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LITERATURE REVIEW

The diagnosis of a cervical tumefaction in children

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ABSTRACT

Neck pathology in children involves many diseases with similar clinical expression. Different disorders in paediatric practice may be clinically manifested by a cervical tumefaction. The cervical tumefaction can be represented by adenopathies or tumors. Cervical adenopathies may be unilateral or bilateral; the cause of their occurrence may be infectious, inflammatory, tumoral or may be associated with rare diseases. Tumoral cervical tumefactions can be represented by: congenital cervical cysts and fistulae, cystic lymphangiomas, thyroid swellings, malformations and cervical tumors detected antenatally. A correct diagnosis is established after a complete clinical and paraclinical assessment. In this article the authors make a review of the types of cervical tumefactions in children, emphasising the diagnostic protocol.
KEYWORDS: cervical tumefactions, cervical adenopathies, congenital cervical cysts, cervical malformations, thyroglossal duct cyst

INTRODUCTION

Neck pathology in children involves many diseases with similar clinical expression. Different disorders in paediatric practice may be clinically manifested by a cervical tumefaction.

The cervical tumefaction can be represented in approximately 95% of cases by adenopathies and in 5% by tumors. Cervical adenopathies may be unilateral or bilateral; the cause of their occurrence may be infectious, inflammatory, tumoral or may be associated with rare diseases. Tumoral cervical tumefactions can be represented by: congenital cervical cysts and fistulae, cystic lymphangiomas, thyroid swellings, malformations and cervical tumors detected antenatally.

CONGENITAL CERVICAL CYSTS AND FISTULAE

Congenital cervical cysts and fistulae must be taken into account when diagnosing a cervical tumefaction in children. They develop from the branchial clefts and may have lateral localization (uni- or bilateral) or median one, when talking about the thyroglossal duct cyst.

A. Lateral congenital cervical cysts and fistulae

These cervical tumefactions originate in the branchial clefts and represent approximately 30% of the total cervical tumefactions[1-9].

By the end of the fourth month of gestation, the four main arches and two rudimentary ones form. From each branchial arch form certain anatomical structures, which is why it is essential to know embryology and anatomy in order to establish a diagnosis and appropriate treatment of these embryonic structures anomalies (Figure 1).

Cervical tumefactions originating in the first branchial cleft represent approximately 8% of the total anomalies of the branchial clefts[10,11].

From the first branchial cleft form the mandible, the external auditory canal, the Eustachian tube, the eardrum cavity, the mastoid cells and a portion of the inner ear.

The cysts and fistulae derived from the first branchial cleft can be classified both from the anatomical and histological points of view. Anatomically, cervical tumefactions originating in the first branchial cleft were classified in 1971 by Arnot as[12,13]:

- Type I – develop in the parotid region;
- Type II – appear at the level of the anterior triangle of the neck, communicating with the external auditory canal (high frequency in children).

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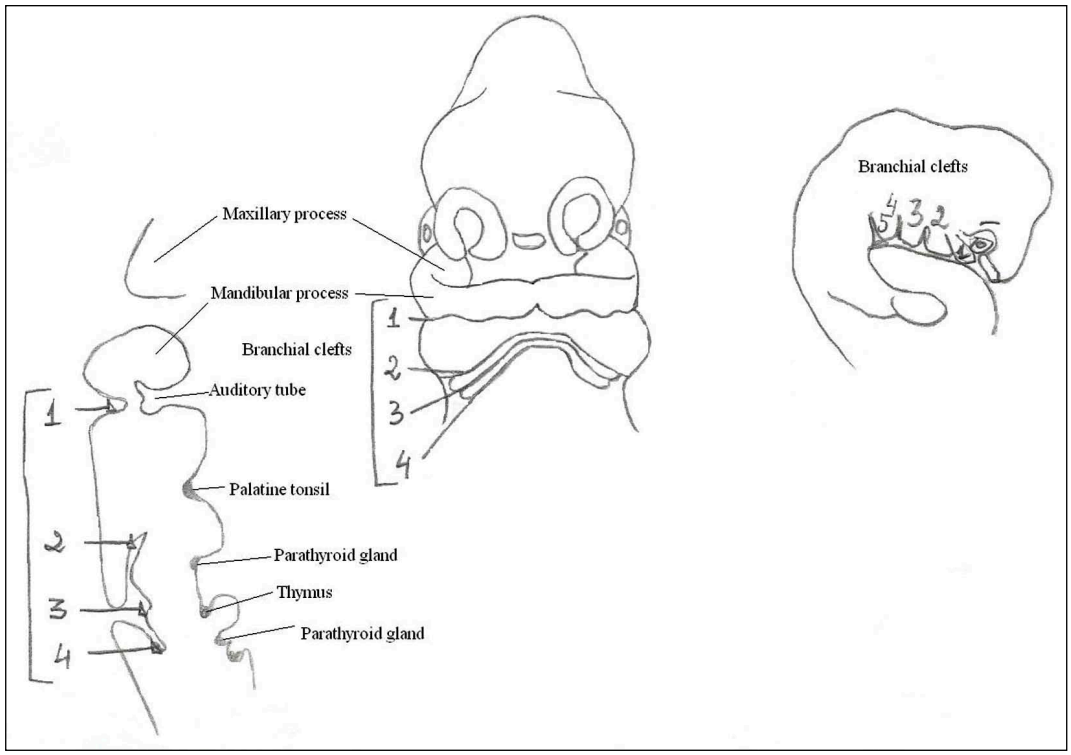

