"Data Base"

"Genodermatoses in Kuwait Published from Kuwait and from Elsewhere"

1. **Skin Manifestations in Primary Immunodeficient Children.**

   Al-Herz W, Nanda A.

   Pediatr Dermatol. 2011 Mar 31. doi: 10.1111/j.1525-1470.2011.01409.x. [Epub ahead of print] PMID: 21453308 [PubMed - as supplied by publisher]

2. **Identical glycine substitution mutations in type VII collagen may underlie both dominant and recessive forms of dystrophic epidermolysis bullosa.**

   Almaani N, Liu L, Dopping-Hepenstal PJ, Lai-Cheong JE, Wong A, Nanda A, Moss C, Martinéz AE, Mellerio JE, McGrath JA.

   Acta Derm Venereol. 2011;91(3):262-6. PMID:21448560 [PubMed - indexed for MEDLINE]

3. **Parental consanguinity and the risk of primary immunodeficiency disorders: report from the Kuwait National Primary Immunodeficiency Disorders Registry.**

   Al-Herz W, Naguib KK, Notarangelo LD, Geha RS, Alwadaani A.

   Int Arch Allergy Immunol. 2011;154(1):76-80. Epub 2010 Jul 27. PMID: 20664281 [PubMed - indexed for MEDLINE]

4. **Multiple milia in a newborn with congenital malformations: oral-facial-digital syndrome type 1.**

   Nanda A, Sharaf A, Alsaleh QA.

   Pediatr Dermatol. 2010 Nov-Dec;27(6):669-70. PMID: 21510010 [PubMed - in process]

5. **Congenital Yellow Nail Syndrome: A Case Report and Its Relationship to Nonimmune Fetal Hydrops.**

   Nanda A, Al-Essa FH, El-Shafei WM, Alsaleh QA.

   Pediatr Dermatol. 2010; 27(5): 533-4. PMID: 20807364 [PubMed – indexed for MEDLINE]

6. **Benign familial pemphigus (Hailey-Hailey disease) responsive to low dose cyclosporine.**

   Nanda A, Khawaja F, Harbi R, Nanda M, Dvorak R, Alsaleh QA. **Indian J Dermatol Venereol Leprol. 2010**;76(4):422-4. No abstract available. PMID: 20657133 [PubMed - indexed for MEDLINE]

7. **GAPO syndrome: a report of two siblings and a review of literature.**

   Nanda A, Al-Ateeqi WA, Al-Khawari MA, Alsaleh QA, Anim JT.

   Pediatr Dermatol. 2010 1;27(2):156-61. Review. PMID: 20537066 [PubMed - indexed for MEDLINE]

8. **Homozygous mutations in the 5' region of the JUP gene result in cutaneous disease but normal heart development in children.**

   Cabral RM, Liu L, Hogan C, Dopping-Hepenstal PJ, Winik BC, Asial RA, Dobson R, Mein CA, Baselaga PA, Mellerio JE, Nanda A, Boente Mdel C, Kelsell DP, McGrath JA, South AP.

   J Invest Dermatol. 2010;130(6):1543-50. Epub 2010 Feb 4. PMID: 20130592 [PubMed - indexed for MEDLINE]

9. **Mutations in PYCR1 cause cutis laxa with progeroid features.**

Reversade B, Escande-Beillard N, Dimopoulou A, Fischer B, Chng SC, Li Y, Shboul M, Tham PY, Kayserili H, Al-Gazali L, Shahwan M, Brancati F, Lee H, O'Connor BD, Schmidt-von Kegler M, Merriman B, Nelson SF, Masri A, Alkazaleh F, Guerra D, Ferrari P, Nanda A, Rajab A, Markie D, Gray M, Nelson J, Grix A, Sommer A, Savarirayan R, Janecke AR, Steichen E, Sillence D, Hausser I, Budde B, Nürnberg G, Nürnberg P, Seemann P, Kunkel D, Zambruno G, Dallapiccola B, Schuelke M, Robertson S, Hamamy H, Wollnik B, Van Maldergem L, Mundlos S, Kornak U.

Nat Genet. 2009;41(9):1016-21. Epub 2009 Aug 2. PMID: 19648921 [PubMed - indexed for MEDLINE]

10. **Germline mutation in the von Hippel-Lindau gene in Kuwait: a clinical and molecular study.**

AlFadhli SM, Mohammed B, Yassin A.

