

Adult-onset Foveomacular Vitelliform Dystrophy

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Summary:

Adult-onset foveomacular vitelliform dystrophy is one of the most common macular dystrophies, classified within the group of pattern dystrophies. The condition typically arises after the fourth decade of life and manifests with subretinal vitelliform deposits in the macula that may progressively enlarge and later undergo resorption, ultimately resulting in retinal and retinal pigment epithelium atrophy. In most patients, the course is mild; however, significant vision loss may occur in older individuals, mainly as a result of chorioretinal atrophy or the development of subretinal neovascularization. On multimodal imaging, these lesions are hyperautofluorescent. On optical coherence tomography, they exhibit a dome-shaped configuration, while electrooculography and full-field electroretinography most often remain normal, indicating focal rather than global retinal dysfunction. The adult-onset foveomacular vitelliform dystrophy phenotype may mimic or coexist with other conditions, including age-related macular degeneration, vitreomacular traction, or central serous chorioretinopathy, which complicates diagnostic evaluation. In some patients, genetic associations have been identified with mutations in *PRPH2*, *BEST1*, *IMPG1*, and *IMPG2*. Clinical variability and inconsistent terminology have often led to misdiagnoses, underscoring the need for standardized diagnostic criteria.

Key words:

vitelliform macular dystrophy, macula/ pathology, retinal pigment epithelium/ physiopathology, optical coherence tomography (OCT), autofluorescence imaging.

Adult-onset foveomacular vitelliform dystrophy (AFVD)

Adult-onset foveomacular vitelliform dystrophy is one of the most common forms of macular degeneration, with a prevalence of approximately 1: 7400 to 1: 8200 [1]. It was first described by Gass in 1974 [2]. It is classified as one of several forms of pattern dystrophy (PD) [3].

Importantly, AFVD and the early stages of Best disease (Best Vitelliform Macular Dystrophy – BVMD) exhibit similar features on blue autofluorescence (BL-AF) and optical coherence tomography (OCT) images, making them difficult to distinguish using routine examinations. In contrast to AFVD, nearly all patients with Best disease show markedly reduced EOG values. Despite morphological similarities, the functional prognosis in most cases is significantly different – BVMD is characterized by an earlier onset and a poorer final level of visual acuity. The final stage of typical BVMD is associated with severe vision loss and may manifest as retinal atrophy or hypopigmentation (IVa), macular scarring (IVb), or choroidal neovascularization beneath or around the macular scar (IVc) [4].

AFVD is classified within the heterogeneous group of pattern dystrophies, which includes butterfly-shaped pigment dystrophy (BPD), reticular dystrophy of the retinal pigment epithelium, pseudo-Stargardt pattern dystrophy (fundus flavimaculatus), and fundus pulverulentus [5]. The term “pattern dystrophy” was originally proposed to describe dystrophies involving the retinal pigment epithelium (RPE) [6, 7]. However, some of the pattern dystrophies listed above, including AFVD, pseudo-Stargardt pattern dystrophy, and butterfly-shaped pigment dystrophy, have been linked to mutations in the same gene, *PRPH2*, which encodes peripherin-2, a protein with a key structural role in the outer segments of photoreceptors rather than in the RPE [8, 9]. Typically, eyes affected by pattern dystrophies demonstrate various patterns of progressive RPE changes, often accompanied by deposits of yellowish-dark subretinal material involving the macula and posterior pole.

AFVD-like secondary phenotypes may also occur in the course of other ocular disorders, such as age-related macular degeneration (AMD), vitreomacular traction, pseudodrusen, or central serous chorioretinopathy. The disease phenotype may arise from dysfunction of the photoreceptors, the retinal pigment epithelium, and/or the extracellular matrix, with the outer segments of the photoreceptors and disturbances in their phagocytosis potentially playing an important role. In a small number of patients with AFVD, mutations have been identified in the *PRPH2*, *BEST1*, *IMPG1*, or *IMPG2* genes, as well as a single nucleotide polymorphism in the *HTRA1* gene [1].

AFVD is characterized by subretinal vitelliform lesions in the macula, typically appearing after the age of 40. However, the age of symptom onset in pattern dystrophies is highly variable. Patients usually remain asymptomatic until the fifth decade of life, and some even throughout their lifetime. These lesions gradually enlarge and then regress, leaving areas of atrophy in the outer retina and retinal pigment epithelium, which is associated with progressive loss of visual acuity. Pattern dystrophies generally follow a relatively mild course; however, substantial visual impairment is observed in up to 50% of patients beyond the age of 70, due to chorioretinal atrophy and/or the development of macular neovascularization (MNV) [10, 11]. Vitelliform lesions are hyperautofluorescent and initially exhibit a dome-shaped appearance on OCT, while electrooculography (EOG) and full-field electroretinography (ERG) usually remain within normal limits, indicating the localized nature of retinal damage.