Figure 1 Branchial clefts – lateral anatomical structures

Work classified these anomalies from a histological point of view[13,14]:

- Type I – has ectodermal origin, developing by an overlay-type cleavage at the level of the external auditory canal (EAC), the eardrum and the mastoid having a normal appearance;
- Type II – ecto- and mesodermal origin – located pretragally, in relation to the EAC being located anteriorly/inferiorly/posteriorly. It can be associated with fistulous tracts that may extend superiorly through the parotid gland up to the EAC.

In approximately 10% of the cases[9,15], cervical anomalies originating in the first branchial cleft are accompanied by a pathognomonic sign – a membranous attachment from the floor of the EAC to the tympanic membrane.

The cysts or fistulae derived from the first branchial cleft are located in the Poncet's triangle, externally bounded by the EAC, anteriorly by the mental region and inferiorly by the hyoid bone. Any swelling occurring in this region should raise the suspicion of an anomaly of a branchial cleft.

The CT exam has a specificity of about 64%[1,10]. In the case of congenital fistula, the fistulogram may be of great use in diagnosis[11].

The differential diagnosis must be done with the parotid tumors, maxillary gland tumors, vascular tumors, and adenopathies (tumoral, infectious).

The treatment is surgical. For a complete excision, especially in type II, it is recommended an exploratory parotidectomy with dissection of the facial nerve and complete excision of the fistulous tract, in order to avoid relapse[9,10,16].

Anomalies of the second branchial cleft

The cysts or fistulae derived from the second branchial cleft represent 95% of the total anomalies of the branchial clefts[17,18]. Their localization is on the anterior edge of the sternocleidomastoid (SCM) muscle, and they may be unilateral or bilateral.

This type of pathology affects children at a rate of 66-75%.

Clinically, the anomalies of the second branchial cleft present in the form of a latero-cervical tumor, soft to the touch and painless. The fistulous tract passes through the bifurcation of the two carotid arteries and reaches up to the tonsillar lodge[9].

The cervical cysts and fistulae derived from the second branchial cleft may be thus classified (Figure 2)[1,19]:

- Type I – located anteriorly to the SCM muscle, however without entering into contact with the carotid artery;
- Type II – are located between the SCM and anteriorly or posteriorly to the carotid artery;
- Type III – pass through the bifurcation of the carotid artery, being adjacent to the pharynx;
- Type IV – located medially to the carotid artery, in close contact with the pharynx; they reach the tonsillar lodge.

