

## Hereditary Thyroglossal Duct Cyst: A Literature Review

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### Abstract

**Introduction:** Thyroglossal duct cysts are the most common form of congenital cyst in the neck, with an incidence around 7%. The precise pathogenesis of thyroglossal duct cyst has not been determined, and despite their relative frequency, reports of familial inheritance are rare. The aim of this article is to gather information about the existing evidence of hereditary thyroglossal duct cysts.

**Material and Methods:** A literature review was performed with keywords: hereditary AND thyroglossal duct, between 1975 and 2018 in MEDLINE database.

**Results:** 34 cases were found worldwide, divided into ten families where the thyroglossal duct cyst is found in one to three generations. The majority of published family thyroglossal duct cyst series suggest an autosomal dominant mode of inheritance with an incomplete penetration.

**Discussion:** The reports of hereditary cases of thyroglossal duct cysts are increasing, in spite of this, its correlation with age of manifestation, gender, geographical distribution, and association with other thyroid anomalies is yet to be established.

**Keywords:** Thyroglossal duct cyst, hereditary, familial, genetic.

### INTRODUCTION

Thyroglossal duct cysts (TGDCs) are the most common form of congenital cyst in the neck, with an incidence around 7% (1). They are cysts of epithelial remnants of the thyroglossal tract, that fails to regress by the 10th week of foetal development, and sporadically present a slowly enlarging midline neck mass at the level of the thyrohyoid membrane, closely associated with the hyoid bone (2). Fifty percent of cases present by the age of 20 years (3), and it seems that there are no differences in terms of gender, although not without some controversy (1,4).

The precise pathogenesis of TGDC has not been determined, and despite their relative frequency, reports of familial inheritance are rare. Few records

of this entity, show that TGDC is found in one to three generations (5,6), but there is evidence of up to ten generations (7). Their mode of genetic transmission is not elucidated yet, but autosomal dominant transmission at infinite distance would be the more likely option (8). This can also be found in other types of congenital alterations in the neck (9). The preponderance of the female sex could however, translate a complex transmission mode called genetic imprinting.

The aim of this article is to gather information about the existing evidence of hereditary TGDC.

### MATERIAL AND METHODS

A literature review was performed with keywords: hereditary AND thyroglossal duct, between 1975

and 2018 in MEDLINE database. No exclusion criteria were applied in terms of language, type of study or text availability.

### RESULTS

Articles obtained show 34 cases reported worldwide. These cases are divided into ten families where the TGDC is found in one to three generations.

Two series report an attack on one generation. Klinet al. report the presence of a TGDC in three siblings (two brothers and one sister) (5) and at two sisters (6). In four publications, the presence of TGDC is found over two generations: a father and his son (5), a mother and son (10), a mother and two daughters (11), two brothers and their sister and a niece (7). Three authors report the presence of a TGDC over three generations. In Ashworth's study, the cyst is found in a grandmother, her daughter and her granddaughter (12). Millikan et al. describe an attack on a grandmother, her two daughters and three of her granddaughters (13). Finally, Ayache demonstrated the presence of hereditary TGDC in a grandmother, her son and two granddaughters (8), and Ramchandani et al. described five cases of three generations of the same family (14).

The genetic mechanism of these familial forms remains poorly established today because of the small number of published cases, but most of these cases have exhibited an autosomal dominant pattern with incomplete penetrance.

Concerning anomalies associated to the TGDC, Millikan et al. reported one case with hypothyroidism and one with anaplastic thyroid carcinoma (13). Issa et al. reported maternal grandmother of patient with autoimmune thyroiditis (Hashimoto's thyroiditis) and maternal grandfather with anaplastic carcinoma of thyroid (6). Finally, Ramchandani et al. described one case with hypothyroidism (14).

### DISCUSSION

TGDC appear in the form of a tumefaction later onset, between the sternal manubrium and the chin. The treatment is surgical and consists of the complete exeresis of the cyst. It must be done as soon as possible, before any episode of superinfection of the lesion. Although thyroglossal duct cysts are the most common congenital midline neck masses encountered in children, reports of familial occurrence are rare.

The most often mentioned mode of transmission is autosomal dominant. This method of genetic transmission has been described in the literature in syndromes with the presence of branchial cysts (9). The majority of published family TGDC series suggest an autosomal dominant mode of inheritance (5–7, 10–14). In some cases, the possibility of incomplete penetrance has been mentioned (5, 12, 14). Issa et al. described a male to male inheritance, rule out X linked transmission. A predominance of the female sex is shown, reporting a rate of 68% (7), contrary to what is described in sporadic cases, where male preponderance is slightly greater.

There are several hypotheses that try to explain this fact. Probably, the most recurrent and probable hypothesis is the so-called genetic imprinting or "fingerprinting phenomenon" (11, 15). This is a complex genetic model demonstrated in mammals and especially the mouse, insects and flowers. Usually, in autosomal transmission, each gene is represented by a copy or "allele" inherited from the father and the mother. Everyone is functional with the same opportunities to express themselves. The mechanism is poorly known. The most commonly accepted is the methylation of DNA segments most often bearing on segments rich in cytosine and guanine. This phenomenon is dynamic. The fingerprint can be erased and restored at each generation.

The association of TGDCs with other thyroid pathologies (cancerous, autoimmune, hypofunction) suggests that genetic factors might be involved and that mutated genes could have multiple and varied effects during thyroid development (6,13,14). In fact, it has been shown that among the first degree relatives of a congenital hypothyroidism population with thyroid dysgenesis, there is an elevated rate of asymptomatic thyroid developmental anomalies, even in euthyroid subjects (8% of cases) when they are systematically screened by ultrasound. These thyroid developmental anomalies include cysts of the thyroglossal duct, thyroid hemiagenesis, additional thyroid tissue with the presence of a pyramidal lobe, and ectopic thyroid tissue. These observations support the hypothesis that all thyroid developmental defects (if they lead to hypothyroidism or not) have a common genetic basis.

### CONCLUSION

The reports of hereditary cases of TGDCs are increasing, in spite of this, its correlation with age of

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manifestation, gender, geographical distribution, and association with other thyroid anomalies is yet to be established.

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