NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

1: Klippel Trenaunay and Proteus Syndrome overlap--a diagnostic dilemma, EDOJ5(2): 10

Editor Information. 1. Emeritus Consultant Dermatologist, Leicester Royal Infirmary, Leicester. 2. Consultant Dermatologist and Senior Lecturer, St John's Institute of Dermatology, St Thomas' Hospital, London, and Consultant Dermatologist, Epsom & St Helier University Hospitals NHS Trust, Epsom, Surrey.

Becker naevus What causes epidermal naevi? If one of these populations of skin cells is abnormal it results in localised areas of thickened skin. Luckily, epidermal naevi very rarely affect more than one member of the family. Epidermal naevi are distributed along the lines of Blashko. These lines are the tracks taken by groups of genetically identical cells in the developing embryo. Skin cells that have the active abnormal gene spread out to form the epidermal naevus, whereas the remaining skin cells form the other areas of apparently normal skin. New research has found point mutations in keratin genes that support this theory. The abnormal gene is found in the epidermal naevus cells but not in the normal skin. The same keratin 1 and keratin 10 gene abnormalities have been found in parents who have epidermolytic epidermal naevus and in their offspring who have bullous ichthyosiform erythroderma a rare form of ichthyosis. So the epidermolytic epidermal naevus is thought to be a mosaic form of this type of ichthyosis. The ATP2A2 gene abnormality that arises in Darier disease has been detected in the affected cells of a patient with acantholytic epidermal naevus, so this type may be a mosaic form of Darier disease. Linear porokeratosis may be a mosaic form of disseminated superficial actinic porokeratosis. The majority are linear epidermal naevi, i. When they first appear at birth or in infancy they are flat tan or brown marks but as the child ages they become thickened and often warty. The naevus may also become more extensive for a few years. Another name for a linear epidermal naevus is naevus unis lateralis. Systematised epidermal naevi are less common and are sometimes known as ichthyosis hystrix. There are multiple lesions that usually arise in a swirled pattern, arising on one or both sides of the body. In some patients, there are also other congenital abnormalities, particularly of the skeleton and central nervous system ENS. On skin biopsy, the epidermal naevus shows increased thickness of the epidermis. The rare epidermolytic epidermal naevus subtype is characterised by a specific histological pattern called epidermolytic hyperkeratosis and resembles that seen in bullous ichthyosiform erythroderma. Acantholytic epidermal naevus has pathology that resembles Darier disease. Linear porokeratosis has pathology that resembles disseminated superficial actinic porokeratosis. Epidermal naevi Epidermal naevus syndromes The epidermal naevus syndromes refer to the association of an epidermal naevus with abnormalities in other organ systems derived from the embryonic ectoderm. These syndromes may involve the eyes, bones or nervous system. Many specific syndromes have been described. The defect causing the skin lesions may also result in disorders of other internal organs such as the brain, eyes and skeleton. When multiple organs are involved the condition is referred to as an epidermal naevus syndrome ENS.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

2: Internet Scientific Publications

Naevi and other Developmental Defects Tony Burns MB, BS, FRCP Emeritus Consultant Dermatologist 3, Stephen Breathnach MA, MB, BChir, MD, PhD, FRCP Consultant Dermatologist 4,5.