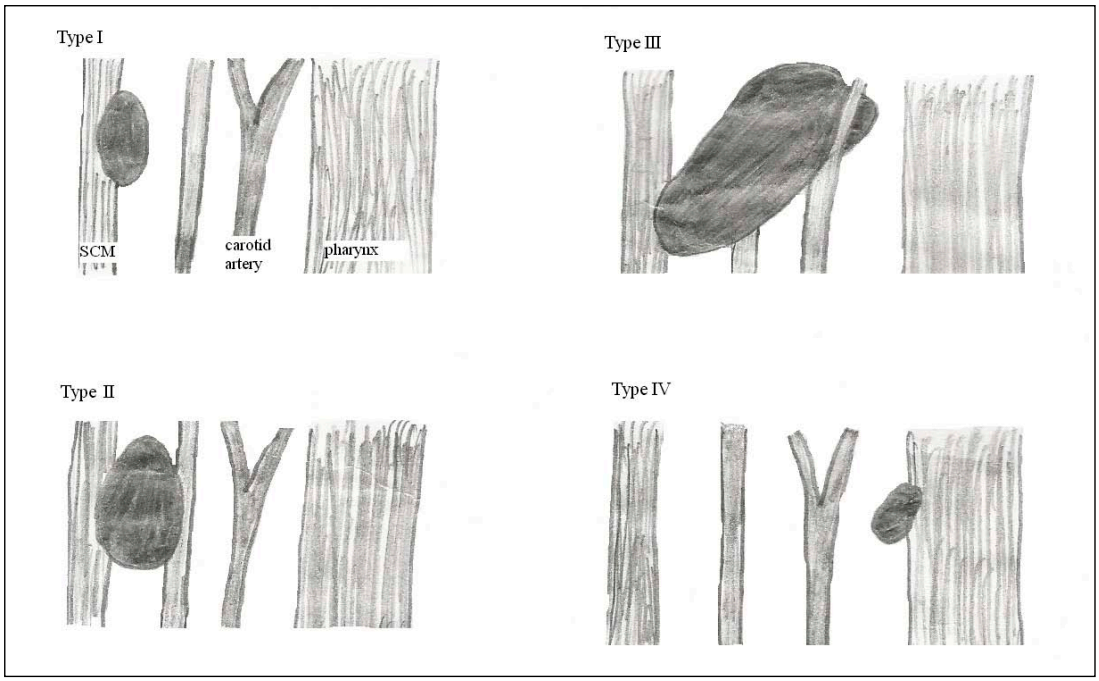

Figure 2 Classification of the cervical cysts derived from the second branchial cleft

Fistulae are much more common in children, the fistulous opening being usually located on the anterior edge of the SCM muscle, in the lower third of the neck.

In most cases, the anomalies of the second branchial cleft are part of the branchio-oto-renal syndrome, also known as the Melnick-Fraser syndrome[20].

The differential diagnosis of this pathology must be done with the lymphangioma, the thymic cyst, the jugulodigastric lymph nodes, the vagal adenoma, etc.

The treatment of choice is the surgical one, consisting of complete excision in order to avoid relapses.

Anomalies of the third and the fourth branchial clefts

The third and the fourth branchial clefts represent the starting point in the formation of the pharynx, the part located below the hyoid. The anomalies of these two clefts are very rare, fistulae opening in the pyriform sinus. The cysts and fistulae are most often located in the left laterocervical region.

In newborns, due to rapid growth of cysts, they can cause a compression phenomenon at the level of the larynx. Also, the cysts may mimic acute thyroiditis, and the infection can fuse to the skull base.

Therefore, the differential diagnosis is essential and must be done with the thyroglossal duct cyst, the thymic cyst, the thyroiditis and the thyroid abscess, lymphangiomas, the parathyroid cyst.

The curative treatment is the surgical one, the endoscopic technique being preferred, by suspension laryngoscopy, with identification of the fistulous orifice at the level of the pyriform sinus. The literature describes chemical cauterizations of these orifices, much less used techniques in medical practice[21]. CO2 laser vaporization seems to give the best results.

Moreover, a hemithyroidectomy can be performed with partial resection of the thyroid cartilage for a perfect exposure of the pyriform sinus and identification of the entire fistulous tract[1,22].

Thymic cysts

Thymic cysts represent approximately 3% of the tumors identified at the level of the neck and the mediastinum in children[23-25]. The new hypotheses argue congenital etiology of thymic cysts, originating at the level of the branchial clefts[24,26,27].

From a clinical point of view, the thymic cyst presents under the form of a voluminous tumor mass, most frequently located in the left laterocervical region. In approximately 75% of the cases, the tumor mass becomes manifest before 20 years of age[27]. Most of the thymic cysts are accidentally discovered, during radiography or CT performed for another pathology.

