

1. Record Nr.	UNINA9910813557103321
Autore	Dan Bernard
Titolo	Angelman syndrome [[electronic resource] /] / Bernard Dan
Pubbl/distr/stampa	London, : Mac Keith Press, 2008
ISBN	1-898683-83-2
Descrizione fisica	1 online resource (192 p.)
Collana	Clinics in developmental medicine ; ; no. 177
Soggetti	Angelman syndrome
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	<p>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;</p>

Methylenetetrahydrofolate reductase deficiency; ATR-X syndrome; Gurrieri syndrome; Mowat-Wilson syndrome; Chromosomal abnormalities; Mosaic imprinting defects of 15q11-q13; Prader-Willi syndrome

Inverted duplication of chromosome 15 Conclusion; 7 BEHAVIOUR

Bernard Dan; Happy disposition; Hyperactivity and impulsivity; Stereotypic behaviours; Autistic features; Social interaction; Behavioural adaptability; Interest profile; Oral behaviours; Conclusion; 8

PERCEPTION AND COGNITION Bernard Dan; Visual perception; Auditory and language perception; Somatosensory perception; Pain; Other perceptual modalities; Attention; 'Mental age'; Memory; Emotion; Conclusion; 9 COMMUNICATION Bernard Dan; General context of communication; Receptive verbal language; Expressive verbal language

Expressive non-verbal language Conclusion; 10 MOVEMENT AND POSTURAL CONTROL Bernard Dan and Guy Cheron; Alternative vs immature motor development; Early motor development; Later motor development; Motor signs; Balance; Oromotor function; Hand function; Walking; Other movements; Tremor vs myoclonus; Pathophysiology; Physical management; Pharmacological management; Conclusion; 11

EPILEPSY Bernard Dan and Stewart G. Boyd; Natural history of the seizure disorder; Seizure types; Epileptic syndrome; Convulsive status epilepticus; Non-convulsive status epilepticus; Management; Conclusion

Conclusion

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.