C ongenital defects of the scalp and skull are rare anomalies. The literature regarding these lesions usually suggests that the appropriate treatment is surgical. Her cranial vault and cutaneous defects have become fully reconstituted without surgical intervention. Case Report The mother of the patient had pelvic disproportion, and had undergone previous Caesarean sections. She had been pregnant three times, with one previous live birth and one abortion. She experienced minor first-trimester bleeding with this pregnancy. She weighed gm. Her head circumference was 32 cm. Abnormal physical findings were restricted to the integument and skull. The defect was covered by a hairless, grayish, vascular, very thin parchment-like membrane, which extended over the entire vault and covered an opaque vascularized tissue, which was thought to represent dura mater. There was no visible cerebral cortex or cerebrospinal fluid. The sagittal sinus could not be identified. Scalp and skull defect at birth. Top view of scalp showing the extent of the defect. The vascular pattern resembled dura, although no saggital sinus could be seen. A narrow strip of epidermis can be seen bordering the normal scalp. Anteroposterior left and lateral right views showing the extent of the bone defect in the frontoparietal region. Other cutaneous defects were noted extending from each axilla to the abdominal wall and over the buttocks and thighs bilaterally Fig. The skull defect included the entire vertex Fig. Initially, some consultants expressed the opinion that these defects should be immediately grafted to avoid severe infection or hemorrhage; however, the patient had no ready source of autogenous grafts and seemed to require full-thickness grafts if possible. It was suggested that a more conservative approach might be feasible. Reverse isolation was employed for 4 weeks. The surface of the scalp defects gradually epithelialized from the margins. Six weeks later, the patient was discharged with almost complete epithelialization of the scalp and trunk lesions. Skin defects at birth. Skin defect extending along shoulder, lateral chest wall, and abdomen. Similar defects involved the left side. Healed defect is seen over the right shoulder. Bilateral defects involving the buttocks also healed spontaneously without contracture. The trunk lesions had healed without contracture Fig. The bone grew from the edges of the skull defect toward the vertex. At 3 years, there was full skin coverage with a normal skull contour Fig. The child has developed normally. The patient at the age of 3 years. Face and profile pictures of the scalp show complete coverage with a full thickness of scalp free of cicatrix. Sparse hair growth is not evident in the illustration. Skull x-ray films illustrate the fully reconstituted cranial vault. Discussion Aplasia cutis congenita of the scalp was first described in , 7, 9, 22â€”24, 26, 28 and since then about cases have been reported. It is believed to be slightly more common in females and first-born infants. Pathologically, these defects may be covered with a flat layer of epithelium or a fibrous membrane devoid of either elastic fibers or skin append-ages. With larger defects that include the skull, the dura may be absent, and the overlying membrane may be covering arachnoid directly. No inflammation or necrosis is present in these lesions at birth. Many familial cases with an autosomal dominant preponderance and few with recessive trait have been cited as evidence of a genetic etiology. In addition, the origin of many of these associated anomalies as ectodermal or mesodermal is not fully worked out, and the incidence of each, associated with congenital scalp and skull defects, is poorly reported in the literature. Hence, a uniform hypothesis regarding etiology remains elusive. The nature of the defect may influence the choice of treatment. Fear of infection has led many authors to advocate early excision and primary repair of small defects. We allowed the skin defect in this patient to close spontaneously, using frequent changes of saline and antibiotic-soaked dressings. We did not think that systemic antibiotic prophylaxis was necessary. Careful protection against trauma and infection in the hospital initially led to adequate epithelialization and early dermal coverage. The patient was allowed to go home under the care of her parents who were trained during her initial period of hospitalization and continued the dressing regimen at home. Sparse hair growth appeared in the area of the scalp defect. Since these defects largely included the full skin thickness, essential baldness will remain. New bone formation gradually reduced the skull defect from the margins. By the end of 3 years,

only a small bone defect remains in the region of the anterior fontanel, emphasizing the fact that bone repair may also occur spontaneously. This illustration of a conservative approach to congenital scalp and skull defects has emphasized the natural course that these lesions may follow given adequate medical care and protection. Some small lesions may not close spontaneously and will require excision. But some very large ones may correct themselves with appropriate medical management and the passage of time.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

3: Embryonic tail Arun C Inamadar - Indian J Dermatol Venereol Leprol

Other developmental defects. (the phenomenon of two cell lines arising early in embryonal development), as it has been observed that many congenital naevi.