In the case of thymic cysts, the differential diagnosis is made with the branchial cysts, the thyroglossal duct cyst, the cystic hygroma, the lymphadenopathy, the lymphoma, etc.

The treatment is represented by the surgical excision. A characteristic of thymic cysts is the extremely easy digital removal. Because of its predominant left location, care should be taken to the recurrent nerve during surgery.

B. Median congenital cervical cysts

Thyroglossal duct cyst

The thyroglossal duct cyst is the second most frequent cervical anomaly, after adenopathy; approximately 7% of the population has this pathology[28]. It occurs due to a closure deficit of the thyroglossal duct between the base of the tongue and the thyroid gland. Thus, two-thirds of thyroglossal duct cysts are diagnosed in the first three years of life, more than half up to the age of 10[1].

Clinically, it presents as a tumor mass located cervically, on the median line, painless, mobile, with cystic consistency on palpation.

In terms of location, the thyroglossal duct cyst is situated mid-cervically; it can be subhyoidian (in 60% of the cases), suprahyoidian (24%), suprasternal (13%) and intralingual (2%)[29,30]. Much more rarely, these tumors may be localized inside the hyoid bone[31] or in the mediastinum[32].

Standard or 3D ultrasound represents the paraclinical examination decisive in diagnosing the thyroglossal duct cyst. The cervical CT scan can be useful in cases where malignant transformation is suspected. Also, a thyroid scintigraphy can be performed in order to establish the level of functioning of the thyroid gland[33].

In approximately 1% of the cases, a malignant transformation of the thyroglossal duct cyst has been reported, the most frequent being the papillary thyroid carcinoma[29,34,35]. Less often, squamous cell carcinoma or Hurthle cell carcinoma is reported to develop[36]. In the literature, the coexistence of two types of carcinomas has also been described, respectively the squamous and the papillary ones[37].

Besides malignant transformation, the cyst may rupture or become superinfected, the most common incriminated germs being: Staphylococcus epidermidis, Haemophilus influenzae, Staphylococcus aureus[38].

The differential diagnosis of the thyroglossal duct cyst must be made with the dermoid cyst, the lipoma, the anomalies of the branchial clefts, the sebaceous cyst, the metastatic thyroid carcinoma, etc.

The treatment of choice is complete surgical excision, the most used technique being the Sistrunk procedure[39,40]. The surgical technique involves resection of the thyroid bone, with tracking under intrabuccal digital inspection and excision of the tract up to the foramen cecum. The degree of relapse in the case of the Sistrunk procedure has decreased from 50% to 2.6-5%[1,4,41]. In case of an incomplete excision, the degree of relapse can reach up to 38-70%[1,4]. The risk is far greater in case of reinterventions, when it can reach 20-35%[2].

CERVICAL CYSTIC LYMPHANGIOMAS

Lymphangiomas are benign tumors, most commonly located in the head and neck region. They are congenital lymphatic malformations, representing approximately 5% of the total lymphatic tumors of the neck in children[42]. The most increased frequency of this pathology is described in the first years of life (80-90%)[43].

In terms of occurrence, lymphangiomas develop through a deficit of connection of the primitive cervical lymph sac with the rest of the lymphatic system[44]. From the location point of view, lymphangiomas may be situated: laterocervically, with a soft consistency, painless; above the hyoid, with a 27-month development and a microcystic structure; below the hyoid, with occurrence at 36 months; oropharyngeally (on the floor of the mouth or the tongue) when it associates with serious functional disorders[45].

Cervical lymphangiomas can be classified as[46-48]:

- Capillary lymphangioma – microcystic consistency;
- Cavernous lymphangioma – macrocystic consistency;
- Cystic hygroma (represent approximately 90% of the total cervical lymphomas[47]).

Clinically, they manifest by the presence of a tumor formation of soft consistency, fluctuating on palpation, and mobile on the superficial and deep levels. Cervical lymphangiomas may be isolated or they can accompany certain syndromes, such as the Turner syndrome, the congenital glaucoma, the Klippel-Trenauny syndrome[49,50].

Paraclinical investigations are particularly important in establishing the diagnosis. These must include simple radiography, cervical ultrasound, CT exam and MRI of the cervical region.

The differential diagnosis in case of cervical lymphangiomas must be made with the adenopathies (inflammatory or infectious), teratomas, malignant tumors with cervical localization, or with the thyroid tumors.

