Appendix 1 search stratergies/Bilaga 1 sökstrategier

Cinahl via EBSCO 19 August 2015
Title: Chromosomal microarray analysis and next generation sequencing for prenatal diagnosis

	Search terms	Items found
Prenatal diagnosis		
1.	MH "Prenatal Diagnosis" OR MH "Genetic Screening"	9416
2.	TI ((prenatal OR "pre-natal" OR antenatal) n4 (diagnos* OR detect* OR predict* OR prognos* OR test OR tests OR testing OR screen*)) OR amniocentesis OR chorionic Villi sampling OR "nuchal translucency measurement" OR "maternal serum screening test") OR AB ((prenatal OR "pre-natal" OR antenatal) n3 (diagnos* OR detect* OR predict* OR prognos* OR test OR tests OR testing OR screen*)) OR amniocentesis OR chorionic Villi sampling OR "nuchal translucency measurement" OR "maternal serum screening test")	3754
3.	TI (NIPT OR NIPD OR NIDT OR NIFTY OR "cell-free dna" OR "fetal dna"OR "cff dna" OR cffdna OR "cf dna" OR cfdna OR "f dna"OR fdna OR "ff dna" OR ffdna OR "maternal serum screening test" OR (maternal w2 plasm*) OR ((noninvasiv* OR "non-invasive") n3 (prenatal OR "pre-natal" OR antenatal OR diagnos* OR detect* OR predict* OR prognost* OR test OR tests OR testing)) OR AB (NIPT OR NIPD OR NIDT OR NIFTY OR "cell-free dna" OR "fetal dna"OR "cff dna" OR cffdna OR "cf dna" OR cfdna OR "f dna"OR fdna OR "ff dna" OR ffdna OR "maternal serum screening test" OR (maternal w2 plasm*) OR ((noninvasiv* OR "non-invasive") n4 (prenatal OR "pre-natal" OR antenatal OR diagnos* OR detect* OR predict* OR prognost* OR test OR tests OR testing))	2291
4.	1 OR 2 OR 3	*13634*
Chromosomal microarray analysis and next generation sequencing		
5.	MH "Nucleic Acid Hybridization+" OR MH "Microarray Analysis+"	2269
6.	TI ("comparative genomic hybridi?ation*" OR "comparative genome hybridi?ation*" OR cgh OR ((chromosom* OR DNA OR cytogenetic OR genomic OR "genome-wide" OR snps OR snp OR "single nucleotide polymorphism*") n4 (array* OR microarray* OR "micro-array*")) OR acgh OR "oligo-aCGH" OR "whole sequenc*" OR "complete sequenc*" OR "entire sequenc*" OR "full sequenc*" OR ((whole OR complete OR entire) n4 (genome OR genomic)) OR "genome sequenc*" OR "genomic sequenc*" OR "massive parallel sequenc*" OR "high-through sequenc*"OR ngs OR nps OR (("next generation" OR exome) n4 sequenc*) OR AB ("comparative genomic hybridi?ation*" OR "comparative genome hybridi?ation*" OR cgh OR ((chromosom* OR DNA OR cytogenetic OR genomic OR "genome-wide" OR snps OR snp OR "single nucleotide polymorphism*") n4 (array* OR microarray* OR "micro-array*")) OR acgh OR "oligo-aCGH" OR "whole sequenc*" OR "complete sequenc*" OR "entire sequenc*" OR "full sequenc*" OR ((whole OR complete OR entire) n4 (genome OR genomic)) OR "genome sequenc*" OR "genomic sequenc*" OR "parallel sequenc*" OR "high-through sequenc*"OR ngs OR nps OR (("next generation" OR exome) n4 sequenc*)	3918
7.	5 OR 6	6013
Combined sets		
8.	**4 AND 7**	**320**
Limits		
9.	**Limiters** - Peer Reviewed	
10.	**8 AND 9**	**308**

The search result, usually found at the end of the documentation, forms the list of abstracts.
AB = Abstract
AU = Author
DE = Term from the thesaurus
MH = Term from the "Cinahl Headings" thesaurus
MM = Major Concept
TI = Title
TX = All Text. Performs a keyword search of all the   database's searchable fields

ZC = Methodology Index
* = Truncation
" " = Citation Marks; searches for an exact phrase

Cochrane Library via Wiley 18 August 2015 (CDSR, DARE & CENTRAL)
Title: Chromosomal microarray analysis and next generation sequencing for prenatal diagnosis

