

1. Record Nr.	UNISA996392965803316
Autore	Rodriguez Alfonso <1526-1616.>
Titolo	A treatise of mentall prayer [[electronic resource]] : With another of the presence of God. Composed by the R. Fa. Alfonsus Rodriguez, of the Society of Iesus. And translated out of the Spanish, into English
Pubbl/distr/stampa	[Saint-Omer, : printed at the English College Press] Permissu superiorum, 1627
Descrizione fisica	[16], 303, [1] p
Altri autori (Persone)	MatthewTobie, Sir, <1577-1655.> WilsonJohn <ca. 1575-ca. 1645?>
Soggetti	Prayer Christian life - Catholic authors
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	A translation, by Sir Tobie Matthew, of selections from: Ejercicio de perfeccioìn y virtudes cristianas. Editor's dedication signed: I.W., i.e. John Wilson. Identification of printer from STC. Identified as item no. 7, reel 2033, unit 64, of the UMI microfilm reel guide "Early English books 1475-1640". Reproduction of original in the Bodleian Library.
Sommario/riassunto	eebo-0014

2. Record Nr.	UNINA9910566479203321
Autore	Millan Jose
Titolo	Genetic Testing for Rare Diseases
Pubbl/distr/stampa	Basel, : MDPI - Multidisciplinary Digital Publishing Institute, 2022
Descrizione fisica	1 online resource (144 p.)
Soggetti	Medicine and Nursing
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Sommario/riassunto	<p>Rare diseases, or orphan diseases, are those that individually affect a small number of patients, but taken together affect over 300 million people worldwide. They are characterized by their etiological, diagnostic and evolutionary complexity, important morbi-mortality, with high levels of disability that entail and hinder the development of a normal vital subject, not only in those who suffer from them, but also their families; therefore, a comprehensive social health approach is necessary to address this problem. About 80% of rare diseases have a genetic origin, mainly monogenic; thus, genetic testing is mandatory for the confirmation of clinical diagnostics and to ensure correct genetic counseling. Next-generation sequencing (NGS) has enabled a revolution in genetic diseases, specially in rare diseases. However, their complexity makes diagnoses difficult even with the advent of NGS. In this Special Issue, we present several examples of the complexity of genetic diagnosis for most of these diseases and the consequences that genetic testing implies for genetic counseling. There are examples of the genetic heterogeneity of hearing loss, some metabolic and lisosomal disorders, ataxia, Prader-Willi syndrome, and three comprehensive reviews on syndromic retinal dystrophies, the complexity of the molecular diagnosis of neuromuscular disorders, and the value of genetic counseling before and after a genetic test.</p>