This search summary contains the results of a literature search undertaken by the Lincolnshire Knowledge and Resource Service librarians in; 

**August 2014**

All of the literature searches we complete are tailored to the specific needs of the individual requester.

If you would like this search re-run with a different focus, or updated to accommodate papers published since the search was completed, please let us know. This literature searching service is available to support public health / health and social care commissioning in Lincolnshire.

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“Google can bring you back 100,000 answers, a librarian can bring you back the right one.”

*Neil Gaiman*
“Google can bring you back 100,000 answers, a librarian can bring you back the right one.”

Neil Gaiman
Lincolnshire Knowledge and Resource Service

Please find below the results of your literature search request. If you would like the full text of any of the abstracts included, or would like a further search completed on this topic, please let us know.

“Google can bring you back 100,000 answers, a librarian can bring you back the right one.”
Neil Gaiman

Literature Search Results

Search request date: 6th August 2014
Search completion date: 7th August 2014
Search completed by: Jan Badcock

Enquiry Details

Can you find me some papers about Aarskog Syndrome and its effects/characteristics? Anything linked to physiotherapy would be ideal but I don’t think you’ll be able to find any.

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Every effort has been made to ensure that this information is accurate, up-to-date, and complete. However it is possible that it is not representative of the whole body of evidence available. No responsibility can be accepted for any action taken on the basis of this information. It is the responsibility of the requester to determine the accuracy, validity and interpretation of the search results.

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Links are given to full text resources where available. For some of the papers, you will need a free NHS & Social Care Athens Account.

If you do not have an account you can register by following the steps at: https://register.athensams.net/nhs/nhseng/

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Guidance on Searching within Online Documents

Links are provided to the full text documents where available. Relevant extracts have been copied and pasted into these Search Results.

Rather than browse through a whole documents, you can search for specific words and phrases as follows:

**Portable Document Format / pdf. / Adobe**
Click on the Search button (illustrated with binoculars). This will open up a search window. Type in the term you need to find and links to all of the references to that term within the document will be displayed in the window. You can jump to each reference by clicking it. You can search for more terms by pressing 'search again'.

**Word documents**
Select Edit from the menu, the Find and type in your term in the search box which is presented. The search function will locate the first use of the term in the document. By pressing 'next' you will jump to further references.
Aarskog-Scott syndrome  
Orphanet (accessed 7th July 2014)

Summary

Aarskog-Scott syndrome (AAS) is a rare developmental disorder characterized by facial, limbs and genital features, and a disproportionate acromelic short stature.

AAS prevalence is not known, but less than 100 cases have been reported in the literature since the first description in 1970. However, prevalence estimates are thought to be around 1/25,000. About 40 molecularly proven cases are published worldwide.

AAS predominantly concerns males. Facial features include widow's peak and hypertelorism, both observed in female carriers, and downslanting palpebral fissures, broad nasal bridge, anteverted nostrils, low set and protuberant ears, maxillary hypoplasia and transverse crease below the lower lip. AAS patients have short and broad hands and feet, interdigital webbing, clinodactyly, and hyperextension of proximal interphalangeal joints and flexion at distal interphalangeal joints causing swan neck deformity of fingers. Size is generally normal at birth, but growth is slow in infancy and childhood, leading to short stature until puberty, which is often delayed. A growth spurt in late teens, generally, results in a moderate short stature. Genital anomalies may include cryptorchidism, macroorchidism, shawl scrotum and, more rarely, hypospadias. Fertility is normal. Female carriers may have only a subtle phenotype with hypertelorism and widow's peak. Patients may present a neurodevelopmental phenotype with learning and behavioural disabilities that are often confined to early childhood. When present, mental impairment is rarely severe.

Although clinically and genetically heterogeneous, the best characterized form of the disorder is caused by mutations in the FGD1 gene (faciogenital dysplasia 1 gene; Xp11.21). Other gene(s) might be involved since most familial cases still do not have identified genetic cause.

Clinical diagnosis is based on physical examination and the recognition of the most distinctive clinical hallmarks. Molecular genetics, based on analysis of the FGD1 gene, may confirm diagnosis.

When molecular diagnosis is not conclusive, all possible options for differential diagnosis should still be considered, including Noonan syndrome, SHORT syndrome, pseudohypoparathyroidism and Robinow syndrome (see these terms).

Prenatal diagnosis for pregnancies at increased risk is technically possible when the disease-causing mutation in the family is known (the majority of mutations are family specific). However, prenatal testing is unlikely to be requested frequently, because usually physical signs can be mild and the clinical heterogeneity makes difficult a prediction of the phenotype, even within the same family.

AAS is an X-linked disease, but autosomal dominant and autosomal recessive transmissions have also been reported. Genetic counseling thus requires in depth investigation of patient's family history.
There is no curative treatment for AAS. Preliminary results of growth hormone administration in childhood do not seem to show a significant effect. Learning problems and attention deficit and hyperactivity disorder (ADHD), in case, may require a neuropsychiatric intervention.

The majority of patients present a good prognosis. Typically, they have a good evolution into adulthood with an age-related improvement of mental status. http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=915

**Research**

**Systematic Reviews & Meta Analyses**
None

**Literature Reviews**
None

**Randomised Controlled Trials**
None

**Other Research**

**Skeletal-specific expression of Fgd1 during bone formation and skeletal defects in Faciogenital Dysplasia (FGDY; Aarskog syndrome)**

Citation: Developmental Dynamics, 2000, vol./is. 218/4(573-586), 1058-8388 (2000)

Author(s): Gorski J.L.; Estrada L.; Hu C.; Liu Z.

