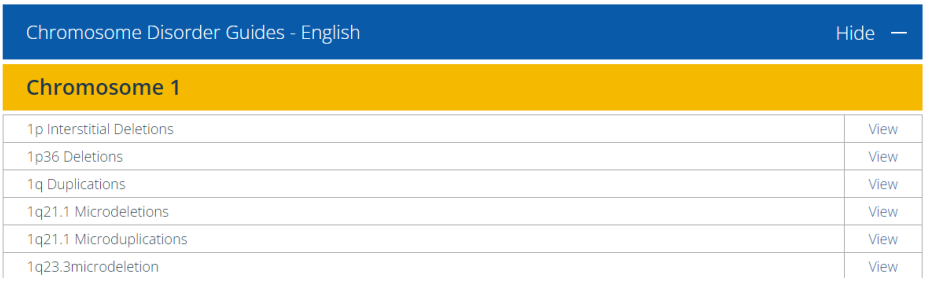
Unique- Rare Chromosome and Single Gene Disorders Website - RareChromo.org

- Unique is a UK-based charity organization established in1984 to support and provide information for families and medical genetics professionals about specific rare chromosomal and single gene disorders. The website also provides resources for families and a genotype database documenting the clinical history of more than 27,000 members of all ages affected by various disorders.
- Select disorder guides are available in multiple languages including Arabic, Chinese, Danish, Deutsch, French, Georgian, Greek, Korean, Lithuanian, Malay, Polish, Portuguese, Romana, Russian, Spanish, Swedish, Turkish, Ukrainian and Vietnamese.

Disorder Guide Booklets- [Disorder Guides - Unique]

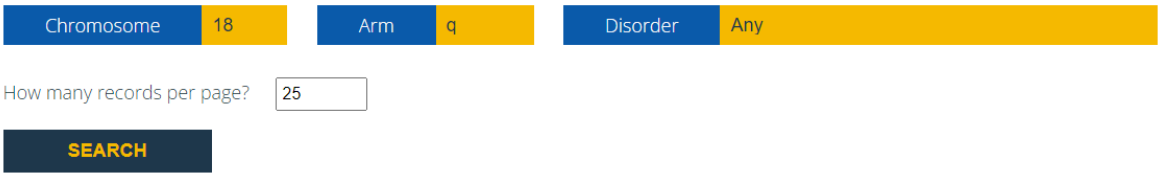


Disorder Database - [Registered Chromosome Disorders, Copy Number Variants & Gene Variants - Unique]

- The database is searchable and provides anonymized phenotypic feature description associated with specific chromosome disorders

- To search for specific chromosomal disorders dependent on the chromosome number, arm designation or disorder type, click the ‘any’ to select your inquiry and click on the

search button. In the example below, a chromosomal disorder in 18q is being researched.



Results

The following list shows the chromosome conditions registered with Unique which match your search criteria. You may click on one to see specific karyotypes, FISH or arrayCGH analysis or DNA sequencing results registered against that condition.

Found 179 results. Page 1 of 8 Next

Chromosomes	Number of Members
10p Duplication;18q Deletion	3
10p;18q (Balanced Translocation)	1
10q Deletion;18q Duplication	1
10q Duplication;17p Duplication;18q Deletion	1

- The inquiry above provided 179 search inquiries, which are categorized into various sections, the sub-section below lists 3 cases that resulted in 10q duplication and 18q loss and provides the ISCN nomenclature for each case. It also provides a code number which can be utilized to request additional information from the Unique organization.

Chromosomes		Number of Members
10p Duplication;18q Deletion		
Genotype	**Code Number**	
46,XX,der(18)t(10;18)(p13;q21.33)pat	19116	3
arr[hg19] 10p15.3p12.2(102539-24060390)x3, 18q22.1q23(61757751-78012829)x1	55484	
arr 10p15.3p15.1(153585_5261427)x3, 18q22.1q23(63948743_78013619)x1	64213	

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Created: March 31st, 2024