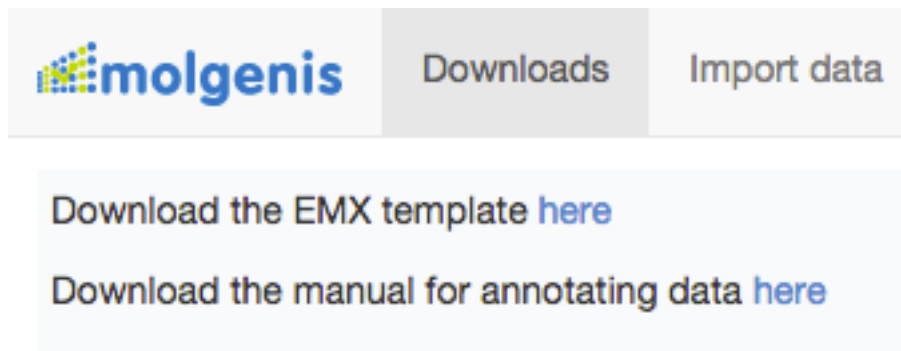


# Manual for Molgenis 5gpm Annotation

1: You start with the file that was downloaded from Cartagena

	A	O	Y	AG	AH	CQ
1	Overerving	Omim Link	Gene (gene)	cDNA (cNomen)	Protein (pNomen)	HGMD Disease
2	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	AIPL1	c.937G>T	p.A313S	Leber congenital
3	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	ALMS1	c.10269G>C	p.K3423N	
4	Comp/Dom	<a href="http://m.nih.gov/omim/17">m.nih.gov/omim/17</a>	ALPL	c.1381G>A	p.V461I	
5	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	ANKRD11	c.6497T>G	p.M2166R	
6	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	ARID1B	c.2419C>T	p.P807S	
7	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	ATIC	c.281A>G	p.N94S	
8	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	ATN1	c.1500G>T	p.Q500H	
9	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	ATN1	c.2260G>A	p.V754I	
10	Comp/Dom	<a href="http://m.nih.gov/omim/11">m.nih.gov/omim/11</a>	BDNF	c.*356-1delA		
11	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	BRAT1	c.1636G>T	p.V546L	
12	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	CACNA1A	c.2749C>T	p.P917S	
13	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	CBS	c.833T>C	p.I278T	Homocystinuria
14	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	COG6	c.1963C>A	p.L655I	
15	Comp/Dom	<a href="http://m.nih.gov/omim/12">m.nih.gov/omim/12</a>	COL18A1	c.1771G>A	p.G591S	
16	Comp/Dom	<a href="http://m.nih.gov/omim/12">m.nih.gov/omim/12</a>	COL4A1	c.367C>T	p.P123S	
17	Comp/Dom	<a href="http://m.nih.gov/omim/12">m.nih.gov/omim/12</a>	COL4A2	c.3448C>A	p.Q1150K	Haemorrhagic
18	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	COQ6	c.62C>T	p.S21F	
19	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	CPA6	c.192+4A>C		
20	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	CREBBP	c.6685G>A	p.G2229S	
21	Homo	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	CREBBP	c.3370-4delT		
22	Comp/Dom	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	EMG1	c.126-1T>C		
23	Comp	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	EPG5	c.5954G>A	p.R1985Q	
24	Comp	<a href="http://m.nih.gov/omim/61">m.nih.gov/omim/61</a>	EPG5	c.2063T>C	p.F688S	
25	Comp/Dom	<a href="http://m.nih.gov/omim/13">m.nih.gov/omim/13</a>	ERCC3	c.1027+3A>G		
26	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	ERCC6	c.2825C>T	p.T942M	
27	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	FKRP	c.1177G>A	p.V393I	
28	Homo	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	GAA	c.858+5_858+6ins		
29	Comp/Dom		GCM2	c.1060A>G	p.M354V	
30	Comp/Dom	<a href="http://m.nih.gov/omim/13">m.nih.gov/omim/13</a>	GNAS	c.1556C>A	p.P519Q	
31	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	GPR98	c.6095C>T	p.A2032V	
32	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	GPR98	c.10039T>C	p.F3347L	
33	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	GPR98	c.18746T>G	p.L6249R	
34	Homo	<a href="http://m.nih.gov/omim/30">m.nih.gov/omim/30</a>	GRIA3	c.380dupG		
35	Homo	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	KAT6B	c.3310_3312delGA	p.E1097del	
36	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	LRP2	c.10804G>A	p.A3602T	
37	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	LRP2	c.7918A>G	p.I2640V	
38	Comp	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	LRP2	c.5293G>A	p.V1765M	
39	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	LYST	c.11268-5delT		
40	Comp/Dom	<a href="http://m.nih.gov/omim/60">m.nih.gov/omim/60</a>	MANBA	c.2351C>G	p.P784R	

2: Download the Molgenis EMX template from the [www.molgenis64.target.rug.nl](http://www.molgenis64.target.rug.nl) homepage (after logging in downloads tab)