Over the years, various terms have been used in the literature to describe AFVD. The general term “adult vitelliform lesion” has also been applied to refer to vitelliform changes occurring in adults that do not necessarily have a genetic basis. This considerable diversity of nomenclature has led to significant confusion among clinicians, researchers, and patients, and is likely partly responsible for the high frequency of misdiagnoses of this condition. The variable terminology reflects the lack of consensus regarding the diagnostic criteria and pathogenesis of AFVD [12].

Clinical course

Gass suggested that AFVD typically becomes apparent between the ages of 30 and 50, presenting with bilateral subfoveal yellowish deposits occupying approximately one-third of the optic disc area, with a centrally located pigmented spot. Over time, lesion pigmentation may intensify, the yellow color may diminish, and the RPE may develop areas of atrophy [2].

AFVD has been described as either asymptomatic or presenting with various visual symptoms, such as reduced visual acuity and/or metamorphopsia, with a slow progression of visual function decline. In some patients from the original series, drusen were also observed in the vicinity of the lesion. Electrophysiological and color-vision assessments were largely within normal limits, although a mild reduction in EOG was noted in some cases. Based on this initial description and a suggestive family history, in some cases it was hypothesized that the disease might follow an autosomal dominant pattern of inheritance. Although most of these initial observations remain valid, it is now known that AFVD often manifests at an older age than described by Gass and is generally not associated with autosomal dominant inheritance.

One of the main limitations of earlier studies is the absence of strict diagnostic criteria for AFVD, which may have led to the inclusion of adults with vitelliform changes secondary to other conditions that differ from AFVD. At that time, the association of vitelliform lesions with many diseases was also unknown, partly due to the lack of appropriate imaging methods such as spectral-domain optical coherence tomography (SD-OCT) and fundus autofluorescence (FAF).

In the past, studies focused on the role of autosomal dominant inheritance, demographic characteristics, visual function, and prognosis in AFVD, as well as on distinguishing this entity from AMD [13, 14]. However, some of these studies did not confirm autosomal dominant inheritance, and a similar phenotype was observed in patients without a family history and with normal EOG, suggesting that AFVD is not always hereditary [15, 16].

In 1990, a larger cohort of 81 patients with a clinical presentation consistent with AFVD was described. They were found to have yellowish lesions in the fovea or pigment clumps within the foveal region, usually surrounded by a hypopigmented halo. The mean age of the patients was 67 years, which is higher than in Gass's original series, and only one patient had a positive family history. In 34 individuals (42%), changes characteristic of early AMD coexisted, particularly the presence of small drusen. Among the 17 patients for whom clinical follow-up was available, visual acuity remained stable in nine, while in eight it deteriorated by at least one line. Based on the late age of onset and the coexistence of features typical of AMD, the authors suggested that the described phenotype is probably degenerative in nature and falls within the spectrum of AMD-related changes [17].

A high prevalence of drusen (60%) was also documented in 53 eyes of 31 patients older than 50 years. Family history was negative in this group, and during follow-up (median 3 years), a deterioration of visual acuity by at least two lines was observed in 53% of eyes. Importantly, eyes with distinctly yellowish lesions showed a greater tendency toward progression of vision loss than those in which pigment-cluster abnormalities predominated [18].

In an analysis of 85 cases, the median age at disease onset was 61 years (range: 37–81 years). Women accounted for 62.3% of the study population, and a positive family history (mother and son) was reported in only one patient. The electrooculogram (EOG) was normal in 67% of cases or only slightly reduced in 33%, distinguishing this presentation from the classic form of Best disease. Vitelliform lesions showed gradual evolution leading to oval or round foci of retinal pigment epithelium atrophy

in the macula. Initially, visual acuity was relatively good: ≥ 0.6 vision was recorded in 43% of eyes. However, maintaining this level of acuity showed a clear downward trend: after one year it was present in 59% of eyes, after two years in 28.5%, after four years in only 20%, and in the group followed for a decade, in just 8%. Visual decline was predominantly linked to progressive retinal atrophy and RPE dysfunction; however, subretinal neovascularization occurred in approximately 15% of cases, with macular edema reported sporadically. Due to the late age of onset, the absence of consistent inheritance, and the frequent overlap with degenerative retinal features, the authors concluded that these changes represent a well-defined phenotype within the spectrum of AMD. At the same time, it was noted that despite the similarity to AMD, the clinical distinctiveness and observed genetic variability may indicate a subtype of AMD with a potential hereditary basis [19].

