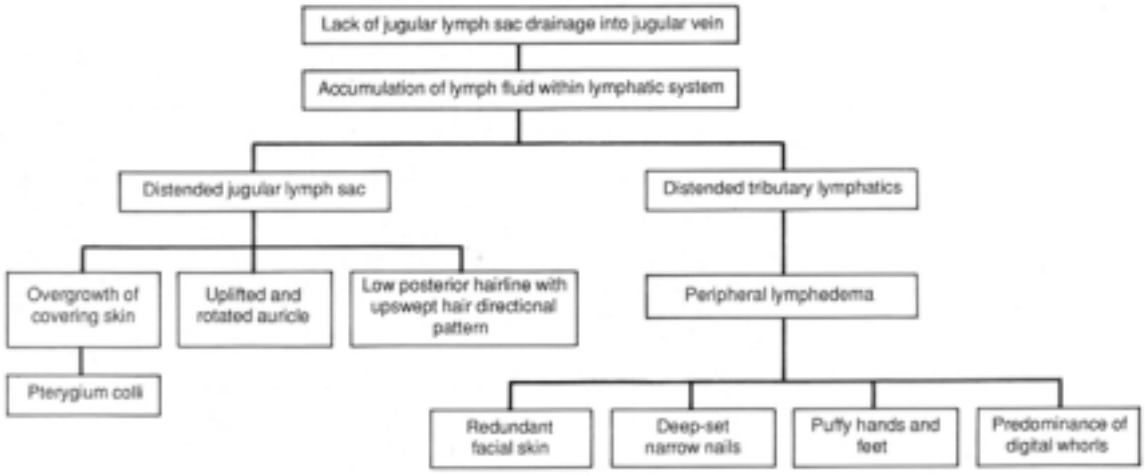

Figure 3-5. Lymphatic obstructive sequence. *(Reproduced with permission from Smith: Recognizable Patterns of Human Malformation: Genetic Embryologic and Clinical Aspects, 3rd ed. Philadelphia, Saunders, 1982, p 472.)*

Figure 3-6. Midtrimester fetus with jugular lymphatic obstructive sequence. Note the presence of ascites, skin edema, and cystic hygromas.

Figure 3-7. Closer view of the fetus shown in Figure 3-6. Cystic hygromas are seen as massive paracervical bilateral masses.

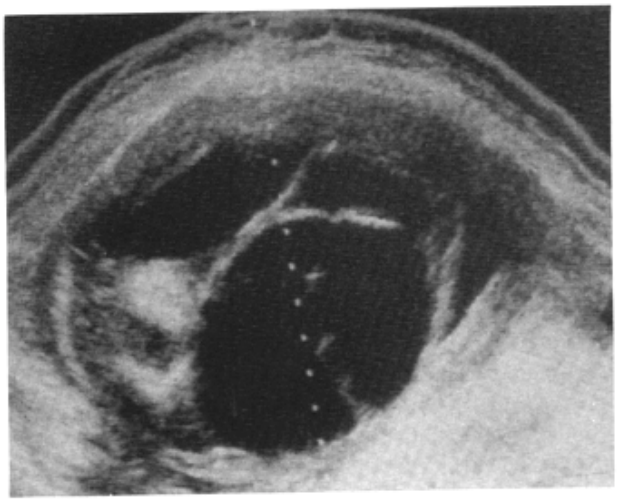

Figure 3-8. Cystic hygroma. Transverse section of the fetal neck. Typical multiseptate appearance.

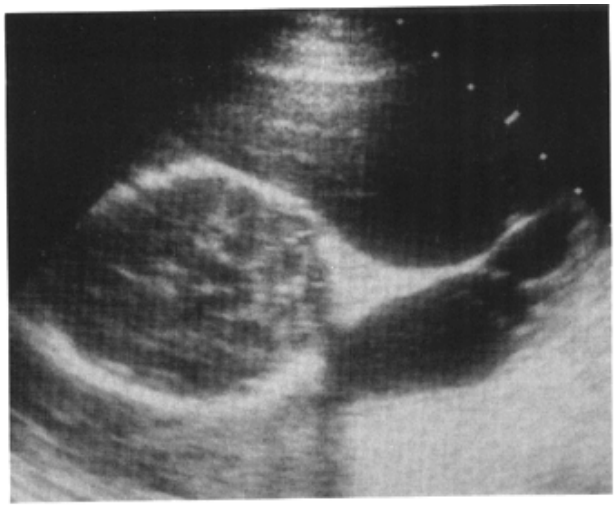

Figure 3-9. Cystic hygroma. Note the presence of septae and an intact skull. This section is an axial scan at the level of the BPD.

