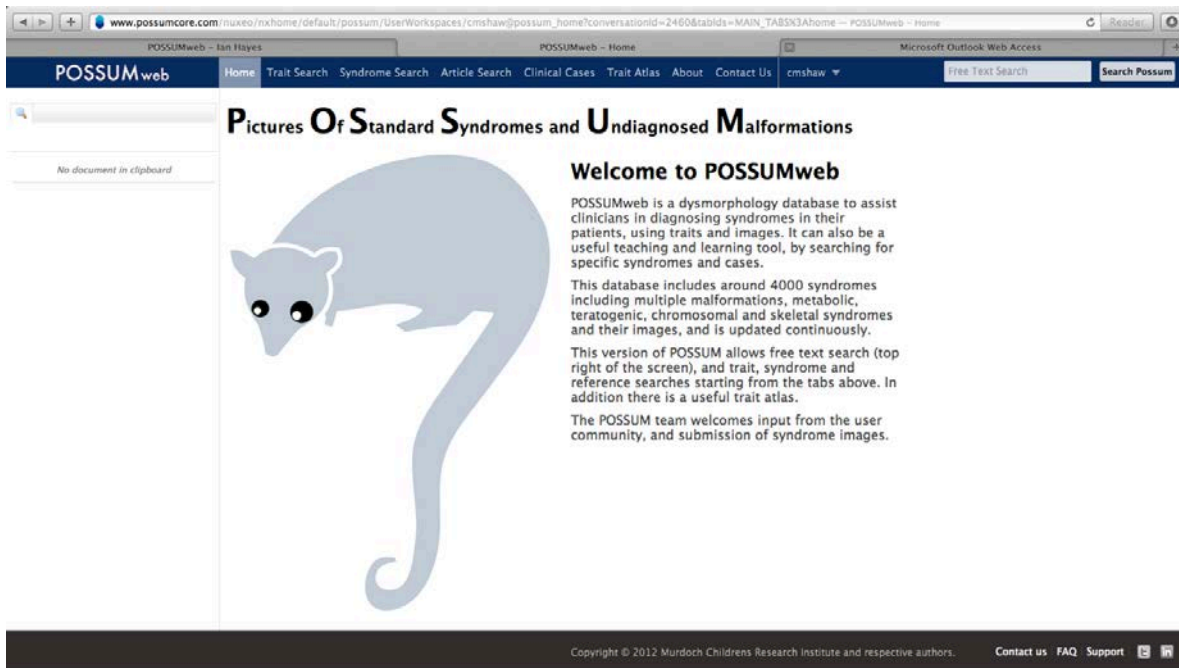


A SHORT GUIDE TO THE LATEST POSSUMweb

1. Searching using traits

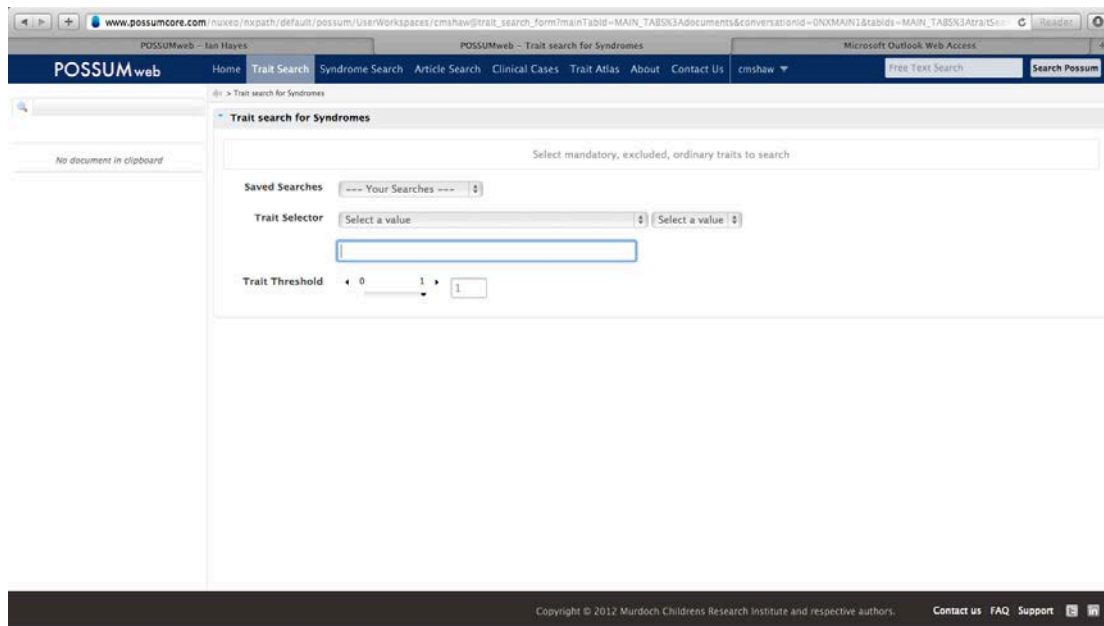
After logging in you come to the Home page:



