

## Summary of search protocol

Category	Search terms
Natural history	'natural history', pattern
Diagnosis 1	diagnosis, presentation
Diagnosis 2	AFP, alpha-fetoprotein, 'alpha fetoprotein', age, delay
Respiratory	respirat*, 'sinopulmonary disease', Bronchi*, 'interstitial lung disease', ILD, 'lung disease'
Neurology	neuroI*, neuroimag*, neurodisabil*, 'nerv* system, 'oculomotor apraxia', cerebell*, ataxia, dysarthria*, 'movement disorder', CT, 'computed tomography', MRI, 'magnetic resonance imaging', bradykinesia, dystonia, myoclonus, choreoathetosis, neurodegener*, tremor, PET, 'positron emission tomography'
Gastrointestinal	growth, nutrition, malnutrition, aspiration, dysphagia, PEG, gastroin*, gastrostomy, 'percutaneous endoscopic gastrostomy'
Malignancy	malignancy, cancer, leukaemia, leukemia, tumour, tumor, carcinoma, neoplasm
Genetics	genot*, phenot*, genet*, ATM, 'ataxia-telangiectasia mutated', 'ataxia-telangectasia mutated', 'ataxia telangiectasia mutated', 'ataxia telangectasia mutated', variant
Immunology	immunol*, 'immune system', infection, lymphop*, lymphc*
Endocrinology	endocrine*, hormone*, diabetes, T1DM, T2DM, 'insulin resistance', 'vit* D' or 'vit* E'
Orthopaedics	'skeletal abnormalit*', scoliosis, 'foot deformit*', fracture, bone
Dermatology	dermatology, skin, hyperpiment*, 'café au lait', hypopigment*, telangiectasia, telangiectasia
Death	death, mortality, survival
Pregnancy	fertility, pregnancy, childbirth
Cognition	neuropsychometric, IQ, 'cognitive function', intel*
Psychiatry	'mental state', mood, anxiety, depression
Lifestyle	'quality of life', lifestyle,

# Search Protocol

Search as per the search terms chart. Each column needs to be searched with the first column (Ataxia-telangiectasia) using 'AND'.

All date ranges, ages and languages.

Searching title, keywords and MeSH terms in each database.

## 1. PubMed

Human only filter.

- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND ("natural history"[Text Word] OR pattern[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (diagnosis[Text Word] OR presentation[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (AFP[Text Word] OR "alpha-fetoprotein"[Text Word] OR "alpha fetoprotein"[Text Word] OR age[Text Word] OR delay[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word]))AND (Respirat\*[Text Word] OR sinopulmonary disease[Text Word] OR Bronchi\*[Text Word] OR interstitial lung disease[Text Word] OR ILD[Text Word] OR lung disease[Text Word])

- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word]))AND (NeuroI\*[Text Word] OR Neuroimag\*[Text Word] OR neurodisabil\*[Text Word] OR "nerv\* system"[Text Word] OR "oculomotor apraxia"[Text Word] OR cerebell\*[Text Word] OR "ataxia"[Text Word] OR dysarthri\*[Text Word] OR "movement disorder"[Text Word] OR CT[Text Word] OR "computed tomography"[Text Word] OR MRI[Text Word] OR "magnetic resonance imaging"[Text Word] OR bradykinesia[Text Word] OR dystonia[Text Word] OR myoclonus[Text Word] OR choreoathetosis[Text Word] OR neurodegener\*[Text Word] OR tremor[Text Word] OR PET[Text Word] OR "positron emission tomography"[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word]))AND (Growth[Text Word] OR Nutrition[Text Word] OR malnutrition[Text Word] OR aspiration[Text Word] OR dysphagia[Text Word] OR PEG[Text Word] OR gastroin\*[Text Word] OR gastrostomy[Text Word] OR "percutaneous endoscopic gastrostomy"[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (Malignancy[Text Word] OR cancer[Text Word] OR leukaemia[Text Word] OR leukemia[Text Word] OR tumour[Text Word] OR tumor[Text Word] OR carcinoma[Text Word] OR neoplasm[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (Genot\*[Text Word] OR phenot\*[Text Word] OR genet\*[Text Word] OR ATM[Text Word] OR "Ataxia-

telangiectasia mutated "[Text Word] OR "Ataxia-telangeectasia mutated "[Text Word]  
OR "Ataxia telangiectasia mutated "[Text Word] OR "Ataxia telangeectasia mutated  
"[Text Word] OR variant[Text Word])

- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR  
"Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-  
Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (Immunol\*[Text Word] OR  
immune system[Text Word] OR infection[Text Word] OR lymphop\*[Text Word] OR  
lymphoc\*[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR  
"Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-  
Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (endocrin\*[Text Word] OR  
hormone\*[Text Word] OR diabetes[Text Word] OR T1DM[Text Word] OR T2DM[Text  
Word] OR "insulin resistance"[Text Word] OR "vit\* D"[Text Word] OR "vit\* E"[Text  
Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR  
"Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-  
Bar"[Text Word] OR "Louis Bar"[Text Word])) AND ("skeletal abnormalit\*\*"[Text Word]  
OR scoliosis[Text Word] OR "foot deformit\*\*"[Text Word] OR fracture[Text Word] OR  
bone[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR  
"Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-  
Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (dermatology[Text Word] OR  
skin[Text Word] OR hyperpigment\*[Text Word] OR "café au lait"[Text Word] OR  
hypopigment\*[Text Word] OR telangiectasia[Text Word] OR telangeectasia[Text  
Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR  
"Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-

Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (death[Text Word] OR mortality[Text Word] OR survival[Text Word])

- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (fertility[Text Word] OR pregnancy[Text Word] OR childbirth[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND (neuropsychometric[Text Word] OR IQ[Text Word] OR "cognitive function"[Text Word] OR intell\*[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND ("mental state"[Text Word] OR mood[Text Word] OR "anxiety"[Text Word] OR depression[Text Word])
- (("Ataxia-telangiectasia"[Text Word] OR "Ataxia-telangeectasia"[Text Word] OR "Ataxia telangiectasia"[Text Word] OR "Ataxia telangeectasia"[Text Word] OR "Louis-Bar"[Text Word] OR "Louis Bar"[Text Word])) AND ("quality of life"[Text Word] OR lifestyle[Text Word])

2. Ovid SP (MEDLINE) 1946- present HUMANS only and 3.) OVID EMBASE 1980 – present (HUMAN ONLY)

- a) Exp Ataxia-telangiectasia/ OR ataxia-telangiectasia.mp. OR ataxia-telangeectasia.mp. OR Louis-Bar.mp.
- b) (natural history or pattern\*).mp.
- c) exp Diagnosis/ OR (diagnosis OR presentation).mp.
- d) exp alpha-fetoprotein/ OR (alpha-fetoprotein OR age OR delay OR AFP).mp.

- e) exp lung disease/ OR (lung disease OR Respirat\* OR sinopulmonary disease OR Bronchi\* OR interstitial lung disease OR ILD).mp.
- f) (Neurol\* OR Neuroimag\* OR Neurodisabil\*OR nerv\* system OR oculomotor apraxia OR cerebell\* OR ataxia OR dysarthri\* OR movement disorder OR CT OR computed tomography OR MRI OR magnetic resonance imaging OR bradykinesia OR dystonia OR myoclonus OR choreoathetosis OR Neurodegen\* OR tremor OR PET OR positron emission tomography).mp.
- g) exp Growth/ OR exp Nutrition/ OR (Growth OR nutrition OR malnutrition OR aspiration OR dysphagia OR PEG OR gastro\* OR gastrostomy OR percutaneous endoscopic gastrostomy).mp.
- h) exp Malignancy/ OR exp cancer/ OR (malignancy OR cancer OR leukaemia OR leukemia OR tumour OR tumor OR carcinoma OR neoplasm).mp.
- i) Exp ataxia-telangiectasia mutated/ OR exp ataxia-telangectasia mutated OR (ataxia-telangiectasia mutated OR ataxia-telangectasia mutated OR Genot\* OR phenot\* OR genet\* OR ATM OR variant).mp.
- j) exp infection/ OR (infection OR Immuno\* OR immune system OR exp lymph\*).mp.
- k) exp endocrin\*/ OR exp diabetes/ OR (endocrine\* OR hormone OR diabetes OR T1DM OR T2DM OR insulin resistance OR vit\* D OR vit\* E).mp.
- l) (skeletal abnormalit\* OR scoliosis OR foot deformit\* OR fracture OR bone).mp.
- m) exp dermatology/ OR (dermatology OR skin OR hyperpigment\* OR cafe au lait OR hypopigment\* OR telangiectasia OR telangiectasia).mp.
- n) exp death/ OR (death OR mortality OR survival).mp.
- o) exp fertility/ OR exp pregnancy/ OR (fertility OR pregnancy OR childbirth).mp.
- p) (neuropsychometric OR IQ OR cognitive function OR intell\*).mp.
- q) (mental state OR mood OR anxiety OR depression).mp.
- r) Exp quality of life/ OR (quality of life OR lifestyle).mp.