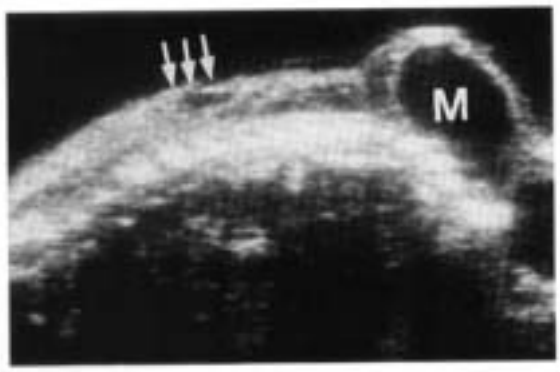

Figure 3-10. Cervical meningocele (M) may mimic a cystic hygroma. A spinal defect extending to the thoracic area is visible *(arrows).* Note the absence of septae in the cystic mass.

Prognosis

The prognosis is different depending on the presence or absence of associated hydrops. In our experience, the mortality rate of cystic hygromas with hydrops is 100 percent. In many cases, intrauterine demise occurs within the first two trimesters of pregnancy. In a review of 40 cases reported in three series in the literature,[3,7,10] most had hydrops. Thirteen (33 percent) died in utero usually within a few weeks of the diagnosis, two (5 percent) died in the early neonatal period, and the rest were electively aborted.

Prognostic data about fetal cystic hygromas without associated hydrops are scanty. Of a total of 41 infants with cystic hygromas of the head and neck reported in two different series,[6,16] 37 (90 percent) required operations. Sixteen (31 percent) developed symptoms of airway obstruction or swallowing difficulties. Facial nerve palsy as a consequence of the operation occurred in 10 (24 percent). In some infants with cystic hygromas, mandibular maldevelopment, problems of malocclusion, and tooth eruption occurred. An early partial glossectomy may be required to allow normal speech development.

The natural history of fetal cystic hygromas has not been clearly established. Some infants diagnosed in the second trimester with isolated hygromas may develop nonimmune hydrops as gestation progresses. Others may undergo canalization of the lymphatic channels and resolution of the hygromas. Regression results in redundant skin in the cervical region and webbed neck.

Obstetrical Management

Determination of fetal karyotype is recommended in all cases. This information is of diagnostic value for the index pregnancy and useful in the counseling of future pregnancies. The option of pregnancy termination should be offered before viability. After viability, fetuses with associated hydrops should probably be managed nonaggressively because of the extremely poor prognosis. In the presence of isolated cystic hygromas, no modification of standard obstetrical management is required. On some occasions, cystic hygromas have been associated with prolongation of the second stage of labor.[6] A cesarean section may be indicated if there are gigantic lesions. Infants must be delivered in a tertiary care center, because there is a high frequency of airway obstruction.[16]

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Goiter

Synonym Thyromegaly.

Definition
Goiter is an enlargement of the thyroid gland.

Incidence
Rare.

Etiology
Goiter can be associated with hyperthyroidism, hypothyroidism, or a euthyroid state. In the newborn period, goiter is most commonly associated with hypothyroidism.

Maternal Grave's disease may result in neonatal thyrotoxicosis and goiter. Two varieties of this condition are recognized. The self-limited type is thought to result from transplacental passage of a thyroid-stimulating substance, such as long-acting thyroid sfimulant (LATS). The second variety appears to be transmitted with an autosomal dominant pattern, and has a predilection for females and a protracted clinical course.[6]

Goiters associated with hypothyroidism can be the result of iodine intoxication, iodine deficiency, or congenital metabolic disorders of thyroid synthesis. Congenital hypothyroidism occurs in 1 in 3600 to 4000 live births.[5] Seventy-four percent of hypothyroid infants have primary thyroid dysgenesis (absent or hypoplastic thyroid gland), 13 percent have thyroid dyshormonogenesis, 3 to 4 percent have secondary (pituitary) or tertiary (hypothalamic) hypothyroidism, and 10 percent have hypothyroidism secondary to intrauterine exposure to antithyroid medications. Of these causes, only thyroid dyshormonogenesis and drug-induced hypothyroidism are associated with goiter in the newborn period. The two drugs primarily responsible for the latter are iodide preparations and propylthiouracil (PTU). Iodide preparations are administered as expectorant medications and radiopaque dyes are used in amniography.[9] Maternal ingestion of as little as 12 mg/day may result in congenital hypothyroidism.[3] The ratio of mothers exposed to PTU to hypothyroid infants is 100:1.[2] These drugs cross the placenta readily and block thyroid synthesis in the fetal gland. Goiter is a rare finding in drug-induced fetal hypothyroidism.