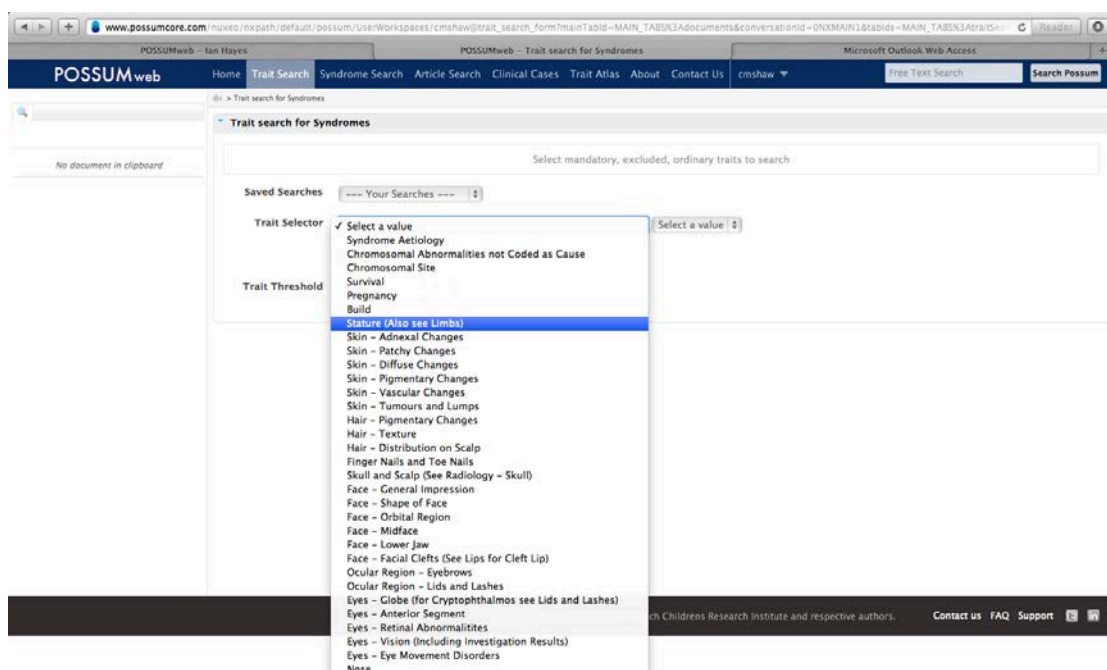
The actions take place using the upper tool bar.

