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A Report on Bazex-Dupre-Christol Syndrome

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Brief Report

Bazex-Dupré-Christol syndrome is a rare genodermatosis that manifests with the classical triad of follicular atrophoderma, basal cell carcinoma and hypotrichosis; it may be accompanied by milia, neurological symptoms, ichthyosis, and visceral malignancies. Bazex-Dupré-Christol syndrome is an X-linked dominant inherited disorder. BCDS is inherited in an X-linked dominant manner. This means the defective gene responsible for the disorder is located on the X chromosome, and only one copy of the defective gene is sufficient to cause the disorder when inherited from a parent who has the disorder. Males are normally hemizygous for the X chromosome, having only one copy.

As a result, X-linked dominant disorders usually show higher expressivity in males than females. Bazex–Dupré–Christol syndrome is a rare X-linked genodermatosis characterized by early-onset nonmelanoma skin cancers, hypotrichosis, hypohidrosis, atrophoderma follicularis, and multiple milia. Its molecular basis remains unknown and nosologic classification is debated. Trichoepitheliomas are an early sign of Bazex–Dupré–Christol syndrome and may guide the diagnosis even before the development of basal cell carcinomas.

The high frequency of hypotrichosis, hypohidrosis and dry skin in Bazex–Dupré–Christol syndrome indicates that it may be better classified as an ectodermal dysplasia. Comparison with other conditions combining features of ectodermal dysplasia and proneness to skin tumors suggests the involvement of a common pathogenic pathway implicated in both skin development and cancer.

Bazex-Dupré-Christol syndrome which results from a mutation in the *ACTRT1* gene causing depletion of the tumour suppressor ARP-T1, a non-coding mRNA. ARP-T1 is required for the assembly of cilia, microtubule organelles. Actin-Related Protein-Testis1 (ARP-T1)/ ACTRT1 gene mutations cause the Bazex-Dupré-Christol Syndrome (BDCS) characterized by hypotrichosis, follicular atrophoderma and basal cell cancer. ARP-T1 participates in the regulation of cilia length and that ARP-T1-associated BDCS is a case of skin cancer with ciliopathy characteristics. Basal cell cancer (BCC) of the skin is the frequent human cancer.

Bazex–Dupré–Christol syndrome (BDCS) is an X-linked dominantly inherited syndrome form of BCC, without male-to-male transmission, affecting primarily the hair follicle, resulting in the triad of hypotrichosis, follicular atrophoderma and BCC. Hypotrichosis and follicular atrophoderma develop around birth and BCC early in adulthood. BDCS less frequently presents with milia, hypohidrosis, facial pigmentation and trichoepithelioma, and thus combines developmental cutaneous anomalies with tumor predisposition. BDCS is an inherited syndromic form of BCC.

The insertion mutation ACTRT1 547_548InsA creates a shift in the reading frame that results in a non-functional truncated protein of 198 amino acids. ARP-T1 was also found depleted in families with germline mutations of non-coding sequences surrounding ACTRT1 postulated to belong to enhancers transcribed as non-coding RNAs. These mutations segregate with the disease, ARP-T1 can be considered a tumor suppressor in BDCS6. ARP-T1 was first isolated as a major acidic component of the cytoskeletal calyx from the head of bull spermatozoa. ARP-T1 is expressed specifically late in spermatid differentiation in testis, where it locates to the postacrosomal region and the centriole, establishing a link to the primary cilium of epidermal cells implicated in BCC development.

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