Iodine deficiency is a cause of endemic hypothyroidism in certain parts of the world. Newborns frecuently have goiters. An enzymatic deficiency impairing the synthesis of T4 or T3 can result in hypothyroidism with goiter, goiter with euthyroidism ,or hypothyroidism without goiter. Six different enzymatic defects have been recognized. They are transmitted as an autosomal recessive trait and, therefore, have a 25 percent recurrence rate. The reader is referred to specialized texts for a full description of these defects.[4] Although some of these enzymatic defects can occur with congenital goiter, in most cases, enlargement of the thyroid gland appears later in life.

Diagnosis
The diagnosis is based on identification of a neck mass that is solid, anterior, and symmetrical and may result in hyperextension of the fetal head.[1,10] Obstruction of the esophagus may lead to polyhydramnios. The differential diagnosis includes cystic hygroma, bronchial cleft cysts, cervical meningocele, and hemangiomas of the neck. Cystic hygromas are purely cystic lesions with a typical honeycomb appearance. Branchial cleft cysts are purely cystic masses located on the anterior border of the sternomastoid muscle. Hemangiomas can appear as cystic or solid masses. Teratomas are generally large tumors that have both solid and cystic elements. A prenatal diagnosis of hypothyroidism was reported recently when a high thyroid-stimulating hormone (TSH) concentration in amniotic fluid was determined in a fetus with a goiter[7,8] (Fig. 3-11). This technique requires concentration of amniotic fluid. A concentration of TSH above 0.8 µU/ml in the second trimester or 0.4 µU/ml in the third trimester are suggestive of hypothyroidism.[8]

Prognosis
Goiter may cause dystocia by extending the fetal head during the normal course of labor. It can also lead to acute respiratory failure if the enlarged gland obstructs the airways. The prognosis depends on the basic cause of the goiter. Neonatal hyperthyroidism is transient in most cases, but hypothyroidism requires treatment. In both instances, the goiter generally resolves. Sixty percent of neonates with Grave's disease have a benign course, 20 percent have symptoms after 6 months, and the remaining 20 percent have neonatal hyperthyroidism leading to death. In most cases, the disease subsides spontaneously within 1 to 3 months. Therapy of hypothyroid infants is extremely important, since untreated infants develop serious mental retardation.

Obstetrical Management
The most serious complication of congenital goiter is respiratory distress due to obstruction of the airway. Therefore, these infants should be delivered in a

