

THIS WEEK in the JOURNAL

ORIGINAL ARTICLE

Screening for Evaluation of the Risk of Down's Syndrome

In a large study comparing different strategies for screening for Down's syndrome, first-trimester combined screening (measurement of nuchal translucency, pregnancy-associated plasma protein A [PAPP-A], and the free beta subunit of human chorionic gonadotropin [β hCG]) at 11 weeks was better than second-trimester quadruple screening (measurement of alpha-fetoprotein, hCG, unconjugated estriol, and inhibin A). Strategies combining first-trimester and second-trimester screening provide high detection rates at acceptable false positive rates. These findings will help guide the choice of screening strategies for Down's syndrome.

SEE P. 2001; EDITORIAL, P. 2068; CME, P. 2097

ORIGINAL ARTICLE

Response of Glioblastomas to EGFR Kinase Inhibitors

A small proportion of glioblastomas respond to gefitinib or erlotinib (tyrosine kinase inhibitors). Some of these responsive tumors have a mutant variant of the epidermal growth factor receptor (EGFR), and some unresponsive tumors lack PTEN, a regulator of the pathway that a mutant EGFR activates. The simultaneous presence in glioblastoma cells of mutant EGFR and PTEN was associated with responsiveness to tyrosine kinase inhibitors.

SEE P. 2012; PERSPECTIVE, P. 1997

ORIGINAL ARTICLE

CPAP for Central Sleep Apnea and Heart Failure

Central sleep apnea is common among patients who have heart failure and increases the risk of death in this setting. In the Canadian Continuous Positive Airway Pressure trial, continuous positive airway pressure (CPAP) therapy administered to patients with heart failure was found to reduce the severity of central sleep apnea and increase the left ventricular ejection fraction, but did not improve survival.

SEE P. 2025; EDITORIAL, P. 2070; CME, P. 2099

ORIGINAL ARTICLE

Obstructive Sleep Apnea as a Risk Factor for Stroke and Death

Obstructive sleep apnea is a form of sleep-disordered breathing in which the upper airway closes repeatedly during sleep. In an observational cohort study, the risk of stroke or death from any cause was significantly increased among patients with sleep apnea, independent of other cardiovascular risk factors. More severe sleep apnea was associated with greater risk.

SEE P. 2034; EDITORIAL, P. 2070

MECHANISMS OF DISEASE

Oxygen-Sensing Mechanisms

This review discusses recent advances in the understanding of the ways in which different organs detect and respond to acute changes in oxygen tension. A group of specialized tissues constitutes an oxygen-sensing system that responds rapidly to even minor changes in oxygen tension.

SEE P. 2042

CASE RECORDS OF THE MASSACHUSETTS GENERAL HOSPITAL

A Girl with a Bullous Skin Eruption and Acute Respiratory Failure

A rash developed in a 10-year-old girl and became confluent and bullous, with sloughing, after she was given carbamazepine for a seizure. Oral and conjunctival sloughing followed, with pancytopenia, gastrointestinal hemorrhage, and respiratory failure. After a protracted course of management in the burn unit, reepithelialization occurred, and the patient recovered fully.

SEE P. 2057; CME, P. 2098

SOUNDING BOARD

Health-Information Altruists

Although we have successfully sequenced the human genome, progress toward defining the genetic basis of disease has been limited by the availability of people willing to share their phenotypic information along with their genotypic information. In this article, the authors offer a proposal to increase participation in genetic studies.

SEE P. 2074

CLINICAL IMPLICATIONS OF BASIC RESEARCH

Of Commensal Bacteria and Immunity

A molecule found on the surface of commensal bacteria is critical to the development of the immune system.

SEE P. 2078