Trait searching

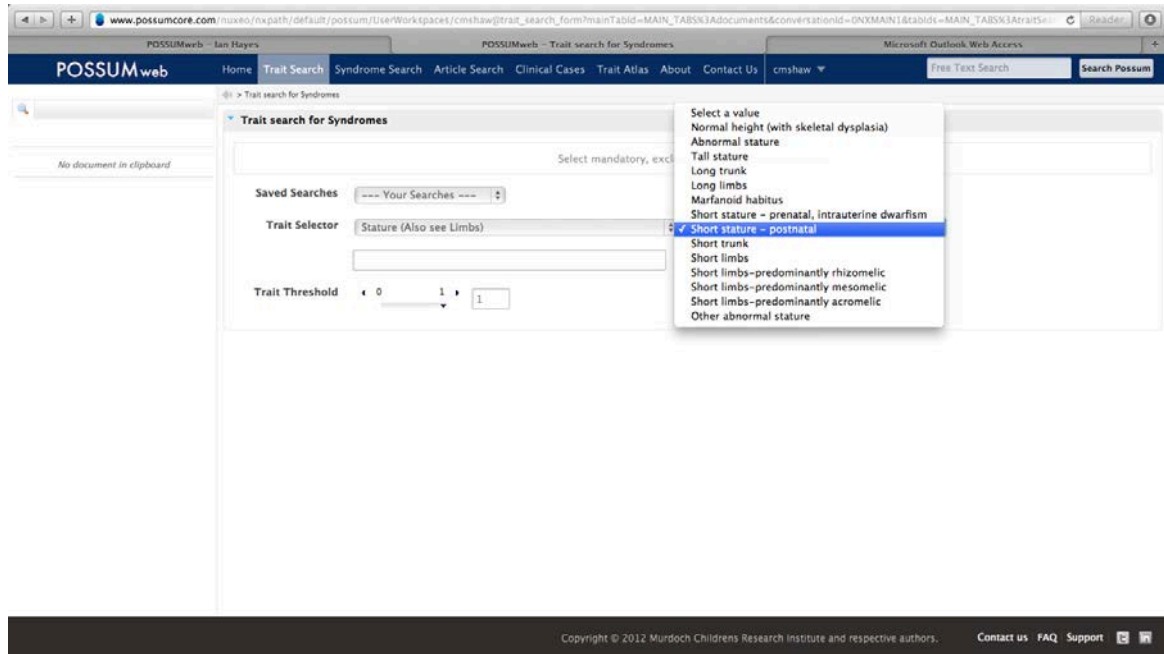
select the Trait search tab from the upper toolbar



Then select a group of traits, in this case **Stature**



And then select the specific trait, in this case **Short stature - postnatal**.



As each trait is added the search is performed.

www.possuimcore.com /nuxeo/oxpath/default/possum/UserWorkspaces/cmshaw/trait_search_form?mainTabId=MAIN_TABSK3Adocuments&conversationId=0NXMAIN1&tabId=MAIN_TABSK3Atraits

POSSUMweb - Ian Hayes

POSSUMweb - Trait search for Syndromes

Microsoft Outlook Web Access

No document in clipboard

Trait search for Syndromes

Select mandatory, excluded, ordinary traits to search

Saved Searches --- Your Searches ---

Trait Selector Stature (Also see Limbs) Short stature - postnatal

Trait Threshold 0 1 1

Selected Traits Short stature - postnatal Ordinary remove

Save Search Save Search

Share this saved search with others

Clear

1382 matching syndromes found

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 1 | 3087 | 147791 | 11q terminal deletion disorder ^{li} |
| 1 | 5271 | 258501 | 3-methylglutaconic aciduria, type 3 ^{li} |
| 1 | 3613 | 273750 | 3M syndrome ^{li} |
| 1 | 6093 | 605756 | 46,XX gonadal dysgenesis, short stature, recurrent metabolic acidosis ^{li} |
| 1 | 3111 | | 49,XXXXY syndrome ^{li} |
| 1 | 6719 | | Aarskog-like syndrome, Xu-Yu type ^{li} |
| 1 | 3001 | 305400 | Aarskog syndrome ^{li} |
| 1 | 3002 | 105650 | Aase syndrome ^{li} |
| 1 | 4318 | 200100 | Abetalipoproteinemia ^{li} |
| 1 | 3919 | 200110 | Ablepharon and macrostoma ^{li} |
| 1 | 5341 | | Abnormal facies, mental retardation, ectomorphic habitus ^{li} |

Keep adding traits to assist in your diagnosis...adding **Pigmented naevi**

www.possuimcore.com /nuxeo/oxpath/default/possum/UserWorkspaces/cmshaw/trait_search_form?mainTabId=MAIN_TABSK3Adocuments&conversationId=0NXMAIN1&tabId=MAIN_TABSK3Atraits