A mosaic activating mutation in AKT1 associated with the Proteus syndrome. N Engl J Med ; 7: Mosaicism of activating FGFR3 mutations in human skin causes epidermal nevi. J Clin Invest ; 8: An epidermal nevus syndrome with cerebral involvement caused by a mosaic FGFR3 mutation. Oncogenic PIK3CA mutations occur in epidermal nevi and seborrheic keratoses with a characteristic mutation pattern. Am J Hum Genet ;90 6: Keratinocytic epidermal nevi are associated with mosaic RAS mutations. J Med Genet ;49 4: HRAS mutation mosaicism causing urothelial cancer and epidermal nevus. N Engl J Med ; J Med Genet ;47 Phenotypic heterogeneity in bullous congenital ichthyosiform erythroderma: Arch Dermatol ; 9: Birthmark due to cutaneous mosaicism for keratin 10 mutation. Epidermal mosaicism producing localised acne: Porokeratotic eccrine nevus may be caused by somatic connexin26 mutations. J Invest Dermatol ; 9: Lethal genes surviving by mosaicism: J Am Acad Dermatol ;16 4: Br J Dermatol ; 6: A clinical, histological and immunohistochemical study. Acta Derm Venereol ;71 4: Linear nevus sebaceus with convulsions and mental retardation. Am J Dis Child ; Phacomatosis pigmentokeratotica is caused by a postzygotic HRAS mutation in a multipotent progenitor cell. J Invest Dermatol ; 8: G12S mutation causes woolly hair and epidermal nevi. J Invest Dermatol ; Am J Med Genet ;84 5: Cutaneous manifestations of proteus syndrome: Arch Dermatol ; 8: Evolution of skin lesions in Proteus syndrome. J Am Acad Dermatol ;52 5: Newly delineated syndrome of congenital lipomatous overgrowth, vascular malformations, and epidermal nevi CLOVE syndrome in seven patients. CNS malformations and seizures may be a component of this disorder. Br J Dermatol ; 1: Systemic epidermal nevus with involvement of the oral mucosa due to FGFR3 mutation. BMC Med Genet ; J Am Acad Dermatol ;64 5: Congenital hemidysplasia with ichthyosiform erythroderma and limb defects. Eur J Pediatr ; 1: Am J Med Genet ;90 4: J Am Acad Dermatol ;46 4: CHILD syndrome without hemidysplasia. Segmentally arranged basaloid follicular hamartomas with osseous, dental and cerebral anomalies: Acta Derm Venereol ;88 4: Bilateral nevus comedonicus syndrome. Yonago Acta Med ;56 2: Pediatr Dermatol ;15 4: Suvremenna Meditsina ;13 5: Management of nevus sebaceous and the risk of basal cell carcinoma: Pediatr Dermatol ;26 6: Pigmented epithelioid melanocytoma developed in a patient with Becker nevus syndrome. J Dermatol ;39 9: Becker naevus and malignant melanoma. Hum Mol Genet ;23 2: J Pediatr ;91 1: Semin Cutan Med Surg ;26 4: The group of epidermal nevus syndromes Part II. Less well defined phenotypes. J Am Acad Dermatol ;63 1: The group of epidermal nevus syndromes Part I. Neurologic complications of the epidermal nevus syndrome. Arch Neurol ;44 2: Treatment of verrucous epidermal nevus: Carbon dioxide laser treatment of epidermal nevi: Actas Dermosifiliogr ; J Invest Dermatol ; Generalized and naevoid epidermolytic ichthyosis in Denmark: Acta Derm Venereol ;93 3: Pediatr Dermatol ;30 3: The incidence and significance of birthmarks in a cohort of 4, newborns. Pediatr Dermatol ;1 1: A survey of birthmarks and cutaneous skin lesions in newborns. J Med Assoc Thailand ;96 Suppl. The incidence of birthmarks in the neonate. Epidemiology of congenital pigmented naevi: Incidence rates and relative frequencies. Br J Dermatol ; 3: J Pediatr ; 6: Part 1 â€" epidemiology, phenotype and outcomes. Large congenital melanocytic nevi and the risk for development of malignant melanoma and neurocutaneous melanocytosis. Pathology of Melanocytic Nevi and Malignant Melanoma. The histogenesis of acquired melanocytic nevi. Based on a new concept of melanocytic differentiation. Am J Dermatopathol ;6 Suppl. The melanocytic differentiation pathway in congenital melanocytic nevi: Pediatr Pathol ;8 3: Schwann cell precursors from nerve innervation are a cellular origin of melanocytes in skin.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

4: Aplasia cutis congenita - Wikipedia

Part 6, Reference for Chapter Congenital Naevi and Other Developmental Abnormalities Affecting the Skin Skip chapter table of contents and go to main content Hide navigation Overview.