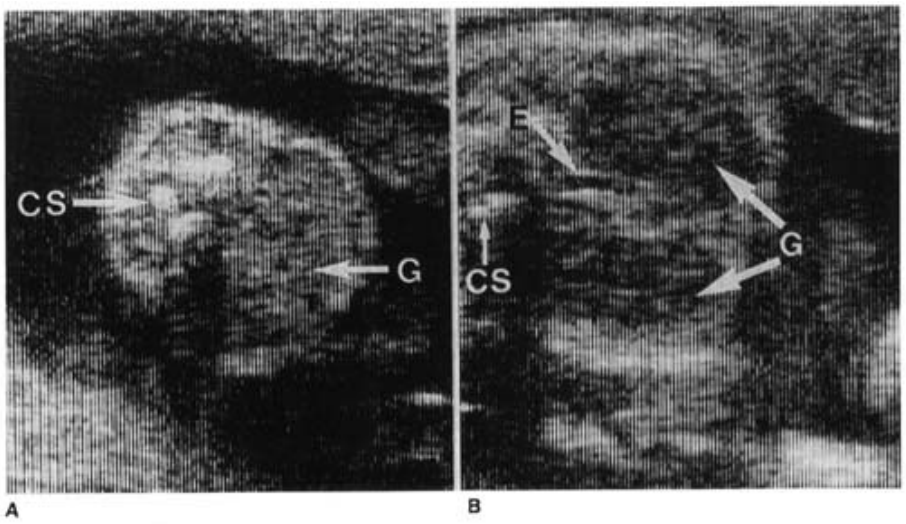

Figure 3-11. A. Fetal goiter at 27 weeks. The back of the neck and cervical spine (CS) are on the left. Echolucent areas are evident within the substance of the goiter (G). **B.** Fetal goiter at 36 weeks. The cervical spine is again on the left. Swallowed amniotic fluid is visible within the esophagus (E). The goiter is clearly bilobed in this view. *(Reproduced with permission from Kourides et al.: J Clin Endocrinol Metab 59:1016, 1984)*

center where resuscitation can be performed immediately after birth. A prenatal diagnosis of the functional state of the thyroid gland (hypothyroidism versus hyperthyroidism) can be attempted by assessing levels of TSH in the amniotic fluid.[7,8] Fetal goiter is not an indication to alter standard obstetrical management.

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Teratoma of the Neck

Synonyms
Cervical teratoma and thyroid teratoma.

Definition
Germ cell tumor located in the neck.

Incidence
One hundred thirty cases have been reported as of 1983.[3] There is no sexual predilection.

Etiology
The anomaly is sporadic, and the causes are unknown. One familial case has been reported in the literature. [5]

Pathology
Ninety percent of all teratomas are recognized at birth or soon thereafter.[7] Tumors are generally unilateral and encapsulated. They vary in size and generally consist of a mixture of cystic and solid components.

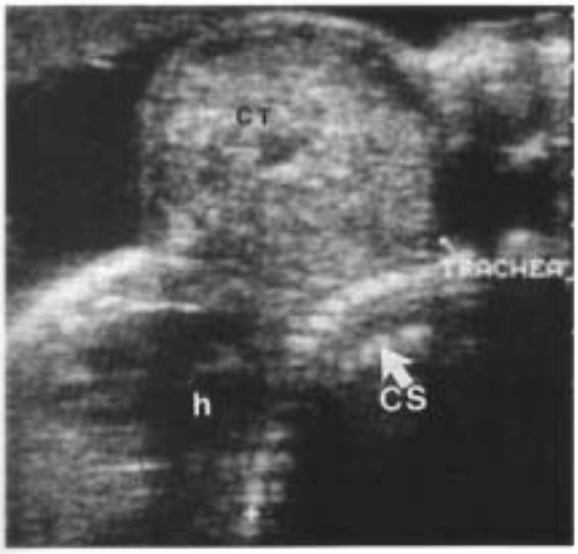
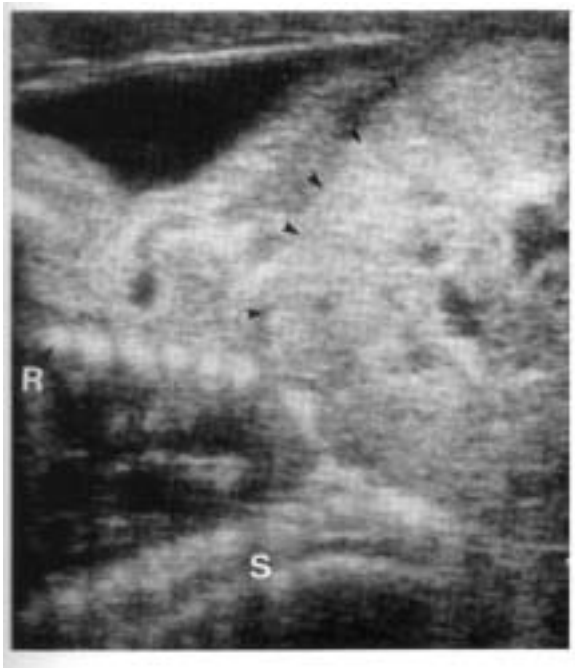


Figure 3-12. Cervical teratoma (CT). A solid mass is seen in the anterior aspect of the fetal neck. CS, cervical spine; h, heart.

Malignant transformation is extremely rare, and there are no reports of recurrence after complete surgical excision in the neonatal period.[8] Calcification can be detected in 50 percent of cases.[3] Neural tissue is the predominant histologic component. Obstruction of the airway by the tumor may lead to acute respiratory failure during the newborn period.[3] Polyhydramnios has been reported as a complication in 30 percent of patients and is thought to result from esophageal obstruction[s] .A correlation between the size of the mass and polyhydramnios has been reported. Masses larger than 10 cm are more likely to be associated with this complication than are smaller tumors.[4]



3-13. Cervical teratoma is seen as a complex mass above apex of the chest. Arrows point to the margins of the tumor. The complex nature of the mass is apparent. S, spine; R, ribs.