Abstract: FGD1 encodes a guanine nucleotide exchange factor (GEF) that specifically activates the Rho GTPase Cdc42; FGD1 mutations result in Faciogenital Dysplasia (FGDY, Aarskog syndrome), an X-linked developmental disorder that adversely affects the formation of multiple skeletal structures. To further define the role of FGD1 in skeletal development, we examined its expression in developing mouse embryos and correlated this pattern with FGDY skeletal defects. In this study, we show that Fgd1, the mouse FGD1 ortholog, is initially expressed during the onset of ossification during embryogenesis. Fgd1 is expressed in regions of active bone formation in the trabeculae and diaphyseal cortices of developing long bones. The onset of Fgd1 expression correlates with the expression of bone sialo-protein, a protein specifically expressed in osteoblasts at the onset of matrix mineralization; an analysis of serial sections shows that Fgd1 is expressed in tissues containing calcified and mineralized extracellular matrix. Fgd1 protein is specifically expressed in cultured osteoblast and osteoblast-like cells including MC3T3-E1 cells and human osteosarcoma cells but not in other mesodermal cells; immunohistochemical studies confirm the presence of Fgd1 protein in mouse calvarial cells. Postnatally, Fgd1 is expressed more broadly in skeletal tissue with expression in the perichondrium, resting chondrocytes, and joint capsule fibroblasts. The data indicate that Fgd1 is expressed in a variety of regions of incipient and active endochondral and intramembranous ossification including the craniofacial bones, vertebrae, ribs, long bones and phalanges. The observed pattern of Fgd1 expression correlates with FGDY skeletal manifestations and provides an embryologic basis for the prevalence of observed skeletal defects.
The observation that the induction of Fgd1 expression coincides with the initiation of ossification strongly suggests that FGD1 signaling plays a role in ossification and bone formation; it also suggests that FGD1 signaling does not play a role in the earlier phases of skeletogenesis. With the observation that FGD1 mutations result in the skeletal dysplasia FGDY, accumulated data indicate that FGD1 signaling plays a critical role in ossification and skeletal development.

**Aarskog syndrome associated with hypermetropia and toe anomaly**

Citation: South African medical journal = Suid-Afrikaanse tydskrif vir geneeskunde, December 1997, vol./is. 87/12(1699-1700), 0256-9574 (Dec 1997)

Author(s): Caksen H.; Kurtoglu S.; Ciftci A.; Cagil N.; Gikrikci V.

Language: English

Abstract: Aarskog syndrome is characterised by a disproportionately short stature and facial, skeletal and urogenital anomalies ('shawl' scrotum and cryptorchidism). Ophthalmic findings include a slight downward slant to the palpebral fissures, hypertelorism, blepharoptosis, strabismus, ophthalmoplegia, hypermetropic astigmatism and a large cornea. Findings on the extremities include joint hyperextensibility, short and broad hands, interdigital webbing, a short fifth finger, clinodactyly and broad feet with bulbous toes. We report on a 7 1/2-year-old boy with typical findings of Aarskog syndrome, hypermetropia and bilateral proximal implantation of the fifth toes. These associated abnormalities have hitherto never been described, to our knowledge.

**Aarskog syndrome in association with mental and psychological retardation, grand mal epilepsy and tardive dyskinesia and apparent radicular paralysis of the fibular nerve in torsion scoliosis [German]**

Citation: Padiatrie und Grenzgebiete, 1993, vol./is. 31/5(345-351), 0030-932X (1993)

Author(s): Fehlow P.; Miosge W.; Walther F.

Language: German

Abstract: The case of an about 22-year-old patient with AARSKOG-syndrome is reported whose scoliosis caused a radicular impairment with resulting severe paresis of the right fibular nerve. The syndrome was also associated with moderate mental retardation, epilepsy, premature craniosynostosis and tardive dyskinesias after neuroleptic therapy because of disorders of behaviour. The risks of idiopathic scoliosis in patients with dysmorphia-retardation syndromes should be taken into consideration and such children should be supervised by an orthopaedist.

Publication Type: Journal: Article

Source: EMBASE
Metatarsus adductus in two brothers with Aarskog syndrome
Citation: Journal of Medical Genetics, 1983, vol./is. 20/6(477), 0022-2593 (1983)
Author(s): Hurst D.L.
Abstract: In 1970, Aarskog described a new syndrome, the facial-digital-genital syndrome, involving seven family members. The syndrome consists of short stature, characteristic facies, 'saddle bag' scrotum, and brachydactyly with mild interdigital webbing. Other features have been less constant, encompassing the skeletal and connective tissue systems. To date, none of the Aarskog patients has had bony deformities requiring surgical correction. A patient with the Aarskog syndrome and severe metatarsus adductus necessitating surgical correction is presented. The patient's brother had the identifying features of the Aarskog syndrome and had a similar foot deformity surgically corrected at the age of 5 years. The mother, with partial features of the syndrome, a widow's peak, and mild hypertelorism, had a mild adductus.
Publication Type: Journal: Article
Source: EMBASE
Full Text: Available from Highwire Press in Journal of medical genetics
Available from National Library of Medicine in Journal of Medical Genetics
Available from Highwire Press in Journal of Medical Genetics

A case of Aarskog syndrome with a review of Japanese literature
Citation: Hiroshima Journal of Medical Sciences, 1982, vol./is. 31/2(87-90), 0018-2052 (1982)
Author(s): Aihara K.; Nishi Y.; Usui T.
Language: English
Abstract: A 9-month-old boy with Aarskog syndrome is described. Eleven cases of this syndrome were collected from the Japanese literature and reviewed. The most characteristic findings are short stature, craniofacial anomalies such a hypertelorism and broad nasal bridge, hand and foot anomalies, and shawl scrotum.
Publication Type: Journal: Article
Source: EMBASE