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Nota di contenuto	CHAPTER 1 Overview and Sensory Issues SANDRA L. H. DAVENPORT, M. D. AND MARGARET A. HEFNER, M.S. ; Humans have receptors for five "input" senses, which allow access to external stimuli, namely vision, hearing, smell, touch, and taste. In addition, the body has other receptors that allow the body to recognize its position in space and in relation to itself, namely vestibular and proprioceptive senses. In CHARGE syndrome, all seven of these senses may be affected. Vision, hearing, smell, and balance CHAPTER 2 The Eye and Vision LEA HYVARINEN, M.D. ; One of the cardinal features of CHARGE syndrome is ocular coloboma, which is a defect in the development of the eye between weeks 5 and 7 of gestation. Colobomas cause defects in the visual field and often lower than normal visual acuity. EARLY DEVELOPMENT OF THE RETINA The optic nerves and retina of the eyes are extensions of the brain. First, finger-like structures develop, one on each side of the neural tube (the embryonic precursor to the cen CHAPTER 3 Hearing JAMES W. THELIN, PH.D. ; AUDIOLOGIC ISSUES In CHARGE syndrome, hearing, balance, and mobility are related to patterns of structural anomalies of the auditory, vestibular, and visual systems. Some patterns of anomalies are common across individuals,

but the group of deficits for an individual can be unique. It is of great value to an individual with CHARGE if the persons who provide care or services understand an individual's specific deficits and challenges. The purpose of the f

CHAPTER 4 Overview of Balance and the Vestibular System CLAES MOLLER, M.D., PH.D. ; The ability to walk upright on two legs and keep equilibrium is dependent on the integrity of a complex system consisting of three major receptor organs: the vestibular, the visual, and the somatosensory systems (see Color Plate 6). The impulses from the vestibular (labyrinth) part of the inner ear, the eyes, and the stimuli from skin muscles, tendons, and joints (somatosensory) are so harmoniously balanced that,

CHAPTER 5 Consequences of Vestibular Dysfunction DAVID M. BROWN, M.A., D.SC. (HON) ; Of all the many sensory impairments associated with CHARGE syndrome, absence of, or significant damage to, the vestibular sense is perhaps the most far-reaching in its implications, the least understood, and often the most overlooked. If people have any idea at all what the term "vestibular sense" means, they usually think that it relates to only the development of good balance, and so they might assume that vest

CHAPTER 6 Balance and Mobility JAMES W. THELIN, PH.D., SARAH E. CURTIS, AU.D., JILL FUSSNER MADDOX, AU.D., AND LORI S. TRAVIS, AU. D.; The anomalies that typically are present in children with CHARGE syndrome often result in the delay of the development of balance and mobility. The causes for these delays may include the following factors: Bilateral aplasia or dysplasia of the semicircular canals of the vestibular mechanism Visual impairment due to ocular colobomas or other anomalies Orthopedic

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## Sommario/riassunto

CHARGE syndrome affects approximately 1:10,000 births worldwide, is extremely complex, and has varied medical and physical manifestations. It was first described in 1979, named in 1981, and only as recently as 2004 was a gene for CHARGE found. In addition to a host of other conditions, most individuals have communication-related problems, including breathing, swallowing, hearing and balance, and speech. Each of the editors is established as an expert on CHARGE syndrome and has received the highest award bestowed by the CHARGE Syndrome Foundation, the Star in CHARGE, and represent four differences

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