POSSUMweb - Ian Hayes

POSSUMweb - Trait search for Syndromes

Microsoft Outlook Web Access

No document in clipboard

Trait search for Syndromes

Select mandatory, excluded, ordinary traits to search

Saved Searches --- Your Searches ---

Trait Selector Skin - Pigmentary Changes

Trait Threshold 0 1 1

Selected Traits Short stature - postnatal Ordinary remove

Save Search Save Search

Share this saved search with others

Clear

Select a value

- Abnormal pigmentary skin changes
- Vitiligo
- Decreased skin pigmentation - diffuse
- Decreased skin pigmentation - irregular
- Decreased skin pigmentation - patchy-streaky
- Increased skin pigmentation - diffuse
- Increased skin pigmentation - irregular
- Increased skin pigmentation - patchy-streaky
- Acanthosis nigricans
- Pigmented naevi**
- Cafe au lait
- Excessive freckling
- Lentiginos
- Other pigmentary skin changes

1382 matching syndromes found

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 1 | 3087 | 147791 | 11q terminal deletion disorder ^{li} |
| 1 | 5271 | 258501 | 3-methylglutaconic aciduria, type 3 ^{li} |
| 1 | 3613 | 273750 | 3M syndrome ^{li} |
| 1 | 6093 | 605756 | 46,XX gonadal dysgenesis, short stature, recurrent metabolic acidosis ^{li} |
| 1 | 3111 | | 49,XXXXY syndrome ^{li} |
| 1 | 6719 | | Aarskog-like syndrome, Xu-Yu type ^{li} |
| 1 | 3001 | 305400 | Aarskog syndrome ^{li} |
| 1 | 3002 | 105650 | Aase syndrome ^{li} |
| 1 | 4318 | 200100 | Abetalipoproteinemia ^{li} |
| 1 | 3919 | 200110 | Ablepharon and macrostoma ^{li} |
| 1 | 5341 | | Abnormal facies, mental retardation, ectomorphic habitus ^{li} |

Adding further traits adds to the matched syndromes. The next screenshot shows two traits, matching to either trait, or to both traits. Number of matches in the left side

The screenshot shows the 'Trait search for Syndromes' interface. The search parameters are: Trait Selector: Skin - Pigmentary Changes, Trait: Pigmented naevi, Trait Threshold: 1. Selected Traits: Short stature - postnatal (Ordinary), Pigmented naevi (Ordinary). The interface shows 1426 matching syndromes found. The following table lists the first 10 results:

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 2 | 3011 | 101800 | Acrodysostosis ^{li} |
| 2 | 3651 | 276820 | Al-Awadi-Raas-Rothschild syndrome ^{li} |
| 2 | 6539 | 612079 | Alopecia, neurological defects, endocrinopathy ^{li} |
| 2 | 3627 | 115150 | Cardiofaciocutaneous syndrome ^{li} |
| 2 | 5620 | | Central sclerosis, bamboo hair, ichthyosis ^{li} |
| 2 | 6530 | 612001 | Chromosome 15, microdeletion 15q13 ^{li} |
| 2 | 4716 | | Chromosome 8, mosaic tetrasomy 8p ^{li} |
| 2 | 4189 | 251260 | Chromosome instability disorder, Maraschio-Peretti type ^{li} |
| 2 | 5572 | | Chromosome X, del Xq ^{li} |
| 2 | 5573 | | Chromosome X, ring (X) ^{li} |

column.

This screenshot shows that there are 40 matches if you want 2 out of 2 matches.

www.possuimcore.com /nuxeo/.../trait_search_form7mainTabId=MAIN_TABS3Adocuments&conversationId=INXMAIN1&tabId=MAIN_TABS3AtraitSe... Reader

POSSUMweb - Ian Hayes POSSUMweb - Trait search for Syndromes Microsoft Outlook Web Access

No document in clipboard

Trait search for Syndromes

Select mandatory, excluded, ordinary traits to search

Saved Searches --- Your Searches ---

Trait Selector Skin - Pigmentary Changes Pigmented naevi

Trait Threshold 0 2 2

Selected Traits Short stature - postnatal Ordinary remove
Pigmented naevi Ordinary remove