P artial agenesis or neonatal ulceration of the skull and scalp is rare. They most commonly occur in the midline, but parietal and posterior auricular lesions also occur. Lesions vary in size from pinpoints to over 8 cm in diameter and may be single or multiple. The lesion may be described as a moist ulcerated defect with a base varying in depth from subcutaneous to the level of the arachnoid. They may occur in infants who are otherwise normal but also may accompany multiple congenital anomalies. Occurrence of the defects in siblings and in several generations of the same family suggests an inherited component. His patient died shortly after birth because of fatal hemorrhage from the superior sagittal sinus. Since then there have been about cases reported. In the past few years we have seen 12 such patients. Summary of Cases We classified our 12 patients into two groups based on the layers involved. Group 1 patients had a partial thickness defect involving the scalp only, Group 2 patients had a full thickness absence of scalp, periosteum, skull, and dura. Partial Thickness Defect Case 1 This newborn white girl was the product of a full-term uncomplicated delivery Fig. She had five scalp defects ranging in diameter from 0. Neurological examination and skull films were normal. There were no other congenital anomalies. The two larger lesions were treated by full thickness excision of the defect and underlying galea followed by primary closure. Five defects ranging in diameter from 0. Cases 2 and 3 These baby girls were identical twins, admitted because of bilateral parietal defects 1 cm from the midline, and 0. There were no associated congenital anomalies. Treatment was by excision and closure. Identical bilateral defects in one of identical twin girls. Treatment was by excision. Cases 4 This white baby boy had two defects; one parietal measuring 1. There were no associated anomalies. The larger lesion was excised; the smaller granulated and healed uneventfully. Skull films and neurological examination were normal. She also had a cleft lip and palate. Treatment was by excision and primary closure. She also had a midline cleft lip and palate, polydactyly, bilateral colobomas, holoprosencephaly, congenital heart disease, and renal anomalies. She died shortly after admission Fig. Case 7 This newborn white girl had several small midline defects in the parietal area. She was a microcephalic child with hypotelorism, bilateral colobomas, and bilateral simian creases. Chromosome analysis demonstrated trisomy D. Treatment was by dressing changes. Case 8 This 2-year-old black girl had been born with multiple lesions of the scalp. She also was severely retarded, and had a lumbosacral lipomyelomeningocele, malrotation of the colon, bilateral corneal opacities, congenital glaucoma, renal anomalies, and congenital heart disease. The lesions had granulated in and healed spontaneously. Sagittal sinus and cortex were visible through the arachnoid. The neurological examination was normal. Treatment was by mobilization of the skin and primary closure. Group 2 full thickness defect. The sagittal sinus and cortex are visible through the arachnoid. There was a large venous bleb just off the midline. Neurological examination was normal; there were no other congenital anomalies. Because of the thickness of the eschar, this lesion was initially treated with dressings. However, when bleeding occurred from the venous bleb, the child was taken to the operating room and the lesion was repaired. Surgicel was placed over the bleb and a periosteal flap swung over this. Skin flaps were then rotated to cover the defect. Large Group 2 defect. Note the venous bleb just off the midline. This was the source of a near fatal hemorrhage. Case 11 This newborn white boy had multiple congenital anomalies, including trisomy D shown by chromosome analysis. There was an irregular horseshoe-shaped defect in the midline-midparietal area with cortex visible through the arachnoid Fig. Associated anomalies included choanal atresia, micrognathia, and supranumerary digits on both hands and the left foot. Treatment was with dressings. The child died shortly after birth. Irregular horseshoeshaped defect in a child with trisomy D syndrome. Case 12 This newborn white boy had a 6cm horseshoe-shaped defect in the midline. Cerebral cortex was visible under the arachnoid. The child also had bilateral congenital hip dislocation, choanal atresia, ambiguous genitalia, imperforate anus, and laryngomalacia. Treatment was with dressings only, and the child died. Discussion The majority of our cases fell into our Group 1 classification. There was a slight predominance of females 7: We have seen cleft lip and

