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Titolo	Imprinting disorders associated with molecular changes on chromosome 11p15 / / Rosanna Weksberg
Pubbl/distr/stampa	London, : Henry Stewart Talks, 2014
Descrizione fisica	1 online resource (1 streaming video file (37 min.) : color, sound)
Collana	Molecular genetics of human disease, , 2056-452X
Soggetti	Epigenetics Genetic disorders Genomic imprinting Medical genetics Molecular genetics Beckwith-Wiedemann Syndrome Chromosomes, Human, Pair 11 - genetics Epigenesis, Genetic Genomic Imprinting - genetics Molecular Diagnostic Techniques Mosaicism Silver-Russell Syndrome
Lingua di pubblicazione	Inglese
Formato	Videoregistrazione
Livello bibliografico	Monografia
Note generali	Animated audio-visual presentation with synchronized narration. Title from title frames.
Nota di contenuto	Contents: What is epigenetics? -- Epigenetic regulation forms the molecular basis for genomic imprinting -- What is genomic imprinting -- Imprinted genes in early development -- Genomic organization of imprinted genes -- Imprinting center -- Imprinted domain 1 on chromosome 11p15.5 -- Complexity of imprinted clusters -- Beckwith-Wiedemann syndrome (BWS) -- BWS: a complex, clinically heterogeneous disorder -- Molecular basis of BWS -- Etiology of the Beckwith-Wiedemann syndrome -- Molecular alterations associated with BWS -- Frequency of molecular alterations in BWS -- The risks to subfertile/ART treated parents -- Subfertility/assisted reproductive

technologies (ART) -- BWS molecular defects -- Multiple phenotypes associated with somatic mosaicism for 11p15 UPD -- Somatic mosaicism -- Isolated hemihyperplasia -- High level constitutional UPD -- BWS molecular testing strategies -- Laboratory testing for BWS -- MS-MLPA -- Expected methylation results from MS-MLPA -- MLPA molecular testing for BWS -- Cancer risk & surveillance -- Prenatal testing options -- Current challenges in BWS molecular testing -- Frequency of CNVs according to methylation pattern -- Monozygotic twins and BWS -- Isolated hemihyperplasia -- Current challenges in chromosome 11p15 molecular testing -- Russell-Silver syndrome (RSS -- Chromosomal regions associated with RSS -- Targeted assays of multiple imprinted loci-BWS -- Different combinations of epigenetic alterations associated with variations in clinical phenotype -- Imprint deregulation causing disease in humans.