

Department of Pathology and Laboratory Medicine

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REF LAB NO: 11111-08

NAME: Doe, Jane

MEDICAL RECORD: 00021210

PATHOLOGY CONSULTATION REPORT

ENDOCRINE AUTOANTI BODY STUDIES

ACCESSION DATE: 02/01/2008

COLLECTION DATE: 01/28/2008

References:

1. *Pediatr Res* (1983) 17:861;
 2. *Clin Endocrinol Metab* (1975) 4:379;
 3. *Endocrinol Metab Clin North Am* (1991) 20:619; 4. *J Pediatr* (1980) 97:191
- Diagnosis of pernicious anemia:**
Gastric parietal cell autoantibodies (PCA) detected in the presence of biochemically defined pernicious anemia and vitamin B12 deficiency identify an autoimmune etiology as the cause of the subject's pernicious anemia. This results from chronic lymphocytic atrophic gastritis with autoimmune destruction of the intrinsic-factor producing gastric parietal cells.

Prediction of pernicious anemia:

Parietal cell autoantibodies (PCA) detected in an asymptomatic individual indicates an increased risk for the subsequent development of chronic lymphocytic gastritis, achlorhydria and pernicious anemia. In asymptomatic PCA-positive individuals it is prudent to consider obtaining yearly measurements of vitamin B12 and ferritin.

Methodology:

PCA are assayed by indirect immunofluorescence using human stomach fundus as substrate.

Islet Cell Autoantibodies

Diagnosis of type 1 diabetes:

The presence of islet cell antibodies (ICA) in patients with diabetes mellitus indicates the presence of autoimmune type 1 diabetes. At the time of diagnosis of type 1 diabetes, ICA are identified in ~70% of Caucasian patients (1). Following the diagnosis of type 1 diabetes, ICA frequency declines.

Prediction of type 1 diabetes:

ICA in an asymptomatic individual places the subject at high risk for developing type 1 diabetes. This is especially true if the ICA-positive nondiabetic individual has a relative with type 1 diabetes (2). First-degree relatives of type 1 diabetes patients have a frequency of type 1 diabetes similar to their ICA frequency: 3-5%. ICA also predict the development of type 1 diabetes in individuals from the general population that lack a family history of type 1 diabetes (3). Higher ICA concentrations more strongly predict the later development of type 1 diabetes in nondiabetic individuals than minimally positive ICA titers (e.g., 10 JDF units). The presence of both ICA and insulin autoantibodies (IAA) greatly increases the risk of type 1 diabetes in asymptomatic individuals.

Relationship of ICA to Beta-cell function following the diagnosis of type 1 diabetes:

Within one year of the diagnosis of type 1 diabetes, the presence or absence of ICA does not appear to influence either insulin dose or endogenous C-peptide secretion (4). However, higher titer ICA have been associated with lower insulin secretion at 2 years following diagnosis (5).

Methodology:

Islet cell cytoplasmic autoantibodies (ICA) are detected by indirect immunofluorescence using human blood group O pancreas as substrate.

References:

1. *Diabetes* (1980) 29:589; 2. *New Engl J Med* (1990) 323:1167; 3. *Journal of Clinical Investigation* (1994) 93:2403; 4. *Diabetes Care* (1992) 15:66; 5. *Diabetes Care* (2000) 23:1072

Steroidal cell autoantibodies

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Diagnosis of primary gonadal failure:

Steroidal cell autoantibodies (SCA) are defined as autoantibodies that react with steroid hormone producing cells in the theca interna/granulosa layer of ovarian graafian follicles, cells of the ovarian corpus luteum, the placental syncytiotrophoblast and/or the Leydig cells of the testes. SCA can also react with cells of the adrenal cortex. In patients with hypergonadotropic hypogonadism, the presence of steroidal cell autoantibodies (SCA) supports the diagnosis of an autoimmune etiology for primary gonadal failure (1). If SCA are present, an independent determination about the presence of adrenal cytoplasmic autoantibodies can not be made because SCA can react with the adrenal gland as well as the gonads or placenta. SCA can be observed in type 1 autoimmune polyglandular syndrome (APS).

Prediction of primary gonadal failure:

Women with SCA are at increased risk of developing primary gonadal failure. It is unusual for men with SCA to develop primary gonadal failure. Patients with SCA are also at high risk to develop adrenal insufficiency and should undergo ACTH- stimulation testing to evaluate the patient for possible concurrent primary adrenal insufficiency.

Methodology:

Indirect immunofluorescence (2).

References:

- 1. J Clin Endocrinol Metab (1981) 52:1137; 2. Lancet (1968) Oct 26; 2(7574): 883**

TMA, TGA or TMA and TGA

Diagnosis of autoimmune thyroid disease (AITD):

The detection of either thyroid microsomal autoantibodies (TMA) or thyroglobulin autoantibodies (TGA) in the presence of biochemically defined hypothyroidism or hyperthyroidism or euthyroid goiter identify an autoimmune etiology as the cause of the subject's thyroid disease. Autoimmune hypothyroidism is most commonly due to Hashimoto thyroiditis (HT). Persistent autoimmune hyperthyroidism results from Graves disease. AITD is very common in the postpartum period in women and may be expressed as transient or permanent hypothyroidism, hyperthyroidism, or mixed sequential cases of hypo and hyperthyroidism. In a subject with a high suspicion for AITD, if TMA and TGA are negative initially, the tests should be repeated at least once in the future to confirm that TMA or TGA have not appeared.

Prediction of autoimmune thyroid disease (AITD):

The detection of TMA and/or TGA in an asymptomatic individual indicates an increased risk for the subsequent development of AITD that can produce goiter and/or either hypothyroidism (Hashimoto thyroid or atrophic thyroiditis) or hyperthyroidism (Graves disease). In asymptomatic TMA or TGA-positive individuals it is prudent to consider obtaining yearly measurements of TSH using a third generation TSH assay. If the TSH is abnormal, a free (unbound) T4 measurement should be obtained. Because thyroid and gastric autoimmunity frequently coexist, patients with either parietal cell autoantibodies (PCA) or pernicious anemia should be screened for TMA. Likewise, AITD or thyroid autoantibodies are very common in subjects with type 1 diabetes.

Methodology:

TMA and TGA are both detected using an agglutination assay.

University of Florida Diagnostic Reference Laboratories

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