palate, polydactyly, colobomas, congenital heart disease, holoprosencephaly, simian creases, lipomyelomeningocele, glaucoma, renal anomalies, choanal atresia, ambiguous genitalia, and laryngomalacia. Four of our 12 cases had trisomy 13â€" Histological Appearance Microscopic section of the lesions excised in Group 1 showed that none contained the stratified squamous epithelium characteristic of scalp. The outer surface was covered by a layer of flat, thin cells. The remainder of the lesion was a collagenous matrix with signs of hemorrhage. There were no flat cells, inflammatory cells, nor skin appendages, such as hair follicles or sebaceous glands. Etiology Many theories have been proposed for the etiology of this lesion. Hoffman 9 in postulated that intrauterine amniotic adhesions formed between the amnion and the skull early in development. With the accumulation of amniotic fluid these bands are torn apart, leaving the skin defect. This theory seems difficult to accept. It is unlikely that such an adhesion would occur in siblings or in successive generations; it is also unlikely that the lesion would occur in the same location in most of the reported cases. For the same reasons, trauma is an unlikely explanation. Pressure necrosis of the scalp during labor has been postulated, 1 but but is not confirmed by microscopic studies. Greig 7 postulated an arrest of midline development in these children; Walz 22 postulated a defective closure of the neural tube. This may be an example of the neuroschistic process described by Padget 15 in which there is incomplete healing and fusion of the nourishing mesoderm. A healthy mesoderm seems to be necessary for proper development and maintenance of the ectoderm. Treatment Treatment of this lesion, as in other congenital anomalies, is based on the total clinical problem. If the child is severely damaged, no surgical treatment is given; chromosome analysis is carried out routinely to aid in this decision. In patients who are felt to be normal but have large scalp defects, therapy consists of primary closure of galea and skin with excision of the lesion when possible. The smaller lesions heal spontaneously with minimal scarring and no surgical therapy is indicated. Surgical intervention is directed to prevent hemorrhage Case 10 , to prevent infection, to protect the underlying brain, and to improve appearance. In the Group 2 lesions, hemorrhage and infection meningitis are the major immediate complications. Infection was not a problem in our series but has been in other cases. As suggested by others 12, 13 we have excised the margins of the lesion or the entire lesion when possible, and closed primarily. When a large area of bone is missing, pericranium is rotated to fill the defect. We have not used split thickness skin grafts. To avoid a large area of alopecia, hair-bearing skin is rotated over the defect. We have been able to protect the brain adequately in all necessary cases. Eleven years later, the patient in Case 9 had only a small 0. We have not seen an excessively large defect requiring a plastic cap as described by Matson.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

5: Bilateral Becker's nevi Bansal R, Sen R - Indian J Dermatol Venereol Leprol

Read chapter 2 Developmental Defects and Their Causes: Scientific Frontiers in Developmental Toxicology and Risk Assessment reviews advances made during t.

Pattern of Naevi in Children in South India. The Internet Journal of Dermatology. Review A total of 4, paediatric cases below the age of 14 years attending the dermatology OPD of our hospital during the study period from August to August were screened for naevi. Out of these, 77 1. Various naevi recorded in decreasing order of frequency were melanocytic naevi 41 cases, The mean age of these cases was 2. Out of the total of 41 cases of melanocytic naevi, 30 Amongst 19 vascular naevi cases, 17 were of haemangiomas of infancy and the remaining two were of Port-wine stain. The subtypes of epidermal naevi included were linear verrucous epidermal naevi 5 cases , naevus sebaceous 4 cases , systematized epidermal naevus 3 cases , inflammatory linear verrucous epidermal naevus 2 cases and one case each of naevus comedonicus and linear porokeratosis. Discussion A naevus is a localized, highly differentiated, proliferative malformation arising from keratinocytes, melanocytes, or appendageal i. These naevi are best described by its origin or location, such as melanocytic naevus, sebaceous naevus, or systematized naevus. In a cross-sectional study in two hospitals in the city of Belo Horizonte, neonatal dermatoses of clinical relevance congenital melanocytic nevus, sebaceous nevus, cafe-au-lait spots, Port-wine stain, ash leaf macules were found in 42 5. Naevi and other developmental defects. Blackwell Science Limited Neonatology-Pathophysiology and management of the Newborn, 5th edn. Dogra S, Kumar B. Epidemiology of skin diseases in school children: Pediatr Dermatol ; 20 6: Skin diseases in Chinese children at a pediatric dermatology center. Pediatr Dermatol ; 21 2: Prevalence of skin disorders in school children in Ibadan, Nigeria. Pattern of pediatric dermatoses in a referral center in South India. Indian Pediatr ; Pereira LB, Gontijo B. Neonatal dermatoses of relevant medical significance: J Pediatr Rio J ; 75 5:

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

6: CONGENITAL NAEVI - Rooks Textbook of Dermatology

Naevi and other Developmental Defects. Rook's Textbook of Dermatology, A Burkhead, G Poindexter, D S Morrell. () A case of extensive Aplasia Cutis.