Evolutionarily, these tumors can extend into the parapharyngeal space, affecting cranial nerves. Some cysts occurring in children may regress in approximately 15% of the cases[51].

The treatment of choice is surgical. In case of complete excision, the relapse rate can be of 10-27% and may reach up to 50-100% in case of incomplete excisions[47,52].

In the literature, alcohol sclerosing injections are also described. Nevertheless, these were abandoned because of the increased risk of complications, which is due to the major vascularisation and innervation, and because of the vital risk at the level of the neck[53].

THYROID TUMEFACTIONS

Thyroid tumefactions in children are always represented by the single, solitary nodule. The prevalence of thyroid nodules in children was reported to be between 0.22 and 1.35%, their incidence being much higher among adults – 13% between 18 – 39 years and 50% over 50 years[45,54].

In 20-25% of the cases, malignant transformation of the thyroid nodules was noticed[55-58].

Clinically, the thyroid nodule appears as a unique, solitary, adherent nodule, which associates with adenopathies. On ultrasounds, the nodule has a hypoechoic appearance, with microcalcifications, a perinodular halo, diffuse boundaries and it is hypervascularized.

The differential diagnosis is made with the thyroglossal duct cyst, Hashimoto's thyroiditis, Graves' disease, nodular hyperplasia.

In order to determine the correct diagnosis, the following are compulsory: ultrasound, thyroid scintigraphy, plasma thyroid hormones levels (TSH, thyroglobulin) and calcitonin. Cytodiagnostic puncture can be positive in 80-100% of the cases.

Surgical treatment of the thyroid nodule found in children is mandatory.

CERVICAL MALFORMATIONS AND TUMORS DETECTED ANTENATALLY

The development of the foetus head and neck begins between the 4th and 5th month of pregnancy. Cervical tumefactions occurring intrapartum develop from the four branchial clefts. The most frequent are cystic hygroma, teratomas, sarcomas, hemangiomas or lymphomas[59,60]. Far more rare are congenital cervical neuroblastomas[61,62].

The main complication of cervical swellings is represented by the phenomenon of compression that they can manifest on the upper airways. Thus, in the literature, a risk of intrauterine fetal death of up to 20% and of 35% immediately after birth is reported[63,64]. For these reasons, early diagnosis of these malformations or tumors is extremely important.

Changes in the neck are usually discovered during the second mandatory ultrasound in pregnancy. The morphological ultrasound is performed in weeks 18-20 of pregnancy, precisely to detect any congenital malformation. The 3D ultrasound provides three-dimensional images of the cervical region; it may therefore be successfully used in detecting cervical tumefactions. In case of suspicion of a vascular anomaly, the Doppler ultrasound can be associated to the battery of tests.

If a cervical tumor formation exists, it is indicated to perform an MRI examination, which can supply more details regarding its characteristics, as well as whether there is a mass effect on the trachea and the esophagus[60,65].

In case of such pathology, periodic evaluation is mandatory and delivery by caesarean section is indicated[66], associated with the ex utero intrapartum treatment (EXIT). If left untreated, fetal mortality can be of 80-100%[59,66]. EXIT consists in uterine exposure to perform a hysterotomy by revealing the fetal head and neck. After ensuring proper fetal breathing, cutting of umbilical cord is performed.

Cervical tumors detected antenatally, besides severity, also represent a matter of medical and medico-legal ethics. Apart from communicating the diagnosis to the parental couple, it also implies deciding upon the course of the pregnancy. The latter is discussed within a pluridisciplinary commission. If the pregnancy is maintained, the presence at birth of the ENT specialist is compulsory, his duty being to stabilize the breathing of the foetus.

CERVICAL ADENOPATHIES

Adenopathies represent approximately 95% of the cervical tumors diagnosed in children. In 38-45% of the healthy paediatric population, the cervical lymph nodes may be palpable[67].

Most frequently, cervical adenopathies are infectious or inflammatory, having an acute and subacute evolution.

Inflammatory cervical adenopathies have a viral etiology, most commonly involved being the viruses responsible for the upper airways infections – rhinoviruses, adenoviruses, the respiratory syncytial virus, the parainfluenza virus[68]. Besides these, are also incriminated: the cytomegalovirus, the rubella and measles virus, the varicella zoster virus, the herpes virus or the human immunodeficiency virus[69,70].

