Etiologi hipotiroid kongenital primer = Etiology of primary congenital hypothyroidism

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Abstrak

[Latar belakang : Hipotiroid kongenital (HK) merupakan penyebab disabilitas intelektual yang dapat dicegah dan pencarian etiologinya belum menjadi prosedur rutin. Pencarian etiologi HK penting untuk dilakukan, karena membantu dalam menentukan derajat keparahan, mempengaruhi dosis substitusi L-tiroksin, terapi jangka panjang, prognosis, dan kemungkinan HK diturunkan pada anak selanjutnya (konseling genetik). Etiologi HK dapat bervariasi antar negara. Saat ini data mengenai etiologi HK di Indonesia masih sedikit.
Tujuan : Mengevaluasi etiologi hipotiroid kongenital.
Metode : Penelitian potong lintang dengan metode total sampling pada semua subjek yang terlibat dalam penelitian “Dampak keterlambatan diagnosis hipotiroid kongenital: disabilitas intelektual dan kualitas hidup pasien” di Jakarta. Penelitian ini dilakukan sejak lolos kaji etik sampai November 2014.
Hasil : Terdapat 19 dari 25 subjek yang dapat dievaluasi etiologinya. Etiologi yang ditemukan adalah disgenesis (16/19) dan dishormonogenesis (3/16). Tipe disgenesis terbanyak berturut-turut adalah hemiagenesis (6/16), athireosis (5/16), hipoplasia (4/16), dan ektopik (1/16). Nilai IQ pada kelompok hipoplasia adalah borderline, sedangkan kategori nilai IQ etiologi lainnya adalah disabilitas intelektual. Rerata nilai IQ 72,7(SD 30,3) untuk kelompok hipoplasia, 58,2 (SD 16) untuk agenesis, 52,5 (SD 16,5) untuk hemiagenesis, 37,3 (SD 8) untuk dishormonogenesis, dan nilai IQ 46 didapatkan pada anak dengan kelenjar tiroid ektopik.
Simpulan : Etiologi HK pada penelitian ini adalah disgenesis tiroid (16/19) dan dishormonogenesis (3/19). Hemiagenesis merupakan etiologi HK terbanyak (6/19). Hipoplasi tiroid merupakan kelompok dengan nilai IQ tertinggi (borderline) daripada kelompok lainnya (disabilitas intelektual).;Latar belakang : Congenital hypothyroidism (CH) is one of the most preventable cause of intellectual disability. Investigation for etiology of CH is not a routine procedure in Indonesia. Congenital hypothyroidism etiology is important for predict severity of hypothyroidism, L-thyroxine dose substitution, prognosis, and genetic counselling. Etiology of CH varies among countries. Current data about CH etiology in Indonesia is limited. This research is part of “Impact of delayed CH diagnosis: intellectual disability and quality of live” research that has been done in RSCM.
Tujuan : To evaluate etiology of primary congenital hypothyroidism.
Metode : A cross sectional study with total sampling of all participants in “Impact of delayed CH diagnosis: intellectual disability and quality of live” research. This research has been done since pass the ethics until November 2014.
Hasil : There were 19 of 25 participants that could be evaluate the CH etiology. The etiology are dysgenesis (16/19) and dyshormomogenesis (3/19). Types of dysgenesis are hemiagenesis (6/16), athireosis (5/16), hypoplasia (4/16), and ectopic (1/16). Mean of total IQ was 72,7 (SD 30,3) for hypoplasia, 58,2 (SD 16) for agenesis, 52,5 (SD 16,5) for hemiagenesis, 37,3 (SD 8) for dyshormonogenesis, and IQ score for ectopic

thyroid is 46.
Simpulan : Etiology of Ch in this research is dysgenesis (16/19) and dyshormonogenesis (3/19). Hemiagenesis is the most common etiology in CH. Hypoplasia thyroid group has the highest IQ score (borderline) among other groups od etiology., Latar belakang : Congenital hypothyroidism (CH) is one of the most preventable cause of intellectual disability. Investigation for etiology of CH is not a routine procedure in Indonesia. Congenital hypothyroidism etiology is important for predict severity of hypothyroidism, L-thyroxine dose substitution, prognosis, and genetic counselling. Etiology of CH varies among countries. Current data about CH etiology in Indonesia is limited. This research is part of “Impact of delayed CH diagnosis: intellectual disability and quality of live” research that has been done in RSCM.
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