Save Search Save Search

Share this saved search with others

Clear

40 matching syndromes found

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 2 | 3011 | 101800 | Acrodysostosis ^{li} |
| 2 | 3651 | 276820 | Al-Awadi-Raas-Rothschild syndrome ^{li} |
| 2 | 6539 | 612079 | Alopecia, neurological defects, endocrinopathy ^{li} |
| 2 | 3627 | 115150 | Cardiofaciocutaneous syndrome ^{li} |
| 2 | 5620 | | Central sclerosis, bamboo hair, ichthyosis ^{li} |
| 2 | 6530 | 612001 | Chromosome 15, microdeletion 15q13 ^{li} |
| 2 | 4716 | | Chromosome 8, mosaic tetrasomy 8p ^{li} |
| 2 | 4189 | 251260 | Chromosome instability disorder, Maraschio-Peretti type ^{li} |
| 2 | 5572 | | Chromosome X, del Xq ^{li} |
| 2 | 5573 | | Chromosome X, ring (X) ^{li} |

You can also choose that a trait be **mandatory**

www.possumcore.com nuxeo/nxpath/default/possum/UserWorkspaces/cmshaw/trait_search_form?mainTabId=MAIN_TABSK3Adocuments&conversationId=0NXMAIN1&tabId=MAIN_TABSK3AtraItS...

POSSUMweb - Ian Hayes POSSUMweb - Trait search for Syndromes Microsoft Outlook Web Access

No document in clipboard

Select mandatory, excluded, ordinary traits to search

Saved Searches --- Your Searches ---

Trait Selector Skin - Pigmentary Changes Pigmented naevi

Trait Threshold 0 2 1

Selected Traits Short stature - postnatal Ordinary remove
Pigmented naevi Mandatory remove

Save Search Save Search

Share this saved search with others

Clear

84 matching syndromes found

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 2 | 3011 | 101800 | Acrodysostosis ^{li} |
| 2 | 3651 | 276820 | Al-Awadi-Raas-Rothschild syndrome ^{li} |
| 2 | 6539 | 612079 | Alopecia, neurological defects, endocrinopathy ^{li} |
| 2 | 3627 | 115150 | Cardiofaciocutaneous syndrome ^{li} |
| 2 | 5620 | | Central scleriosis, bamboo hair, ichthyosis ^{li} |
| 2 | 6530 | 612001 | Chromosome 15, microdeletion 15q13 ^{li} |
| 2 | 4716 | | Chromosome 8, mosaic tetrasomy 8p ^{li} |
| 2 | 4189 | 251260 | Chromosome instability disorder, Maraschio-Peretti type ^{li} |
| 2 | 5572 | | Chromosome X, del Xq ^{li} |
| 2 | 5573 | | Chromosome X, ring 00 ^{li} |
| 2 | 3965 | | Cohen-Scott-Smith syndrome ^{li} |

You can keep adding traits

www.possumcore.com nuxeo/nxpath/default/possum/UserWorkspaces/cmshaw/trait_search_form?mainTabId=MAIN_TABSK3Adocuments&conversationId=0NXMAIN1&tabId=MAIN_TABSK3AtraItS...

POSSUMweb - Ian Hayes POSSUMweb - Trait search for Syndromes Microsoft Outlook Web Access

No document in clipboard

Select mandatory, excluded, ordinary traits to search

Saved Searches --- Your Searches ---

Trait Selector Neurological - Functional Abnormalities Muscular hypertonia, spasticity, rigidity, brisk refl

Trait Threshold 0 3 2

Selected Traits Short stature - postnatal Ordinary remove
Pigmented naevi Ordinary remove
Muscular hypertonia, spasticity, rigidity, brisk refl Ordinary remove

Save Search Save Search

Share this saved search with others

Clear

1662 matching syndromes found

| Score | Id | OMIM | Name |
|-------|------|--------|---|
| 3 | 3627 | 115150 | Cardiofaciocutaneous syndrome ^{li} |
| 3 | 5620 | | Central scleriosis, bamboo hair, ichthyosis ^{li} |
| 3 | 3234 | 227650 | Fanconi pancytopenia syndrome ^{li} |
| 3 | 4101 | | Mental retardation, short stature, unusual facies ^{li} |
| 3 | 3329 | 310465 | Multiple anomalies, mental retardation, deafness ^{li} |
| 3 | 6604 | 613078 | Nijmegen breakage syndrome-like disorder ^{li} |
| 3 | 3257 | 309580 | X-linked mental retardation-hypotonic facies syndrome ^{li} |
| 2 | 5271 | 258501 | 3-methylglutaconic aciduria, type 3 ^{li} |
| 2 | 4241 | 231550 | Achalasia-adrenal-alacrima syndrome ^{li} |
| 2 | 4321 | 201100 | Acrodermatitis enteropathica ^{li} |
| 2 | 3011 | 101800 | Acrodysostosis ^{li} |
| 2 | 3651 | 276820 | Al-Awadi-Raas-Rothschild syndrome ^{li} |

A SHORT GUIDE TO THE LATEST POSSUMweb

2. Searching for syndromes by name, OMIM number, POSSUM number or Reference

After logging in you come to the Home page:



The actions take place using the upper tool bar.