Med Princ Pract. 2008;17(5):395-9. Epub 2008 Aug 6. PMID: 18685280 [PubMed - indexed for MEDLINE]

11. **Susceptible and protective endothelial nitric oxide synthase gene polymorphism in alopecia areata in the Kuwaiti population.**

Alfadhli S, Kharrat NJ, Al-Tememy B, Nanda A, Rebai A.

Autoimmunity. 2008;41(7):522-5. PMID: 18608176 [PubMed - indexed for MEDLINE]

12. **Autoimmune diseases associated with neurofibromatosis type 1.**

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13. **Primary immunodeficiency disorders in Kuwait: first report from Kuwait National Primary Immunodeficiency Registry (2004--2006).**

Al-Herz W.

J Clin Immunol. 2008;28(2):186-93. Epub 2007 Nov 16. PMID: 18008151 [PubMed - indexed for MEDLINE]

14. **Gerodermia osteodysplastica/wrinkly skin syndrome: report of three patients and brief review of the literature.**

Nanda A, Alsaleh QA, Al-Sabah H, Marzouk EE, Salam AM, Nanda M, Anim JT.

Pediatr Dermatol. 2008;25(1):66-71. Review. PMID: 18304158 [PubMed - indexed for MEDLINE]

15. **Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2.**

Kornak U, Reynders E, Dimopoulou A, van Reeuwijk J, Fischer B, Rajab A, Budde B, Nürnberg P, Foulquier F; ARCL Debré-type Study Group, Lefeber D, Urban Z, Gruenewald S, Annaert W, Brunner HG, van Bokhoven H, Wevers R, Morava E, Matthijs G, Van Maldergem L, Mundlos S.

Nat Genet. 2008;40(1):32-4. Epub 2007 Dec 23. PMID: 18157129 [PubMed - indexed for MEDLINE]

16. **A novel mutation in the VDR gene in hereditary vitamin D-resistant rickets.**

Arita K, Nanda A, Wessagowit V, Akiyama M, Alsaleh QA, McGrath JA.

Br J Dermatol. 2008;158(1):168-71. Epub 2007 Oct 26. No abstract available. PMID: 17970811 [PubMed - indexed for MEDLINE]

17. **Anterior cervical hypertrichosis (hairy throat): is it a sign to worry about?**

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Clin Exp Dermatol. 2007;32(1):112-4. Epub 2006 Sep 27. No abstract available. PMID: 17004983 [PubMed - indexed for MEDLINE]

18. **Keutel syndrome with overlapping features of cutis laxa: a new variant.**

Nanda A, Anim JT, Al-Gareeb M, Alsaleh QA.

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19. **Surgical treatment of an unusual patient with Proteus syndrome.**

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J Craniofac Surg. 2006;17(1):175-8. PMID: 16432429 [PubMed - indexed for MEDLINE]

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Nanda A, Lionel J, Al-Tawari AA, Anim JT.

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25. **Infantile systemic hyalinosis.**

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J Craniofac Surg. 2003;14(5):719-23. PMID: 14501336 [PubMed - indexed for MEDLINE]

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34. **A new variant of 1,25-Dihydroxyvitamin D-receptor-defect rickets and alopecia associated with mental retardation and hearing loss.**

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Hanna RM, Dahniya MH, al-Marzouk N, Grexa E.

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36. **Gerodermia osteodysplastica in a Bedouin sibship: further delineation of the syndrome.**

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Clin Dysmorphol. 1997;6(1):51-5. PMID: 9018419 [PubMed - indexed for MEDLINE]

37. **Hand anomalies in fetal-hydantoin syndrome: from nail/phalangeal hypoplasia to unilateral acheiria.**

Sabry MA, Farag TI.

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39. **Familial richner-Hanhart syndrome in Kuwait: twelve-year clinical reassessment by a multidisciplinary approach.**

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40. **Autosomal recessive disorders among Arabs: an overview from Kuwait.**

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43. Further delineation of Costello syndrome.

Teebi AS, Shaabani IS.

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51. **Craniofacial anomalies, abnormal hair, camptodactyly, and caudal appendage.**

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54. **New autosomal recessive faciodigitogenital syndrome.**

Teebi AS, Naguib KK, Al-Awadi S, Al-Saleh QA.

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55. **Primary hypogonadism and partial alopecia in three sibs with müllerian hypoplasia in the affected females.**

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Created in: ***June, 2011***