All searches in Ovid carried out with keywords. Then all searches combined with search 2a with 'AND'.

4 Web of Science core collection

- a) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=("natural history" OR pattern)
- b) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS= (diagnosis OR presentation)
- c) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(AFP OR "alpha-fetoprotein" OR "alpha fetoprotein" OR age OR delay)
- d) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Respirat\* OR sinopulmonary disease OR Bronchi\* OR interstitial lung disease OR ILD OR lung disease)
- e) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Neurol\* OR Neuroimag\* OR Neurodisabil\* OR "nerv\* system" OR "oculomotor apraxia" OR cerebell\* OR "ataxia" OR dysarthri\* OR "movement disorder" OR CT OR "computed tomography" OR MRI OR "magnetic resonance imaging" OR bradykinesia OR dystonia OR myoclonus OR choreoathetosis OR Neurodegen\* OR tremor OR PET OR "positron emission tomography")
- f) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Growth OR Nutrition OR malnutrition OR aspiration OR dysphagia OR PEG OR gastro\* OR gastrostomy OR "percutaneous endoscopic gastrostomy")

- g) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Malignancy OR cancer OR leukaemia OR leukemia OR tumour OR tumor OR carcinoma OR neoplasm)
- h) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Genot\* OR phenot\* OR genet\* OR ATM OR "Ataxia-telangiectasia mutated " OR "Ataxia-telangeectasia mutated " OR "Ataxia telangiectasia mutated " OR "Ataxia telangeectasia mutated " OR variant)
- i) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(Immuno\* OR immune system OR infection OR lymph\*)
- j) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(endocrin\* OR hormone OR diabetes OR T1DM OR T2DM OR "insulin resistance" OR "vit\* D" OR "vit\* E")
- k) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=("skeletal abnormalit\*" OR scoliosis OR "foot deformit\*" OR fracture OR bone)
- l) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(dermatology OR skin OR hyperpigment\* OR "café au lait" OR hypopigment\* OR telangiectasia OR telangeectasia)
- m) TS=("Ataxia-telangiectasia" OR "Ataxia-telangeectasia" OR "Ataxia telangiectasia" OR "Ataxia telangeectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS= (death OR mortality OR survival)



- n) TS=("Ataxia-telangiectasia" OR "Ataxia-telangectasia" OR "Ataxia telangiectasia" OR "Ataxia telangectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(fertility OR pregnancy OR childbirth)
- o) TS=("Ataxia-telangiectasia" OR "Ataxia-telangectasia" OR "Ataxia telangiectasia" OR "Ataxia telangectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=(neuropsychometric OR IQ OR "cognitive function" OR intell\*)
- p) TS=("Ataxia-telangiectasia" OR "Ataxia-telangectasia" OR "Ataxia telangiectasia" OR "Ataxia telangectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=("mental state" OR mood OR anxiety OR depression)
- q) TS=("Ataxia-telangiectasia" OR "Ataxia-telangectasia" OR "Ataxia telangiectasia" OR "Ataxia telangectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome") AND TS=("quality of life" OR lifestyle)

## 5. Scopus

Only the following categories were searched: medicine, 'biochemistry, genetics and molecular biology', 'immunology and microbiology', neuroscience, 'pharmacology, toxicology and pharmaceuticals', health professions'