Syndrome search..... select the syndrome search tab.

The screenshot shows the POSSUM web application interface. The top navigation bar includes links for Home, Trait Search, Syndrome Search, Article Search, Clinical Cases, Trait Atlas, About, Contact Us, Document Management, Admin Center, Studio, and Cathie. The main content area is titled "Search Results" and displays a table of search results. The table has columns for Syndrome Id, Title, OMIM, Modified, Last contributor, and Version State. The results list various syndromes such as "11q terminal deletion disorder", "15q overgrowth syndrome", and "3-hydroxyacyl-CoA dehydrogenase deficiency".

| Syndrome Id | Title | OMIM | Modified | Last contributor | Version State |
|-------------|--|--------|------------|------------------|---------------|
| 3087 | 11q terminal deletion disorder | 147791 | 10/17/2012 | Cathie Rose | 0.0 Approved |
| 6308 | 15q overgrowth syndrome | | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 4926 | 3-hydroxyacyl-CoA dehydrogenase deficiency | 143450 | 9/8/2012 | Cathie Rose | 0.0 Approved |
| 3727 | 3-hydroxyisobutyric aciduria | 236795 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 6870 | 3-methylglutaconic aciduria, deafness, encephalopathy, and Leigh-like syndrome | 614739 | 10/18/2012 | Cathie Rose | 0.0 Approved |
| 5266 | 3-methylglutaconic aciduria, type 1 | 250950 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 5271 | 3-methylglutaconic aciduria, type 3 | 258501 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 5272 | 3-methylglutaconic aciduria, type 4 | 250951 | 9/8/2012 | Cathie Rose | 0.0 Approved |
| 3613 | 3M syndrome | 273750 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 5580 | 3-Phosphoglycerate dehydrogenase deficiency | 601815 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 6367 | 3q29 microdeletion syndrome | 609425 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 6529 | 3q29 microduplication syndrome | 611936 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 6093 | 46,XX gonadal dysgenesis, short stature, recurrent metabolic acidosis | 605756 | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 5483 | 48XYXX syndrome | | 9/7/2012 | Cathie Rose | 0.0 Approved |
| 3111 | 49,XXXXY syndrome | | 9/7/2012 | Cathie Rose | 0.0 Approved |

And either type in a name, OMIM number or POSSUM number.

In this example I searched for **Noonan syndrome**.

The screenshot shows the POSSUMweb search results page. The search term 'Noonan' is entered in the search box. The results table lists several syndromes, with 'Noonan syndrome' (OMIM 163950) highlighted. The table columns include Syndrome Id, Title, OMIM, Modified, Last contributor, Version, and State.

| Syndrome Id | Title | OMIM | Modified | Last contributor | Version | State |
|-------------|--|--------|------------|------------------|---------|----------|
| 4342 | Baraitser-Patton syndrome | | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 3965 | Cohen-Scott-Smith syndrome | | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 3286 | McDonough syndrome | 248950 | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 4205 | Neurofibromatosis-Noonan syndrome | 601321 | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 3972 | Noonan-like, contractures, myopathy, hyperpyrexia | | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 6742 | Noonan-like syndrome, CBL tumour-suppressor gene mutations | 613563 | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 6287 | Noonan-like syndrome, loose anagen hair | 607721 | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 4719 | Noonan-like with digital anomalies | | 9/7/2012 | Cathie Rose | 0.0 | Approved |
| 3335 | Noonan syndrome | 163950 | 10/16/2012 | Cathie Rose | 0.0 | Approved |
| 3390 | Short rib-polydactyly syndrome, Saldino-Noonan type | 263530 | 9/7/2012 | Cathie Rose | 0.0 | Approved |

You can scroll down and select your syndrome to look at, in this case Noonan syndrome.