Search terms		Items found
Prenatal diagnosis		
1.	[mh "prenatal diagnosis"]	971
2.	(prenatal or "pre-natal" or antenatal) and (diagnos* or detect* or predict* or prognos* or test or tests or testing or screen*) or amniocentesis or " chorionic Villi sampling" or "nuchal translucency measurement":ti,ab,kw (Word variations have been searched)	3231
3.	*1 OR 2*	*3390*
4.	nipt or nipd or nidt or nifty or cell next/1 free next/1 dna or cell next/1 free next/1 fetal next/1 dna or "cff dna" or "cf dna" or "ff dna" or "f dna" or maternal plasm*:ti,ab,kw OR (noninvasiv* OR non NEXT/1 invasiv*) AND (prenatal OR pre next/1 natal OR diagnos* OR detect*):ti,ab,kw OR [mh "prenatal diagnosis"]	3908
5.	*3 OR 4*	*6239*
Chromosomal microarray analysis and next generation sequencing		
6.	[mh "comparative genomic hybridization"] or [mh "microarray analysis"] or [mh "genome-wide association study"] or [mh "genotyping techniques"]	401
7.	("comparative genomic" next hybridi?ation*) or ("comparative genome" next hybridi?ation*) or cgh or bac or ("bacterial artificial" next chromosome*):ti,ab,kw or ((chromosomal* or dna or cytogenetic or genomic or 'genome-wide' or snps or "single nucleotide polymorphisms") near/4 (array* or microarray* or 'micro-array' or 'micro-arrays')) or acgh or oligo-aCGH or ((whole or complete or entire or full or parallel or 'next generation') near/4 sequenc*):ti,ab,kw (Word variations have been searched)	475
8.	*6 OR 7*	*1252*
Combined sets		
9.	5 AND 8	**CDSR/3 DARE/3 Central/18 CRM/3 HTA/0 EED/0**

The search result, usually found at the end of the documentation, forms the list of abstracts.

CDSR = Cochrane Database of Systematic Review
CENTRAL = Cochrane Central Register of Controlled Trials, "trials"
CRM = Method Studies
DARE = Database Abstracts of Reviews of Effects, "other reviews"
EED = Economic Evaluations
HTA = Health Technology Assessments

Embase via Elsevier 18 August 2015

Title: Chromosomal microarray analysis and next generation sequencing for prenatal diagnosis

Search terms		Items found
Prenatal diagnosis		
1.	'prenatal diagnosis'/exp/mj	41437
2.	((prenatal OR 'pre natal' OR antenatal) NEAR/4 (diagnos* OR detect* OR predict* OR prognos* OR test OR tests OR testing OR screen*)):ti,ab	40616
3.	1 OR 2	65406
4.	(nipt:ab,ti OR nipd:ab,ti OR nidt:ab,ti OR nifty:ab,ti OR 'cell-free dna':ab,ti OR 'fetal dna':ab,ti OR 'cff dna':ab,ti OR cffdna:ab,ti OR 'cf dna':ab,ti OR cfdna:ab,ti OR 'ff dna':ab,ti OR ffdna:ab,ti OR 'f dna':ab,ti OR fdna:ab,ti OR ('maternal serum screening' NEXT/1 test*):ab,ti OR (noninvasiv* OR non NEXT/1 invasiv*)) AND ('prenatal diagnosis'/exp/mj OR prenatal:ab,ti OR (pre NEXT/1 natal):ab,ti OR antenatal:ab,ti OR diagnos*:ab,ti OR detect*:ab,ti OR predict*:ab,ti OR prognost*:ab,ti)	102266
5.	3 OR 4	164164
Chromosomal microarray analysis and next generation sequencing		
6.	'comparative genomic hybridization'/exp/mj OR 'microarray analysis'/exp/mj OR 'dna microarray'/exp/mj OR 'genetic association'/exp/mj OR 'genotyping technique'/exp/mj	35863
7.	(('comparative genomic' NEXT/1 hybridi?ation*) OR ('comparative genome' NEXT/1 hybridi?ation*) OR cgh OR bac OR ('bacterial artificial' NEXT/1 chromosome*)):ab,ti OR ((chromosomal* OR dna OR cytogenetic OR genomic OR 'genome-wide' OR snp OR snps OR 'single nucleotide polymorphism') NEAR/4 (array* OR microarray* OR 'micro-array' OR 'micro-arrays')):ti,ab OR (acgh:ab,ti OR oligo-aCGH OR ((whole OR complete OR entire OR full OR 'next generation' OR exome OR parallel OR 'high-through' OR ngs OR nps) NEAR/4 sequenc*)):ab,ti	[122071](#)
8.	6 OR 7	149482
Combined sets		
9.	5 AND 8	2016
Limits:		
10.	([article]/lim OR [article in press]/lim/)	
11.	**9 AND 10**	**974**

/de= Term from the EMTREE controlled vocabulary
/exp= Includes terms found below this term in the EMTREE hierarchy
/mj = Major Topic
:ab = Abstract
:au = Author
:ti = Article Title
:ti:ab = Title or abstract
* = Truncation
" " = Citation Marks; searches for an exact phrase " " = Citation Marks; searches for an exact phrase

PubMed via NLM 17 August 2015

Title: Chromosomal microarray analysis and next generation sequencing for prenatal diagnosis

