

THIS WEEK in the JOURNAL

ORIGINAL ARTICLE

Recurrent Genomic Event Associated with Autism

The causes of autism are largely unknown. This study establishes that aberrant dosage of a large genomic segment is associated with autism spectrum disorder. Deletion or duplication of the segment, which encompasses 25 known genes, was present in approximately 1% of case subjects and less than 0.1% of unscreened control subjects.

SEE P. 667; EDITORIAL, P. 737

ORIGINAL ARTICLE

B-Cell Depletion with Rituximab in Relapsing–Remitting Multiple Sclerosis

In this phase 2 trial involving 104 patients with relapsing–remitting multiple sclerosis, patients who received rituximab on days 1 and 15 had fewer gadolinium-enhancing lesions on magnetic resonance imaging and fewer relapses during 48 weeks of follow-up than patients who received placebo. Rituximab was associated with more adverse events within 24 hours after the first infusion. The study was too small and short to assess uncommon adverse events or long-term safety.

SEE P. 676; PERSPECTIVE, P. 664

ORIGINAL ARTICLE

Local Delivery of Paclitaxel during Angioplasty

In a randomized trial, 154 patients with femoropopliteal-artery disease who were undergoing angioplasty were assigned to treatment with a paclitaxel-coated angioplasty balloon, an uncoated balloon with paclitaxel dissolved in the contrast medium, or an uncoated balloon without paclitaxel (control treatment). Late lumen loss, restenosis at 6 months, and target-lesion revascularization at 6, 12, and 24 months were significantly reduced with the paclitaxel-coated balloon but not with paclitaxel-containing contrast medium.

SEE P. 689; CME, P. 763

ORIGINAL ARTICLE

Nasal CPAP or Intubation for Very Preterm Infants

In this trial, preterm infants who needed ventilatory assistance were randomly assigned to receive initial treatment with nasal continuous positive pressure (CPAP) ventilation, followed by intubation if their condition deteriorated, or immediate intubation and mechanical

support. Although there were more pneumothoraces and fewer days of mechanical ventilation in the CPAP group, there was no difference in mortality or bronchopulmonary dysplasia between the two groups.

SEE P. 700

BRIEF REPORT

A *GPR54*-Activating Mutation in a Patient with Central Precocious Puberty

GPR54, a G protein–coupled receptor, and its ligand, kisspeptin, constitute an excitatory neuroregulator system for the secretion of gonadotropin-releasing hormone and appear to be important in the onset of puberty. This report describes an autosomal dominant GPR54 mutation, Arg386Pro, in an adopted girl with idiopathic central precocious puberty.

SEE P. 709

MECHANISMS OF DISEASE

Acute Lower Respiratory Tract Infection

Acute lower respiratory tract infections are a major cause of death and disability, yet the mechanisms that make such infections so virulent are not entirely understood. This review emphasizes the roles of inflammation and the response of the innate immune system and explains how these two processes interact to rid the lung of microbes but also how they can bring the elimination of infection in the lung to a perilous climax.

SEE P. 716; CME, P. 762

CASE RECORDS OF THE MASSACHUSETTS GENERAL HOSPITAL

An 18-Month-Old Girl with Neck Contracture

An 18-month-old Chinese girl was seen for management of a life-threatening neck contracture due to burns to the face and torso at 12 months of age. The wounds had healed with contractures that resulted in fused nares, microstomia, and sleep apnea. Tracheal intubation was impossible because of the fused nares and microstomia, and scar tissue prevented tracheotomy.

SEE P. 729; CME, P. 761

CLINICAL IMPLICATIONS OF BASIC RESEARCH

Variation in the Human Genome

Large-scale genomic deletions, duplications, and inversions represent a major source of variation among persons; thus, new approaches to probing disease susceptibility are warranted.

SEE P. 740