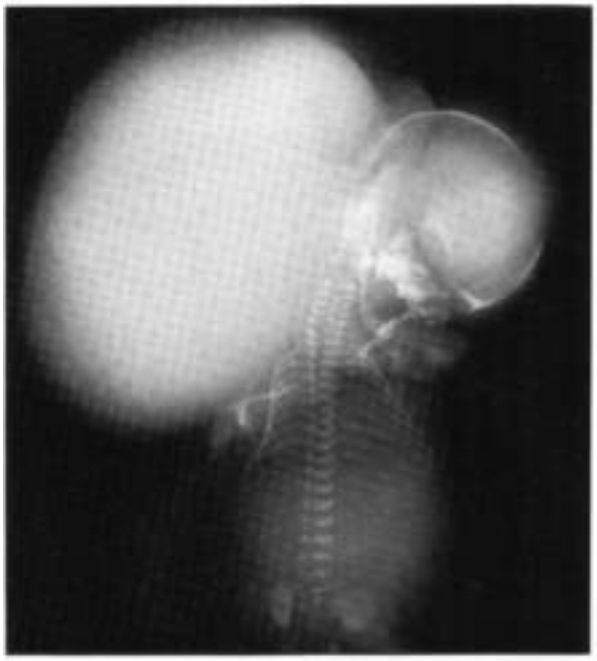


Figure 3-14. Postnatal radiograph showing cervical teratoma.

Associated Anomalies

These are very uncommon. Isolated patients have had pulmonary hypoplasia, imperforate anus,[9] trisomy 13,[1] and chondrodystrophia fetalis[9] in association with these tumors.

Diagnosis

The diagnosis relies on demonstration of a complex mass in the cervical region (Figs. 3-12 to 3-14).[10] The differential diagnosis includes cystic hygroma, goiter, bronchial cysts, cervical meningocele, neuroblastoma of the neck, and hemangiomas of the neck. Cystic hygromas are purely cystic lesions with a typical honeycomb appearance. Cervical meningoceles appear as masses of the neck, and a spinal defect can be demonstrated. Cervical myelomeningoceles can give a mixed pattern, but their location and the presence of an associated spinal defect should be diagnostic.[6] Goiters are solid, generally devoid of cystic compo-

nents, symmetrical, anterior, and generally do not reach the size achieved by some teratomas. Branchial cleft cysts are purely cystic masses located in the anterior border of the sternomastoid muscle. Hemangiomas can appear as cystic or solid masses. A differentiation between mesenchymal tumors and teratomas may not be possible because the former are generally solid tumors. Neuroblastomas of the neck can produce masses of mixed consistency.[2] Polyhydramnios is a frequent finding. Calcifications are present in 40 to 45 percent of the cases. [8]

Prognosis

A stillbirth rate of 17 percent has been reported.[4,7] The mortality rate of untreated infants has varied from 80 to 100 percent.[3,8] Upper airway obstruction is the major cause of death. Operative mortality is 9 to 15 percent.[3,8] Most tumors are benign, and no recurrences have been reported after total excision in the neonatal period.

Obstetrical Management

Large tumors may cause dystocia, and a cesarean section is indicated in these instances. In the presence of small tumors, standard obstetrical care should not be altered. Serial scans are indicated to monitor tumor growth and amniotic fluid volume. Delivery in a tertiary care center is mandatory, and a pediatric team must be prepared to intubate the infant immediately after birth if necessary. An amniogram may be considered in patients with severe polyhydramnios as an indirect mean to assess the degree of tracheoesophageal obstruction.

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Fetal Nuchal Skin Thickening

Definition

Increased soft tissue thickening in the posterior aspect of the neck.