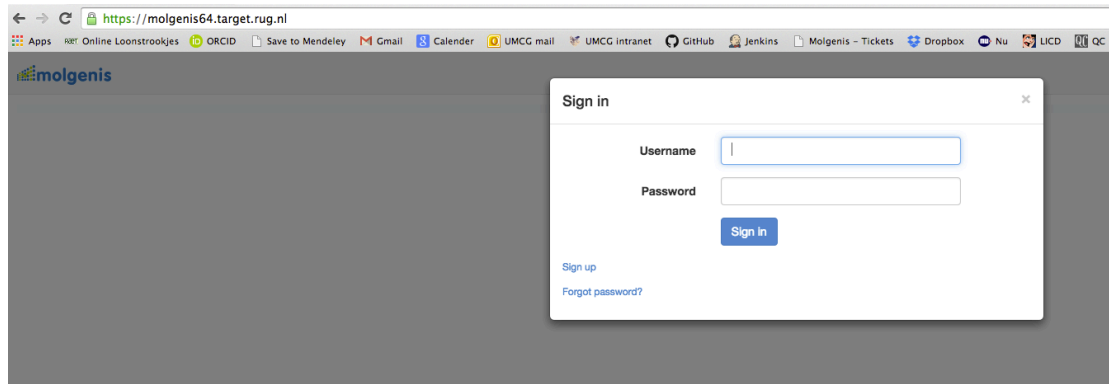
3: Copy paste your Cartagenia data into the EMX template, starting in the second column

1	Identifier	Overerving	#CHROM	POS	Stop	REF	Allele_1	Allele_2	ALT	Transcript	Classification	Confirmed Class	dbSNP	OMIM_refs	OMIM	AF_Allele_1	AF_Allele_2	AD_Allele_1	AD_Allele_2
2	1	Comp/Dom	16	77323312	77323312	C	C	G	G	NM_19935.2			rs11643553	607512	<a href="http://omim.org/entry/607512">http://omim.org/entry/607512</a>	0,5		28	13
3	2	Dom	18	12337348	12337348	C	C	T	T	NM_00679.2			rs13946978	604581	<a href="http://omim.org/entry/604581">http://omim.org/entry/604581</a>	0,5		21	20
4	3	Comp/Dom	11	27679011	27679011	T	T	.	.	NM_17073.5			rs3838785	113505	<a href="http://omim.org/entry/113505">http://omim.org/entry/113505</a>	0,5		19	15
5	4	Comp/Dom	11	117280516	117280516	A	A	C	C	NM_01495.6.4			614848		<a href="http://omim.org/entry/614848">http://omim.org/entry/614848</a>	0,5		17	13
6	5	Dom	3	190106074	190106074	G	G	C	C	NM_00658.0.3			rs76555381	603959	<a href="http://omim.org/entry/603959">http://omim.org/entry/603959</a>	0,5		15	11
7	6	Comp/Dom	16	3820723	3820723	T	T	C	C	NM_00438.0.2			rs14324768	600140	<a href="http://omim.org/entry/600140">http://omim.org/entry/600140</a>	0,5		22	17
8	7	Dom	9	131708133	131708133	T	T	C	C	NM_01490.8.3			rs14763097	7	<a href="http://omim.org/entry/7">http://omim.org/entry/7</a>	0,5		25	16
9	8	Homo	12	7080212	7080212	T	C	C	C	NM_00633.1.7			rs11428482	611531	<a href="http://omim.org/entry/611531">http://omim.org/entry/611531</a>	1	1	31	31
10	9	Comp/Dom	16	53860052	53860052	G	G	A	A	NM_00108.0432.2			rs79206939	610966	<a href="http://omim.org/entry/610966">http://omim.org/entry/610966</a>	0,5		29	24
11	10	Homo	17	78081526	78081526	.	GCAGCGG	GCAGCGG	GCAGCGG	NM_00015.2.3			rs3071247	606800	<a href="http://omim.org/entry/606800">http://omim.org/entry/606800</a>	1	1	30	30

4: The name of the tab your Cartagenia data is in should be the same as the name in the entity column in the attribute sheet.

IMPORTANT!! Every row needs an unique identifier, so fill in 1, 2, 3, etc... in the identifier column.