Search terms		Items found
Prenatal diagnosis		
1.	"Prenatal Diagnosis"[Mesh] OR "Amniocentesis"[Mesh] OR "Chorionic Villi Sampling"[Mesh] OR "Maternal Serum Screening Tests"[Mesh] OR "Ultrasonography, Prenatal"[Mesh] OR "Nuchal Translucency Measurement"[Mesh]	61954
2.	((prenatal[Title/Abstract] OR pre-natal[Title/Abstract] OR antenatal[Title/Abstract] AND (diagnos*[Title/Abstract] OR detect*[Title/Abstract] OR predict*[Title/Abstract] OR prognos*[Title/Abstract] OR test[Title/Abstract] OR tests[Title/Abstract] OR testing[Title/Abstract] OR screen*[Title/Abstract])) OR amniocentesis[Title/Abstract] OR chorionic Villi sampling[Title/Abstract] OR nuchal translucency measurement[Title/Abstract] OR maternal serum screening test*[Title/Abstract]	59077
3.	1 OR 2	93904
4.	NIPT[Title/Abstract] OR NIPD[Title/Abstract] OR NIDT[Title/Abstract] OR NIFTY[Title/Abstract] OR cell-free dna[Title/Abstract] OR fetal dna[Title/Abstract] OR cff dna[Title/Abstract] OR cffdna[Title/Abstract] OR cf dna[Title/Abstract] OR cfdna[Title/Abstract] OR f dna[Title/Abstract] OR fdna[Title/Abstract] OR ff dna[Title/Abstract] OR ffdna[Title/Abstract] OR maternal serum screening test*[Title/Abstract] OR maternal plasm*[Title/Abstract] OR ((noninvasiv*[Title/Abstract] OR non-invasiv*[Title/Abstract]) AND ("Prenatal Diagnosis"[Mesh] OR prenatal[Title/Abstract] OR pre-natal[Title/Abstract] OR antenatal[Title/Abstract] OR diagnosis[Title/Abstract] OR diagnos*[Title/Abstract] OR detect*[Title/Abstract] OR predict*[Title/Abstract] OR prognos*[Title/Abstract] OR test[Title/Abstract] OR tests[Title/Abstract] OR testing[Title/Abstract]))	83479
5.	*3 OR 4*	*174428*
Chromosomal microarray analysis and next generation sequencing		
6.	"Comparative Genomic Hybridization"[Mesh] OR "Microarray Analysis"[Mesh:NoExp] OR "Oligonucleotide Array Sequence Analysis"[Mesh] OR "Genotyping Techniques"[Mesh]	85249
7.	comparative genomic hybridization*[Title/Abstract] OR comparative genomic hybridisation*[Title/Abstract] OR comparative genome hybridization*[Title/Abstract] OR comparative genome hybridisation*[Title/Abstract] OR cgh[Title/Abstract] OR bac[Title/Abstract] OR bacterial artificial chromosome*[Title/Abstract] OR ((chromosom*[Title/Abstract] OR DNA[Title/Abstract] OR cytogenetic[Title/Abstract] OR genomic[Title/Abstract] OR genome-wide[Title/Abstract] OR snp[Title/Abstract] OR snps[Title/Abstract] OR single nucleotide polymorphism*[Title/Abstract]) AND (array*[Title/Abstract] OR microarray*[Title/Abstract] OR micro-array*[Title/Abstract])) OR acgh[Title/Abstract] OR oligo-aCGH[Title/Abstract] OR whole sequenc* [Title/Abstract] OR complete sequenc* [Title/Abstract] OR entire sequenc* [Title/Abstract] OR full sequenc* [Title/Abstract] OR ((whole[Title/Abstract] OR complete[Title/Abstract] OR entire[Title/Abstract]) AND (genome[Title/Abstract] OR genomic[Title/Abstract]) AND sequenc*[Title/Abstract]) OR genome sequenc*[Title/Abstract] OR genomic sequenc*[Title/Abstract] OR parallel sequenc*[Title/Abstract] OR high-through sequenc*[Title/Abstract] OR ngs[Title/Abstract] OR nps[Title/Abstract] OR ((next generation*[Title/Abstract] OR exome[Title/Abstract]) AND sequenc*[Title/Abstract])	159060
8.	6 OR 7	218074
Reviews		
9.	Systematic[sb]	258201
Combined sets		
10.	5 AND 8	2042
11.	9 AND 10 Reviews	40
Limits		
12.	(animals [MeSH] NOT humans [MeSH])	4031755
	10 NOT 12	**1954**

The search result, usually found at the end of the documentation, forms the list of abstracts.

[MeSH] = Term from the Medline controlled vocabulary, including terms found below this term in the MeSH hierarchy
[MeSH:NoExp] = Does not include terms found below this term in the MeSH hierarchy
[MAJR] = MeSH Major Topic
[Title/Abstract] = Title or abstract
[Ti] = Title
[AU] = Author
[TW] = Text Word
Systematic[SB] = Filter for retrieving systematic reviews
* = Truncation