Natural history, treatment, and long-term follow up of patients with multiple endocrine neoplasia type 2B: an international, multicentre, retrospective study.

Titre	Natural history, treatment, and long-term follow up of patients with multiple endocrine neoplasia type 2B: an international, multicentre, retrospective study.
Type de publication	Article de revue
Auteur	Castinetti, Frederic [1], Waguespack, Steven G [2], Machens, Andreas [3], Uchino, Shinya [4], Lazaar, Kornelia [5], Sanso, Gabriella [6], Else, Tobias [7], Dvorakova, Sarka [8], Qi, Xiao Ping [9], Elisei, Rossella [10], Maia, Ana Luisa [11], Glod, John [12], Lourenço, Delmar Muniz [13], Valdes, Nuria [14], Mathiesen, Jes [15], Wohllk, Nelson [16], Bandgar, Tushar R [17], Drui, Delphine [18], Korbonits, Marta [19], Druce, Maralyn R [20], Brain, Caroline [21], Kurzawinski, Tom [22], Patocs, Atila [23], Bugalho, Maria Joao [24], Lacroix, André [25], Caron, Philippe [26], Fainstein-Day, Patricia [27], Chazot, Francoise Borson [28], Klein, Marc [29], Links, Thera P [30], Letizia, Claudio [31], Fugazzola, Laura [32], Chabre, Olivier [33], Canu, Letizia [34], Cohen, Régis [35], Tabarin, Antoine [36], Uroic, Anita Spehar [37], Maiter, Dominique [38], Laboureau, Sandrine [39], Mian, Caterina [40], Peczkowska, Mariola [41], Sebag, Frédéric [42], Brue, Thierry [43], Mirebeau-Prunier, Delphine [44], Leclerc, Laurence [45], Bausch, Birke [46], Berdelou, Amandine [47], Sukurai, Akihiro [48], Vlcek, Petr [49], Krajewska, Jolanta [50], Barontini, Marta [51], Vargas, Carla Vaz Ferrei [52], Valerio, Laura [53], Ceolin, Lucieli [54], Akshintala, Srivandana [55], Hoff, Ana [56], Godballe, Christian [57], Jarzab, Barbara [58], Jimenez, Camilo [59], Eng, Charis [60], Imai, Tsuneo [61], Schlumberger, Martin [62], Grubbs, Elizabeth [63], Dralle, Henning [64], Neumann, Hartmut P [65], Baudin, Eric [66]
Editeur	Elsevier
Type	Article scientifique dans une revue à comité de lecture
Année	2019
Langue	Anglais
Date	2019 Jan 16
Titre de la revue	Lancet Diabetes Endocrinol
ISSN	2213-8595

Résumé en anglais	BACKGROUND: Multiple endocrine neoplasia type 2B is a rare syndrome caused mainly by Met918Thr germline RET mutation, and characterised by medullary thyroid carcinoma, phaeochromocytoma, and extra-endocrine features. Data are scarce on the natural history of multiple endocrine neoplasia type 2B. We aimed to advance understanding of the phenotype and natural history of multiple endocrine neoplasia type 2B, to increase awareness and improve detection. METHODS: This study was a retrospective, multicentre, international study in patients carrying the Met918Thr RET variant with no age restrictions. The study was done with registry data from 48 centres globally. Data from patients followed-up from 1970 to 2016 were retrieved from May 1, 2016, to May 31, 2018. Our primary objectives were to determine overall survival, and medullary thyroid carcinoma-specific survival based on whether the patient had undergone early thyroidectomy before the age of 1 year. We also assessed remission of medullary thyroid carcinoma, incidence and treatment of phaeochromocytoma, and the penetrance of extra-endocrine features. FINDINGS: 345 patients were included, of whom 338 (98%) had a thyroidectomy. 71 patients (21%) of the total cohort died at a median age of 25 years (range <1-59). Thyroidectomy was done before the age of 1 year in 20 patients, which led to long-term remission (ie, undetectable calcitonin level) in 15 (83%) of 18 individuals (2 patients died of causes unrelated to medullary thyroid carcinoma). Medullary thyroid carcinoma-specific survival curves did not show any significant difference between patients who had thyroidectomy before or after 1 year (comparison of survival curves by log-rank test: p=0·2; hazard ratio 0·35; 95% CI 0.07-1.74). However, there was a significant difference in remission status between patients who underwent thyroidectomy before and after the age of 1 year (p<0·0001). There was a significant difference in remission status between patients who underwent thyroidectomy before and after the age of 1 year (p<0·0001). In the other 318 patients who underwent thyroidectomy after 1 year of age, biochemical and structural remission was obtained in 47 (15%) of 318 individuals. Bilateral phaeochromocytoma was diagnosed in 156 (50%) of 313 patients by 28 years of age. Adrenal-sparing surgery was done in 31 patients: three (10%) of 31 patients had long-term recurrence, while normal adrenal function was obtained in 16 (62%) patients. All patients with available data (n=287) had at least one extra-endocrine feature, including 106 (56%) of 190 patients showing marfanoid body habitus, mucosal neuromas, and gastrointestinal signs. INTERPRETATION: Thyroidectomy done at no later than 1 year of age is associated with a high probability of cure. The reality is that the majority of children with the syndrome will be diagnosed after this recommended age. Adrenal-sparing surgery is feasible in multiple endocrine neoplasia type 2B and affords a good chance for normal adrenal function. To improve the prognosis of such patients, it is imperative that every health-care provider be aware of the extra-endocrine signs and the natural history of this rare syndrome. The implications of this research include increasing awareness of the extra-endocrine symptoms and also recommendations for thyroidectomy before the age of 1 year. FUNDING: None.
URL de la notice	http://okina.univ-angers.fr/publications/ua18726 [67]
DOI	10.1016/S2213-8587(18)30336-X [68]
Autre titre	Lancet Diabetes Endocrinol
Identifiant (ID) PubMed	30660595 [69]

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