The screenshot shows the detailed view of 'Noonan syndrome' on the POSSUMweb site. The page includes sections for Syndrome Details, Description, and Clinical Cases.

SYNDROME DETAILS

- Name: Noonan syndrome
- Syndrome Id: 3335
- OMIM Link: 163950
- Gene Location: 12q24.1, 2p22-p21, 12p12.1, 3p25, 1p32.2, 7q34
- Alternate Names: Noonan syndrome, Male Turner-like syndrome, Pseudo-Turner syndrome

DESCRIPTION

Clinical
Short stature with often short webbed neck, pectus carinatum/excavatum and a distinctive faces including hypertelorism, downslanting palpebral fissures, ptosis and low-set posteriorly rotated ears with a thickened helix. Cardiac anomalies, especially pulmonary valve stenosis, are frequent. The facial appearance evolves with age and may be more difficult to recognise in the adult (note prominent naso-labial folds). Keratotic papules of eyebrows and pulmonary lymphangiectasis reported.

For a review of facial phenotype versus genotype see (Allanson 2010). Three cases reported with SLE.

Intelligence often normal; about 30% of cases require special education. Some cases with Arnold-Chiari malformation.

Association with myeloproliferative disorder in childhood reported (Bader-Meunier 1997, Kratz 2006).

Report and review (Nisbet 1999) of cases of Noonan syndrome presenting prenatally with ultrasound abnormalities including increased nuchal fluid, short femora, pleural effusions, hydrops, cardiac and renal anomalies.

Noonan syndrome 4 with SOS1 mutations (Roberts 2007):

CLINICAL CASES

- Case Id - 655
 - Case At Age: 2 years and 10 months
- Case Id - 1732
 - Case At Age: 13 years
- Case Id - 1955
 - Case At Age: 4 years and 3 months
- Case Id - 259
 - Case At Age: 5 years
 - Case At Age: 13 years and 6 months
 - Case At Age: 2 years
- Case Id - 565
 - Case At Age: 7 years and 3 months
 - Case At Age: 3 years and 6 months
 - Case At Age: 12 months
 - Case At Age: 3 months
- Case Id - 569
 - Case At Age: 3 years
 - Case At Age: 7 years
- Case Id - 563
 - Case At Age: 13 years and 4 months
 - Case At Age: 6 years and 4 months
- Case Id - 12037
 - Case At Age: 7 months
- Case Id - 932
 - Case At Age: 7 days
- Case Id - 394
 - Case At Age: 8 years and 6 months

Scroll down for images.

The screenshot shows a web browser window with the URL www.possuimcore.com/muxeo/nextpath/default/possuimcore/syndromes/Noonan%20syndrome/view_documents?tabId=93A&conversationId=6NXMAIN1. The page title is "POSSUIMweb - Noonan syndrome". The main content area is titled "PICTURES" and contains a grid of 20 small photographs of children's faces, arranged in 5 rows and 4 columns. The children are of various ages and ethnicities, illustrating the clinical features of Noonan syndrome. To the right of the grid is a list of references, including:

- Carvalho, Daniel.Rocha., et al. 'Noonan syndrome associated with unilateral iris coloboma and congenital chylothorax in an infant', *Clin Dysmorph*, 12: 143-144, 2003
- Cesàrini, Laura., et al. 'Cognitive Profile of Disorders Associated With Dysregulation of the RAS/RAF/Signaling Cascade', *Am J Med Genet*, 149A: 140-146, 2009
- Cristea, I.C., et al. 'A restricted spectrum of NRAS mutations cause Noonan syndrome', *Nature Genet*, 42: 27-29, 2010
- Collins, E., et al. 'The Noonan syndrome - a review of the clinical and genetic features of 27 cases.', *J Pediatr*, 83: 941-950, 1973
- Croonen, Ellen.A., et al. 'Electrocardiography in Noonan syndrome PTPN11 gene mutation - phenotype characterization', *Am J Med Genet*, 146A: 350-353, 2008
- Derbent, Murat., et al. 'Clinical and Hematologic Findings in Noonan Syndrome Patients With PTPN11 Gene Mutations', *Am J Med Genet*, 152A: 2768-2774, 2010
- Dollfus, Hélène., et al. 'Bilateral Iridoretinal colobomas in a child with a Noonan phenotype', *Clin Dysmorph*, 10: 299-300, 2001
- Elalaoui, S.Chafai., et al. 'Germinal Mosaicism in Noonan Syndrome: A Family With Two Affected Siblings of Normal Parents', *Am J Med Genet*, 152A: 2850-2853, 2010
- Fabretto, Antonella., et al. 'Two cases of Noonan syndrome with severe respiratory and gastro-intestinal involvement and the S051 mutation f623f', *Europ J Med Genet*, 53: 322-324, 2010
- Ferrero, Giovanni.Battista., et al. 'Clinical and molecular characterization of 40 patients with Noonan syndrome', *Europ J Med Genet*, 51: 566-572, 2008
- Fryns, J.P., et al. 'Progressive hydrocephalus in Noonan syndrome', *Clin Dysmorph*, 6: 379, 1997
- Fukushima, Y., et al. 'Noonan syndrome and its related disorders.', *Acta Paediatr Jpn*, 38: 102-104, 1996

Individual clinic cases at different ages, and the references, are located to the right of the webpage.

Opening a reference takes you to a link to the pubmed entry.

The screenshot shows a PubMed search result page. The search query is "Am J Med Genet[jour] AND 2011[pdat] AND Alfieri[author] AND Visual processing in Noonan syndrome[dorsal+and+ventral+].". The search results show one entry:

Am J Med Genet. 2011 Oct;155A(10):2459-64. doi: 10.1002/ajmg.a.34229. Epub 2011 Sep 9.

Visual processing in Noonan syndrome: dorsal and ventral stream sensitivity.

Alfieri P, Cesàrini L, De Rose P, Ricci D, Selicomi A, Menchini A, Guzzetta A, Baranello G, Tinelli F, Mallardi M, Zampino G, Vicari S, Atkinson J, Mercuri F. *Child Neuropsychiatry Unit, Department of Neuroscience, Bambino Gesù, Children's Hospital, Rome, Italy.*

Abstract
Global spatial and motion processing abilities were assessed in 18 patients with Noonan syndrome (NS) and in 43 matched controls using form and motion coherence testing, respectively. We observed a discrepancy between the two groups since the study group had significantly lower performances than the control group for form coherence while there was no impairment on motion coherence. All the patients were also assessed on the Movement Assessment Battery for Children (M-ABC) to evaluate visuospatial skills. Thirteen of the 18 (72%) also had global poor performances on the M-ABC. The results show that children with NS have a specific impairment in the global processing of visuospatial information and are likely to have a specific ventral stream deficit as also suggested by the frequent visuospatial perceptual difficulties. Testing form and motion coherence thresholds may be a useful diagnostic tool for this group of patients, despite their normal cognitive abilities, since aspects of global form processing and visuospatial perceptual difficulties can be identified and potentially targeted for a specific rehabilitation program.

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PMID: 21910245 [PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms
LinkOut - more resources

Related citations in PubMed
Neurobiological models of visuospatial cognition in children with Williams [Dev Neuropsychol. 2003]
Form and motion coherence processing in dyspraxia: evidence of a global [Neuroreport. 2002]
Differential vulnerability of global motion, global form, and biological motion [Neuropsychologia. 2009]
From genes to brain development to phenotypic behavior: 'top' [Prog Brain Res. 2011]
Normal and anomalous development of visual motion processes [Neuropsychologia. 2003]

Search details
[Am J Med Genet [journal] OR Am J Med Genet C Semin Med Genet [journal] OR Am J Med Genet B Neuropsychiatr Genet [journal] OR Am J Med Genet A [journal] AND 2011[pdat] AND Alfieri[author] AND (Visual[Title] AND processing[Title] AND ...]

Recent activity
Visual processing in Noonan syndrome: dorsal and ventral stream sensitivity. PubMed
Am J Med Genet [jour] AND 2011[pdat] AND Alfieri[author] AND Visual...

There are [links](#) in the text of syndromes to [OMIM](#) and to [related syndromes](#). The links open in a new web page.

[Free text search](#) also possible using top right search feature. This search will find syndromes and articles with the search term.