Another large clinical analysis included 61 consecutive patients meeting the criterion for AFVD, defined as the presence of a central vitelliform lesion smaller than one optic disc diameter in at least one eye. A total of 120 eyes were evaluated. The mean age at diagnosis was 54.6–55 years, and visual acuity was highly variable, with a median of approximately 0.6 (range 1.25–0.05). In the vast majority of cases (92%), the family history was negative. Only five patients had first-degree relatives with similar changes, which emphasizes the lack of typical inheritance and suggests a sporadic phenotype. Around half of the patients had normal color vision and visual fields, while the rest presented with highly variable defects. In autofluorescence imaging, increased fluorescence within the yellow macular lesion was observed in 76% of cases, which was interpreted as lipofuscin accumulation. In most patients, electrophysiological results in both full-field and multifocal ERG were reduced. Molecular analysis identified a potentially pathogenic mutation in the RDS/peripherin gene in only one patient, confirming the sporadic nature of most AFVD cases. In the group with documented follow-up (25 eyes, period: 0.9–5.3 years), approximately half showed disease progression leading to deterioration of visual acuity, most often accompanied by metamorphopsia, central scotomas, or other visual disturbances. Based on the results, the authors defined AFVD as a late-onset disorder, usually with a slow course and a relatively favorable functional prognosis, but with considerable variability in both morphological and functional presentation, which contributes to frequent diagnostic errors. It was also indicated that the clinically visible vitelliform lesion corresponded to lipofuscin accumulation, and that cone system dysfunction was generalized, with greater severity in the macular region [12].

In a retrospective study involving adult patients with AFVD, progression of changes on SD-OCT was assessed over a follow-up period of at least 12 months. The analysis included 46 eyes of 31 patients with a mean age of 74.6 years; the mean follow-up duration was 16 months. Visual acuity deteriorated significantly, from 0.32 to 0.39 logMAR ($p = 0.03$). At baseline, the vitelliform stage predominated (60.8%), followed by the pseudohypopyon stage (15.2%) and the “scrambled egg” stage (23.9%). During follow-up, the proportion of the “scrambled egg” stage increased (28.2%), and an atrophic stage appeared (10.9%). Stability of the disease stage – and its reflection in SD-OCT findings as well as continuity of the IS/OS junction – was associated with preserved visual acuity, whereas deterioration in these parameters correlated with vision loss ($p = 0.03$). In patients with disease progression, central retinal thickness and subretinal lesion parameters were markedly reduced. The findings indicate that resorption of the vitelliform material and the associated disruption of the IS/OS junction are key processes leading to deterioration of visual function in this disease [20].

Imaging

Fluorescein angiography

AFVD is diagnosed and analyzed using a variety of imaging techniques and functional tests. In his original description of the phenotype, Gass characterized the changes observed in fluorescein angiography (FA), and since then, many other researchers have confirmed his observations. At early disease stages, vitelliform lesions may cause fluorescence blockage due to the presence of vitelliform material and pigment within the fovea, and the lesion may be surrounded by a ring of hyperfluorescence corresponding to an area of RPE atrophy.

In the recirculation phase, staining of the vitelliform material with fluorescein dye is often observed. Therefore, in FA, this may falsely produce an image resembling poorly defined type 1 choroidal neovascularization [21]. Choroidal neovascularization (CNV) may develop in eyes with AFVD; in such cases, confirmation or exclusion of CNV coexisting with the vitelliform lesion may require a combination of clinical evaluation and multimodal imaging.

Fundus autofluorescence

A characteristic feature of vitelliform changes – although not specific to AFVD – is an increased fundus autofluorescence (FAF) signal. Abnormal hyperautofluorescence corresponds to the clinically visible material [21]. FAF imaging shows considerable phenotypic variability, presenting various forms (including focal, ring-like, or patchy patterns); however, the pattern type does not correlate with best corrected visual acuity (BCVA), suggesting limited prognostic value of FAF alone in assessing visual function [22].

In another study, blue-light autofluorescence (BL-AF) and near-infrared autofluorescence (NIR-AF) were evaluated, along with microperimetry assessing retinal sensitivity. In BL-AF, three pattern types were distinguished: normal, focal, and heterogeneous, whereas in NIR-AF imaging two patterns were observed: focal and heterogeneous. Abnormal autofluorescence images were present in the vast majority of patients – in 86% with BL-AF and in 100% with NIR-AF. The poorest functional outcomes, assessed both by visual acuity and retinal sensitivity in microperimetry, were observed in patients exhibiting a heterogeneous pattern in both imaging modalities. The results indicate that AFVD is characterized by considerable variability in autofluorescence patterns, and the assessment of FAF features may reflect the functional status of the retina. The heterogeneous pattern, in particular, is associated with poorer macular function, and the combined use of BL-AF and NIR-AF serves as a valuable tool for assessing disease severity as well as monitoring its progression. As the disease advances and atrophy progresses, areas of hypoautofluorescence appear in most patients, which likely reflects degeneration or loss of the RPE. It is worth noting, however, that central hypoautofluorescence can also be observed in the vitelliform stage of AFVD, indicating that autofluorescence changes do not always correlate clearly with the degree of atrophy, but may partly result from the accumulation or masking of subretinal material [20, 21, 23].

