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Nota di contenuto	CONTENTS; AUTHORS' APPOINTMENTS; FOREWORD; 1 DR ANGELMAN'S SYNDROME Bernard Dan; Original report of Angelman syndrome; Dr Harry Angelman (Fig. 1.2); From puppet children to Angelman syndrome; Illustrations and derivations; From patient descriptions to diagnostic criteria; Angelman syndrome as a model; Conclusion; 2 NATURAL HISTORY Karine Pelc and Bernard Dan; Antenatal and perinatal period; Neonatal period; Infancy; Childhood; Adolescence; Adulthood; Conclusion; 3 A PERSONAL ACCOUNT Patrick Haverbeke; How an angel changed our life; Early problems with integration Options for Henri's educationThe road to diagnosis; Tools for communication; The difficult choice: putting Henri in a residential institution; Reorganising our family life; Time is passing: Henri becomes an adult; Henri, ambassador across the world; 4 MEDICAL GENETICS Bernard Dan; Chromosome 15q11-q13 deletion; Uniparental disomy; UBE3A mutation; Imprinting defect; Chromosomal aberration; Other identified genetic abnormalities; Genetic testing; Genetic counselling; Prenatal diagnosis; Risks associated with in vitro fertilisation; Conclusion; 5 MOLECULAR BIOLOGY Bernard Dan; Genomic imprinting Genomic imprintingUBE3A gene expression; Ubiquitination; UBE3A gene expression; Ubiquitination; An integrative hypothesis; An integrative hypothesis; Possible targets for management; Possible targets for management; Conclusion; Conclusion; 6 DIFFERENTIAL DIAGNOSIS Bernard Dan; Cerebral palsy; Autistic spectrum; Lennox-Gastaut syndrome; Rett syndrome; Untreated phenylketonuria;

Methylenetetrahydrofolate reductase deficiency; ATR-X syndrome; Gurrieri syndrome; Mowat-Wilson syndrome; Chromosomal abnormalities; Mosaic imprinting defects of 15q11-q13; Prader-Willi syndrome
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12 SLEEP Bernard Dan and Stewart G. Boyd

Sommario/riassunto

A comprehensive review of clinical and genetic issues, natural history, possible pathophysiological pathways, specific clinical problems (motor impairment, behaviour, learning difficulties, communication, sleep, epilepsy), clinical neurophysiology, neuropathology, rehabilitation and basic research in the field of Angelman syndrome.