

Supplementary Table 1. TLR search terms

Search Term Group	Number	Search Terms	TLR Results	TLR Update Results (Embase/MEDLINE)
Cerebrotendinous xanthomatosis	1	"cerebrotendinous xanthomatosis".tw.	1368	765/630
	2	(cerebr* adj (cholesterosis or cholesterinosis)).tw.	3	1/1
	3	"van Bogaert-Scherer-Epstein".tw.	11	8/5
	4	"Sterol 27-hydroxylase deficiency".tw.	32	17/15
	5	(Cholestanol\$ adj (lipidosis or storage)).tw.	6	2/3
	6	(Thibaut\$ syndrome).tw.	3	1/1
	7	Or/1-6	1383	772/638
Exclusion terms	8	exp animals/ not exp humans/	9168178	4332509/4514185
	9	("conference abstract" or "conference review" or comment or letter or editorial).pt.	6236002	4848266/1671196
	10	8 or 9	15034246	8843639/6123276
Combined	11	7 not 10	1178	610/577
TLR limits	12	Remove duplicates from 11	664	-
TLR update limits	13	Limit 11 to dd=20180510-20181113 (Embase; 10 th May 2018–13 th November 2018)	-	8 (Embase)
		Limit 11 to yr=2018-current (MEDLINE; 1 st January 2018– 13 th November 2018)	-	25 (MEDLINE)

TLR searches (rows 1–12) run on 18th May 2018. Embase 1974 to 2018 May 17, Ovid MEDLINE(R) Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid

MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to Present. TLR update searches (rows 1–11 and row 13) run on 13th November 2018. Embase 1974 to 2018 November 12,

Ovid MEDLINE(R) Epub Ahead of Print, In-Process & Other Non-Indexed Citations, Ovid MEDLINE(R) Daily and Ovid MEDLINE(R) 1946 to 2018 November 12. TLR: targeted

literature review.