

1. Record Nr.	UNINA9910298438203321
Titolo	Polyglutamine Disorders [[electronic resource] /] / edited by Clévio Nóbrega, Luís Pereira de Almeida
Pubbl/distr/stampa	Cham : , : Springer International Publishing : , : Imprint : Springer, , 2018
ISBN	3-319-71779-0
Edizione	[1st ed. 2018.]
Descrizione fisica	1 online resource (469 pages) : illustrations, tables
Collana	Advances in Experimental Medicine and Biology, , 0065-2598 ; ; 1049
Disciplina	616.80442
Soggetti	Neurosciences Neurology Medical genetics Neurology Gene Function
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references at the end of each chapters and index.
Sommario/riassunto	This book provides a cutting-edge review of polyglutamine disorders. It primarily focuses on two main aspects: (1) the mechanisms underlying the pathologies' development and progression, and (2) the therapeutic strategies that are currently being explored to stop or delay disease progression. Polyglutamine (polyQ) disorders are a group of inherited neurodegenerative diseases with a fatal outcome that are caused by an abnormal expansion of a coding trinucleotide repeat (CAG), which is then translated in an abnormal protein with an elongated glutamine tract (Q). To date, nine polyQ disorders have been identified and described: dentatorubral-pallidoluysian atrophy (DRPLA); Huntington's disease (HD); spinal-bulbar muscular atrophy (SBMA); and six spinocerebellar ataxias (SCA 1, 2, 3, 6, 7, and 17). The genetic basis of polyQ disorders is well established and described, and despite important advances that have opened up the possibility of generating genetic models of the disease, the mechanisms that cause neuronal degeneration are still largely unknown and there is currently no

treatment available for these disorders. Further, it is believed that the different polyQ may share some mechanisms and pathways contributing to neurodegeneration and disease progression.