A crude distinction can be made between intrinsic and extrinsic causes. Intrinsic causes include genetic defects mutations , endogenous chromosomal imbalances e. Extrinsic causes include the enormous variety of environmental inputs such as infection, nutritional deficiencies and excesses, life-style factors e. Other environmental factors, such as hyperthermia, ultraviolet irradiation, and X-rays, should be included. As noted before, developmental defects comprise all structural and functional deficits detected in the implanted embryo, fetus, neonate, infant, or child. The committee was asked to consider environmental agents that might cause developmental defects. Such agents include mercury, lead, and polychlorinated biphenyls. Natural plant and animal products and toxins have long been recognized as agents that can cause toxicity. They were some of the first environmental agents to be identified as teratogens. Agents can enter the environment by either deliberate e. Pharmaceuticals and food additives generally would not be considered environmental agents; however, many of the issues under consideration for environmental agents can also apply to these agents. Additionally, it is possible that they incidentally enter the environment at significant concentrations and become environmental agents. What fraction of developmental defects can be attributed to extrinisic or intrinsic causes? Page 21 Share Cite Suggested Citation: The National Academies Press. The infants were in a surveillance program at a university hospital and were not from the population at large and, therefore, these percent-ages should be viewed cautiously. The term is used when geneological studies indicate that a physical trait, disease, or developmental defect occurs at a higher rate within families than expected in the general population, but the patterns of inheritance do not follow strict Mendelian segregation rules. To explain the departure from Mendelian rules, the genetic variant of a gene is said to predispose the individual, but further circumstances, either environmental or other genetic factors, are needed for the production of the disease. An example of multifactorial inheritance is the relationship between maternal smoking, transforming growth factor TGF polymorphisms, and oral cleft Hwang et al. This example is described in detail in Chapter 5. Such a departure from Mendelian rules might be attributable to environmental factors, but the departure could as well be due to the requirement for a combination of particular alleles of two or more genes to produce the trait a polygenic trait or to genomic imprinting. Specific genes and environmental exposures have been associated in multifactorial inheritance in only a few instances, but increased information is becoming available. As the Nelson and Holmes figures indicate, however, the knowledge about the causes and prevention of developmental defects continues to be limited Mattison It is generally recognized that extrinsic agents probably have acted as human developmental toxicants and that more than 1, chemical and physical agents produce developmental defects in experimental animals Shepard ; Schardein It should be noted that much of the developmental toxicity testing on experimental animals was conducted at up to maternally toxic doses and, therefore, observed effects at those doses might not be the same as effects observed after exposure to environmentally relevant doses. It is not known how many of the 1, agents actually produce developmental defects in humans, and the figure is not obtainable by direct testing in humans. In light of the experimental animal results, many of the agents have never entered the marketplace or environment, and others are handled with great caution according to preventive public-health and workplace-safety guidelines. Page 22 Share Cite Suggested Citation: Table lists several representative human developmental toxicants. There is information available on proposed mechanisms of action for these toxicants, however, it is infrequently synthesized into a cohesive and comprehensive mechanistic explanation. For example, the epidemiological methods for identifying toxicants are inherently insensitive and depend on the systematic examination of large human populations. Such large-scale examination is difficult to do Selevan Thalidomide was an exception. Because it caused such a distinctive outcome bilateral limb shortening of a rare human malformation, its effects were recognized in small patient groups. Even though the frequency of fetal alcohol syndrome is high compared with other developmental disorders, it took many years to identify alcohol as a human teratogen because the

physical alterations are subtle, and the learning and social adjustment problems are sometimes not detectable until several years after birth. When a human exposure problem is suspected, epidemiological testing can be performed to assess toxicity, but few of the 1, agents have been so examined. Also, the number of agents that cause developmental toxicity might be higher if the multifactorial inheritance category of birth defects contains, as indeed is suspected, cases of human variants who are genetically more susceptible predisposed to particular environmental conditions than are others. Finally, the number might be higher if some toxicants extrinsic causes produce malformations as a consequence of their primary effect in causing genetic damage intrinsic cause. In conclusion, although it is recognized that environmental agents can, and some do, act as developmental toxicants, it is still unclear how large a role these agents play in producing human congenital anomalies relative to other sources of developmental toxicants such as pharmaceuticals and food additives, and relative to intrinsic causes such as genetic differences. This variety is theoretically infinite if no limit is set on the molecular size of chemicals, because new and more complex compounds can always be made by coupling together simpler chemical units.

7: Facts About Developmental Disabilities | CDC

The exact cause of Klippel-Trenaunay syndrome remains to be elucidated, although several theories exist like intrauterine damage to the sympathetic ganglia or intermediolateral tract, or deep vein abnormalities, or a mesodermal defect during fetal development.