- a) TITLE-ABS-KEY ( *"Ataxia-telang\*ectasia"* OR *"Ataxia telang\*ectasia"* OR *"Louis-Bar syndrome"* OR *"Louis Bar syndrome"* ) AND TITLE-ABS-KEY ( *"natural history"* OR *pattern* )
- b) TITLE-ABS-KEY ( *"Ataxia-telang\*ectasia"* OR *"Ataxia telang\*ectasia"* OR *"Louis-Bar syndrome"* OR *"Louis Bar syndrome"* ) AND TITLE-ABS-KEY ( *diagnosis* OR *presentation* )
- c) TITLE-ABS-KEY ( *"Ataxia-telang\*ectasia"* OR *"Ataxia telang\*ectasia"* OR *"Louis-Bar syndrome"* OR *"Louis Bar syndrome"* ) AND TITLE-ABS-KEY ( *AFP* OR *"alpha-fetoprotein"* OR *"alpha fetoprotein"* OR *age* OR *delay* )

- d) TITLE-ABS-KEY ( "Ataxia-telang\*ectasia" OR "Ataxia telang\*ectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome" ) AND TITLE-ABS-KEY (Respirat\* OR "sinopulmonary disease" OR Bronchi\* OR "interstitial lung disease" OR ILD OR "lung disease")
- e) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY (Neurol\* OR Neuroimag\* OR Neurodisabil\* OR "nerv\* system" OR "oculomotor apraxia" OR cerebell\* OR ataxia OR dysarthri\* OR "movement disorder" OR CT OR "computed tomography" OR MRI OR "magnetic resonance imaging" OR bradykinesia OR dystonia OR myoclonus OR choreoathetosis OR Neurodegen\* OR tremor OR PET OR "positron emission tomography")
- f) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( *growth* OR *nutrition* OR *malnutrition* OR *aspiration* OR *dysphagia* OR *peg* OR *gastro\** OR *gastrostomy* OR "*percutaneous endoscopic gastrostomy*")
- g) TITLE-ABS-KEY ( "Ataxia-telang\*ectasia" OR "Ataxia telang\*ectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome" ) AND TITLE-ABS-KEY (Malignancy OR cancer OR leukaemia OR leukemia OR tumour OR tumor OR carcinoma OR neoplasm)
- h) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY (Genot\* OR phenot\* OR genet\* OR ATM OR "Ataxia-telangiectasia mutated" OR "Ataxia-telangectasia mutated" OR "Ataxia telangiectasia mutated" OR "Ataxia telangectasia mutated" OR variant)
- i) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( *immuno\** OR "*immune system*" OR *infection* OR *lymph\**)

- j) TITLE-ABS-KEY ( "Ataxia-telang\*ectasia" OR "Ataxia telang\*ectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome" ) AND TITLE-ABS-KEY (endocrin\* OR hormone OR diabetes OR T1DM OR T2DM OR "insulin resistance" OR "vit\* D" OR "vit\* E")
- k) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( "*skeletal abnormalit\**" OR *scoliosis* OR "*foot deformit\**" OR *fracture* OR *bone* )
- l) TITLE-ABS-KEY ( "Ataxia-telang\*ectasia" OR "Ataxia telang\*ectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome" ) AND TITLE-ABS-KEY (dermatology OR skin OR hyperpigment\* OR "café au lait" OR hypopigment\* OR telangiectasia OR telangectasia)
- m) TITLE-ABS-KEY ( "Ataxia-telang\*ectasia" OR "Ataxia telang\*ectasia" OR "Louis-Bar syndrome" OR "Louis Bar syndrome" ) AND TITLE-ABS-KEY (death OR mortality OR survival)
- n) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( *fertility* OR *pregnancy* OR *childbirth* )
- o) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( *neuropsychometric* OR *iq* OR "*cognitive function*" OR *intell\** )
- p) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( "mental state" OR mood OR anxiety OR depression)
- q) TITLE-ABS-KEY ( "*Ataxia-telang\*ectasia*" OR "*Ataxia telang\*ectasia*" OR "*Louis-Bar syndrome*" OR "*Louis Bar syndrome*") AND TITLE-ABS-KEY ( "quality of life" OR lifestyle)

Prospero and Cochrane Library - 28/3/18 – no systematic reviews completed or registered