Bacterial cervical adenopathies are, in most of the cases, the result of infection with streptococci and staphylococci. Involvement of anaerobic bacteria may be suspected in cervical adenopathies that accompany dental abscesses or periodontitis.

Bacterial infection with mycobacteria or Bartonella henselae is rarely described[71,72]. The infection with Bartonella hensalae, also known as the cat-scratch disease, is characterised by the appearance of a purple, fugitive pustule, associated to a unilateral jugular-carotid adenopathy. The spot of inoculation can suppurate, the adjacent tegument being tumefied. The disease can heal spontaneously in about 3 months, the basic treatment being however the antibiotic.

Mycobacteria are ubiquitous bacteria that are found in soil, water, air, vegetable and animal kingdoms. The most affected by this infection is the immunosup-

pressed child. The most common location is ganglionary; the infection can be achieved by cutaneous, digestive or pulmonary route.

Another cause of occurrence of a cervical adenopathy is infection with Mycobacterium tuberculosis. Cervical lymph node tuberculosis appears especially in endemic areas, the most affected being the immunosuppressed, malnourished children, coming from families or entourage with TB. Positive diagnosis is established by identifying the Koch bacillus in the sputum, IDR positivity and significant radiological imaging.

Cervical adenopathies caused by parasites may be the only manifestation of the disease in approximately 50% of the cases[73], the most commonly incriminated being toxoplasmosis.

If a cervical adenopathy persists despite correct antibiotic and anti-inflammatory treatment indicated and carried out, if it becomes of pasty consistency, and teguments change their appearance, *a rare etiology of the adenopathy* has to be considered. Kawasaki disease manifest by cervical adenopathy, conjunctival erythema, severe inflammatory syndrome, polymorphous rash, oral mucosal ulcerations and palmoplantar desquamation. In this disorder, immunoglobulins have the most effective therapeutic effect. Histiocytic necrosis of cervical ganglia or the Kikuchi-Fujimoto disease manifest by lymphadenopathy associated with fever, nausea, weight loss, arthralgia, with or without hepatosplenomegaly.

In the Kimura disease, besides adenopathy, the patient also presents hypereosinophilia.

Malignant a*denopathies* are the most common malignant tumors diagnosed in the paediatric population[73]. Neuroblastoma, leukemia or rhabdomyosarcoma are the most frequent malignancies diagnosed in children less than 6 years of age, while after the age of 6, Hodgkin lymphoma is more frequently found, being followed by non-Hodgkin lymphoma and rhabdomyosarcoma[69,73].

From a clinical point of view, the adenopathy becomes fixed on the superficial and deep levels, hard on palpation. Fever and impaired general condition are suggestive of lymphoma.

An accurate diagnosis necessitates a cervical ultrasound, a cervical MRI, as well as a scintigraphy and monitoring of urinary catecholamines when suspecting a neuroblastoma.

CONCLUSIONS

The cervical tumefaction in children may have an infectious and inflammatory cause in 95% of the cases, the rest of 5% being cysts, fistulae or tumors.

Most of the inflammatory or infectious adenopathies succumb in 3 weeks if treatment is correctly performed. Persistence of the adenopathy or modification of its characteristics associated with an impaired general condition suggests a rare etiology disease or a complication. In such cases, it is compulsory to know the possible causes of a cervical adenopathy in the paediatric population.

A correct diagnosis is established after a complete clinical and paraclinical assessment. The clinical assessment implies a complete clinical examination, knowing the family history, embryology and anatomy. The cytopuncture with extemporaneous examination can determine the diagnosis and may help in establishing the differential diagnosis.

From a paraclinical point of view, are recommended: the standard ultrasound, the 3D ultrasound or the eco-Doppler, the MRI exam and/or a magnetic resonance angiography of the cervical region. If a neuroblastoma is suspected, the scintigraphy and monitoring of the urine catecholamines and the LDH may help in establishing the diagnosis.

The diagnosis and especially the right treatment of the cervical tumefactions discovered in children require an interdisciplinary collaboration – otorhinolaryngologist, pediatrician, infectionist, neonatologist, obstetrician, radiologist and anatomopathologist.

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