Klippel Trenaunay syndrome is a rare mixed vascular malformation characterized by a triad of port wine stain, varicose veins, and bony, soft tissue hypertrophy involving an extremity [1]. A rare case involving face and all the limbs is being reported. This is a case of interest also because of having clinical overlap with Proteus syndrome. Introduction The exact cause of Klippel-Trenaunay syndrome remains to be elucidated, although several theories exist like intrauterine damage to the sympathetic ganglia or intermediolateral tract, or deep vein abnormalities, or a mesodermal defect during fetal development. Most cases are sporadic, although a few cases in the literature report an autosomal dominant pattern of inheritance [2]. This syndrome is characterized by a triad of extensive capillary malformation, underlying venous varicosities, and underlying soft-tissue or bony hypertrophy. The port-wine stain is typically of the nevus flammeus type. Other vascular malformations can be angiokeratomas, angiodermatitis or even lymphangioma circumscriptum [3]. The lower limb is the site of malformation in the majority of patients. The hypertrophy involves the length as well as the circumference of the involved extremity and is caused by local hyperemia and venous stasis secondary to the vascular anomaly [4]. Proteus syndrome is a hamartomatous disorder which is characterized by asymmetrical overgrowth of any part of the body, verrucous epidermal naevi, vascular malformations and lipoma like subcutaneous hamartomas. Macrodactyly has been regarded characteristic. Often, cerebriform thickening of the soles and palms is also present [5]. Case report A 20 years old female born out of non- consanguineous parents presented with a history of hemihypertrophy of left side of face, tongue and also disproportionate hypertrophy of all the limbs, more pronounced on left side since birth. She also had history of presence of Macrodactyly of left middle finger; port wine stain over the trunk, and all the limbs which did not show any change in colour till now. Patient also had venous varicosities over both the lower limbs as well as left upper limb since early childhood. Since last two years the patient had developed grouped vesiculo-papular lesions on left lower leg with history of frequent bleeding from them. There was no family history of such lesions and no history of any other significant illness in patient. On cutaneous examination hemihypertrophy of the left side of face including the palate and tongue was present with a linear central depression over the left side of forehead. Showing hemihypertrophy of the tongue. Showing macrodactyly of left middle finger. Hypertrophy of both upper and lower limbs was present more pronounced on the left side. Port wine stain on back and scoliosis. Showing extensive port wine stain. Dilated tortuous veins were present on all the four limbs. Similar growths were diffusely present over the left popletial fossa, dorsum of left foot and also the ankle. Showing syndactyly of 2nd and 3rd toes and velvety plaques. Both the lower legs showed non pitting type of oedema. All routine investigations were in normal limits. X ray both feet confirmed the syndactyly and showed overgrowth of 2nd left metatarsal. X ray both hands showed overgrowth of left ring finger phalanges. Colour Doppler both lower limbs showed varicosities and sapheno-femoral incompetence. Showing histopathology suggestive of lymphangioma circumscriptum. Discussion In Klippel Trenaunay syndrome characteristic cutaneous lesions comprise of port wine stain which are usually present at birth. They occur on the affected limb but may sometimes extend beyond it involving several limbs. They have a tendency to stop in the midline. Various other rare vascular malformations include- small angio-keratomas, angiodermatitis, lymphangioma circumscriptum, lymphoedema. The hypertrophy present is of more commonly legs than arms and involves the growth of soft tissue and the bone. Rarely involvement of the face may be seen. Compensatory scoliosis is present when there is a leg length difference. Venous varicosity of the affected limb is a common presentation which often presents in the early childhood. There is a high risk of deep vein thrombosis to occur. Other associated features include- verrucous epidermal naevi, polydactyly, syndactyly [5]. Proteus syndrome comprises characteristically of asymmetrical hypertrophy of face, part or whole of one or both limbs, trunk. Macrodactyly is another characteristic feature. Soft tissue growth over the palms and soles

in a cerebriform pattern is found [6 , 7]. The three main types of skin findings include- epidermal naevi which is generally of linear verrucous variety, vascular malformations which are similar to Klippel Trenaunay syndrome, and soft subcutaneous masses which are highly characteristic and may represent complex hamartomatous malformations. Other associated features include scoliosis, kyphosis, cataract, strabismus, hypodontia with normal intelligence [5]. Our patient whose clinical features are summarized in Table -1 exhibits the characteristic feature of hemihypertrophy of limbs a feature shared by both the syndromes but involvement of face and all the limbs favours Proteus syndrome. Presence of port- wine stain is again a feature common to both but its extensive involvement is a rarer presentation in Klippel Trenaunay syndrome than in proteus syndrome. Varicosities is present in both syndromes but more so in Klippel Trenaunay syndrome. Macrodactyly is a characteristic feature suggesting Proteus whereas presence of syndactyly and lymphangioma circumscriptum suggested more likely a possibility of Klippel Trenaunay Syndrome. Absence of verrucous naevi and sub cutaneous lipoma like nodules negated Proteus syndrome.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