Diagnosis

Diagnosis is made by the detection of soft tissue nuchal thickness above 5 mm in a fetus between 15 and 20 weeks of gestation. The measurement is generally taken in an axial plane at the level of the thalami, but it can also be taken in sagittal sections of the fetal neck (Figs. 3-15, 3-16)[1,2]

Significance

Increased soft tissue thickening in the nuchal region is one of the eight major criteria for the diagnosis of trisomy 21 in newborn infants.[5,9,12] It is present in 80 percent of newborns with trisomy 21[5,9] and in 45 percent of midtrimester fetuses with trisomy 21 and in only 0.06 percent of normal fetuses. These data are based on a series of 1704 consecutive midtrimester amniocenteses, of which there were 11 fetuses with trisomy 21.[1,2] If these data are confirmed, fetal nuchal thickening can be used as a means of identifying fetuses at risk for trisomy 21 in a low-risk population (maternal age below 35 years). The value of this approach though, has been challenged recently.[13]

Other conditions associated with redundancy of the skin at the level of the neck in the newborn period and, therefore, skin thickening include:

Chromosomal Syndromes[10]

1. 13q syndrome
2. XXXX syndrome
3. XXXXY syndrome
4. Trisomy 18
5. 18p- syndrome

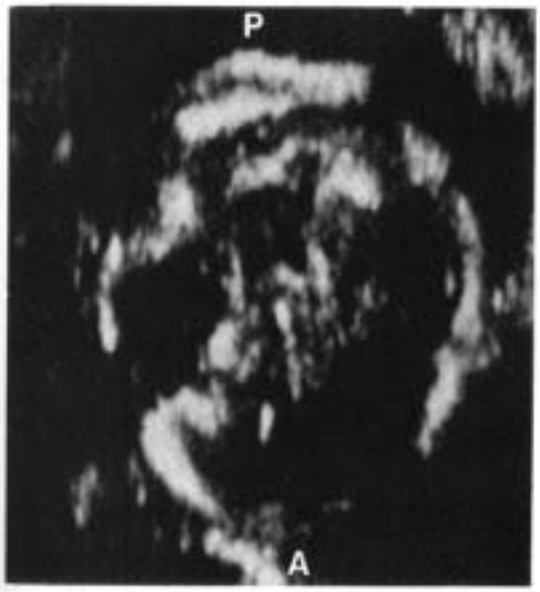

Figure 3-15. Axial scan of a normal fetal head close to the base of the skull. Note the occipital region devoid of soft tissue. P, posterior; A, anterior.

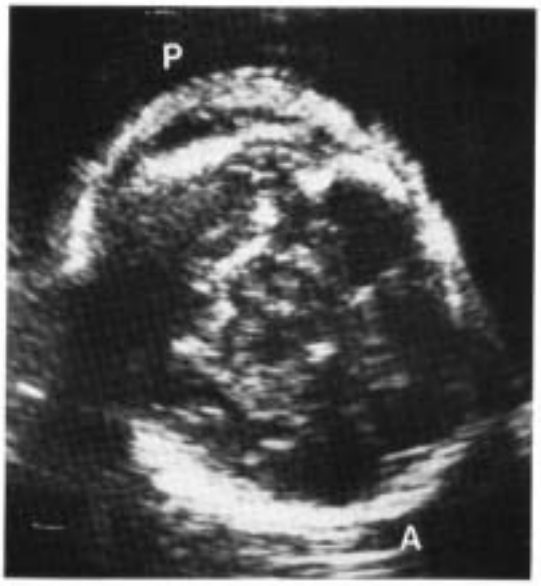

Figure 3-16. Down syndrome fetus. The head is scanned at the same level as in Figure 3-15. Note the nuchal skin thickening. P, posterior; A, anterior. *(Courtesy of Dr. B. Benacerraf.)*

Nonchromosomal Disorders

1. Multiple pterygium syndrome (Escobar syndrome): Characterized by pterygia of the neck, axillae, elbows, and knees, micrognathia, campodactyly, syndactyly, and rocker-bottom feet (autosomal recessive). [4]
2. Klippel-Feil sequence: consists of fusion of the cervical vertebrae; other associated anomalies include congential heart defects (ventricular septal defect is the most common),[6] deafness (30 percent),[8] and cleft palate (sporadic in most cases, autosomal dominant with variable expression)[6,8]
3. Zellweger syndrome (cerebrohepatorenal syndrome): large forehead with shallow supraorbital ridges, flat facies (have been confused with infants with Down syndrome), mild micrognathia macrogyria, polymicrogyria, hepatomegaly, cystic kidney disease, contractures in extremities, equinovarus, simian crease, elevated serum iron (autosomal recessive)[3,7,11]

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