5: Go to molgenis64.target.rug.nl and log in with your password and username



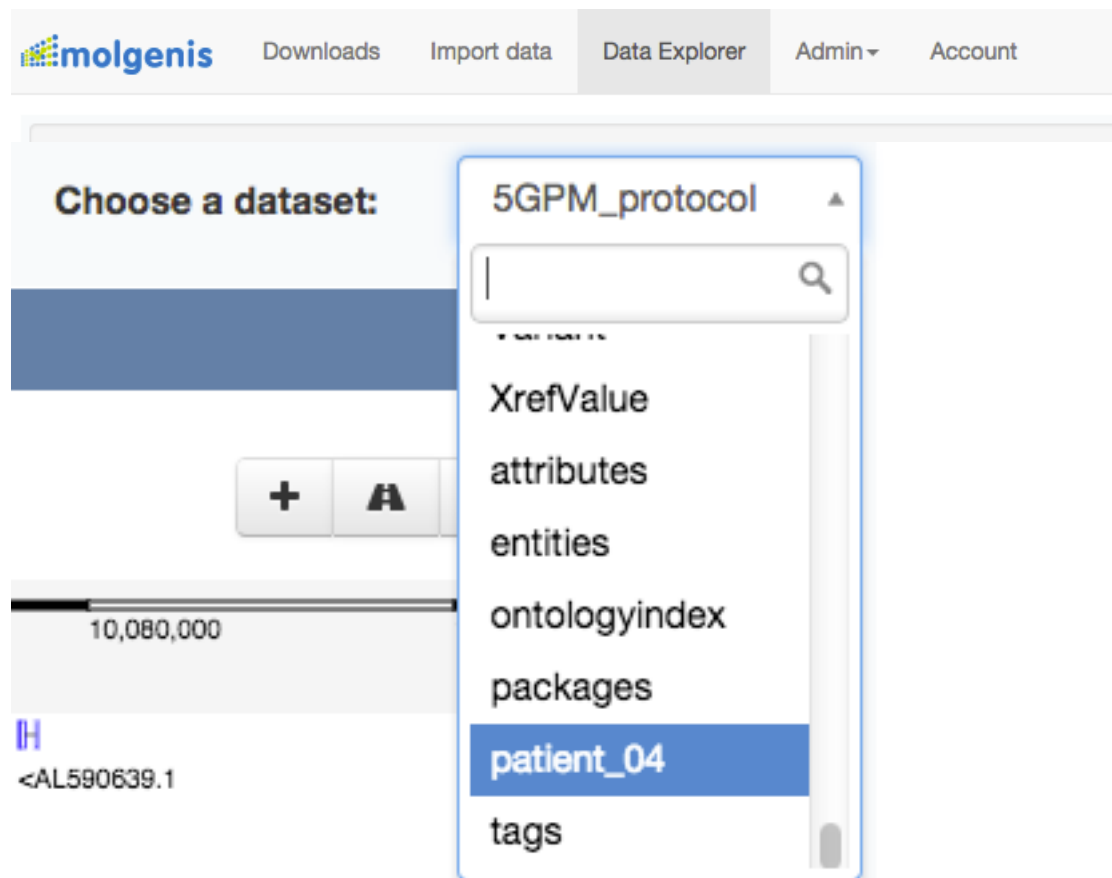
6: navigate to the import data tab and select a file to upload

The screenshot shows the Molgenis web interface for data import. At the top, there is a navigation bar with the Molgenis logo and links for Downloads, Import data (selected), Data Explorer, Admin, and Account. Below this is a progress indicator with four steps: 1 Upload file (active), 2 Options, 3 Validation, and 4 Result. The main content area is titled 'Upload a file' and contains a 'Select a file...' button. A file explorer window is open, showing the contents of a folder named '5GPM\_EMX\_Template'. The file explorer lists two files: 'patient\_04\_raw.xlsx' and '5gpm\_emx\_template'. At the bottom of the interface, there are three buttons: '← Previous', 'Restart', and 'Next →'.

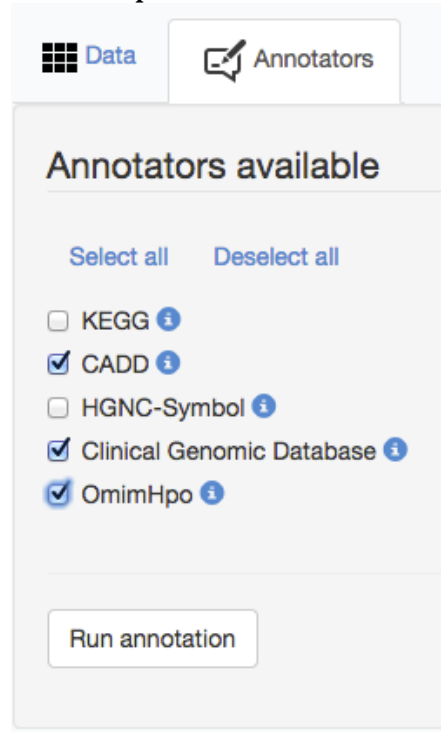
7: Go through steps by pressing next and it should validate and import your data

The screenshot shows the Molgenis web interface after the data import process. The progress indicator at the top now shows '4 Result' as the active step. The main content area displays a green banner with the text 'Import success'. Below this banner, a white box contains the text 'Imported 80 patient\_04 entities.'

8: Navigate to the data explorer tab and select your patient in the dropdown



9: Go to the annotator module (in the dataexplorer), select CGD and OmimHpo and CADD and click run annotation



10. After the success message you can browse and filter your data, or download it as a csv

The screenshot shows a search bar containing the text "Seizures" with a clear (X) and search (Q) icon to its right. Below the search bar is a pagination control with a left arrow, page numbers 1, 2, 3, 4, and a right arrow. The number 1 is highlighted in blue. To the right of the pagination control, the text "80 items found" is displayed. At the bottom right of the interface is a button labeled "Download as CSV".