8: Nonsurgical approach to congenital scalp and skull defects : Journal of Neurosurgery

Birthmarks present at birth or soon after are a source of parental anxiety. Moss c, Shahidullah h. Naevi and other developmental defects. In: Burns T, Breathnach.

Minus Related Pages Developmental disabilities are a group of conditions due to an impairment in physical, learning, language, or behavior areas. Children reach milestones in how they play, learn, speak, behave, and move for example, crawling and walking. However, the developmental milestones give a general idea of the changes to expect as a child gets older. As a parent, you know your child best. At each well-child visit, the doctor looks for developmental delays or problems and talks with the parents about any concerns the parents might have. This is called developmental monitoring. Any problems noticed during developmental monitoring should be followed up with developmental screening. Developmental screening is a short test to tell if a child is learning basic skills when he or she should, or if there are delays. If a child has a developmental delay, it is important to get help as soon as possible. Most developmental disabilities begin before a baby is born, but some can happen after birth because of injury, infection, or other factors. Most developmental disabilities are thought to be caused by a complex mix of factors. These factors include genetics; parental health and behaviors such as smoking and drinking during pregnancy; complications during birth; infections the mother might have during pregnancy or the baby might have very early in life; and exposure of the mother or child to high levels of environmental toxins, such as lead. For some developmental disabilities, such as fetal alcohol syndrome, which is caused by drinking alcohol during pregnancy, we know the cause. Following are some examples of what we know about specific developmental disabilities: Some of the most common known causes of intellectual disability include fetal alcohol syndrome ; genetic and chromosomal conditions, such as Down syndrome and fragile X syndrome ; and certain infections during pregnancy. Children who have a sibling with autism are at a higher risk of also having autism spectrum disorder. Low birthweight, premature birth, multiple birth, and infections during pregnancy are associated with an increased risk for many developmental disabilities. Untreated newborn jaundice high levels of bilirubin in the blood during the first few days after birth can cause a type of brain damage known as kernicterus. Children with kernicterus are more likely to have cerebral palsy, hearing and vision problems, and problems with their teeth. Early detection and treatment of newborn jaundice can prevent kernicterus. It is currently the largest study in the United States to help identify factors that may put children at risk for autism spectrum disorders and other developmental disabilities.

9: Congenital naevi | DermNet New Zealand

Functional or developmental birth defects are related to a problem with how a body part or body system works or functions. These problems can include: Some birth defects affect many parts or processes in the body, leading to both structural and functional problems. This information focuses on.

NAEVI AND OTHER DEVELOPMENTAL DEFECTS pdf

Origins of the other Focusing on the Young Child Place of property Structure of the American court and legal systems The Party Animals Colorful Picnic (Honey Bear Books) Economic reform and alternatives for North Korea Thomas F. Cargill and Elliott Parker Political succession in Eastern Europe 6. Beyond the party. Back-of-the-Envelope Physics 2015 gmc canyon service manual The Two New Yorks Training in family planning for health personnel From a writers notebook. Management of student aid History of Birmingham Mass transport in solids Accion Gramatica! (Action Grammar A-Level) The No Sweat Exercise Plan, 3-cd Set Real book jazz gratis Controllin the planet Married women who love women A brief history of straight Its not always about you The ethics of self-management The Handbook of Communication Skills Tourism and agriculture Investigation of Interior Department, etc. Buddhist influence in T. S. Eliots / Revolutionary party Manase relax please part 2 tamil The mirror with a memory chapter 9 A bear in war The crossing of the Hellespont and the first victory Virtual States (The Internet and the Boundaries of the Nation State) Evidence and objections Alcohol and Pleasure (Series on Alcohol in Society) Better Play Times Why do cats have whiskers? A study of history by arnold toynbee Mission and materiel: The Army and acquisition, 1945-1953