Optical coherence tomography (OCT)

Spectral-domain OCT (SD-OCT) offers much higher resolution than time-domain OCT, allowing precise localization of the hyperreflective structure that is clinically visible as yellowish material in the subretinal space above the RPE. In most patients, this material was clearly demarcated, often with the overlying photoreceptor layer preserved. This suggests partial retinal functionality in the early stages of the disease.

In SD-OCT, the most commonly observed morphology of vitelliform deposits was heterogeneous (60%) and less frequently

homogeneous (23%). Occasionally, the deposits were hypo- or areflective (17%). Focal elevations of the RPE were commonly observed (48%), as well as hyperreflective foci within the outer plexiform and outer nuclear layers (47%). Irregularities of the photoreceptor inner/outer segment (IS/OS) junction were another common finding. In 45% of eyes, the IS/OS line was interrupted and disrupted, indicating progressive photoreceptor damage, whereas in 15% of eyes it retained a distinct hyperreflective form surrounding the vitelliform deposits. RPE abnormalities, including localized structural and reflectivity disturbances, were found in 23% of eyes [24]. Complementary *en face* OCT imaging confirmed a concentric arrangement of changes: in 77% of eyes, a continuous hyperreflective ring corresponded to the preserved IS/OS junction, while in 67% of eyes a darker, hyporeflective ring separated this junction from the central deposit. The central element (the vitelliform material) remained highly reflective. In 27% of eyes, a characteristic superior, crescent-shaped hyporeflective area (“*croissant*”) was observed, corresponding to sedimentation of vitelliform material. Additionally, distinct RPE thickening was detected in 83% of eyes, and its location corresponded to intense foci in near-infrared imaging [25].

In some patients, subretinal fluid was additionally observed. As vitelliform changes progressed, the outer nuclear layer underwent gradual thinning and photoreceptor structures progressively disappeared, ultimately becoming undetectable in more advanced cases, which is consistent with photoreceptor degeneration. Thinning of the neurosensory retina and accumulation of subretinal deposits correlated with visual acuity deterioration, whereas the best visual function was observed when the external limiting membrane and the continuity of the photoreceptor inner/outer segment (IS/OS) junction were preserved, confirming the crucial role of photoreceptor integrity in prognosis [21, 22].

SD-OCT enables quantitative assessment of the spatiotemporal properties of changes in AFVD. Querques et al. observed the natural course of the disease in 46 eyes of 31 patients (mean age: 75 years, mean follow-up time: 16 months). At baseline, most eyes were in the vitelliform stage, with smaller groups in the pseudohypopyon or “scrambled egg” stage. The mean maximum lesion diameter was 1,605 μm . During follow-up, most vitelliform lesions (61%) remained unchanged, whereas approximately 11% progressed to the atrophic stage. In eyes in which progression was observed, the lesion dimensions decreased: the mean lesion height (from the RPE to the photoreceptors) declined from 277 μm to 105 μm , while the mean width decreased from 2.324 μm to 1.751 μm . Pseudohypopyon lesions initially had a larger diameter than vitelliform lesions and gradually underwent resorption, progressing to the “scrambled egg” stage and atrophy. Stabilization of disease stage and integrity of the photoreceptor inner/outer segment (IS/OS) junction was associated with maintenance of visual acuity, whereas deterioration of these features resulted in vision loss (the mean visual acuity declined from 0.32 logMAR to 0.39 logMAR). In addition, SD-OCT revealed hyperreflective nodules and elevations at the level of the RPE in approximately 40% of cases, and reticular drusen were identified in two patients. These results indicate that the progression of AFVD, involving partial or complete resorption of material, is associated with disruption of ellipsoidal layer continuity, changes in the RPE, and gradual loss of visual function [20].

Another study evaluated the correlation between clinical observations and multimodal imaging findings in patients with acquired vitelliform lesions, and analyzed their characteristics and natural course. The retrospective analysis included 90 eyes from 67 patients (mean age: 73 years). Integrity of the ellipsoid zone and the photoreceptor inner/outer segment (IS/OS) junction correlated with visual acuity, whereas its disruption led to visual deterioration.

At baseline, the mean lesion diameter was 975 μm and decreased to 747 μm during follow-up. The mean deposit height declined from 185 μm to 152 μm , demonstrating an inverse correlation with visual acuity. The outer nuclear layer above the lesion became thinned, indicating gradual photoreceptor loss, although the thinning itself did not directly correlate with visual function. In 39% of patients, visual acuity remained stable during a mean follow-up of 11 months. In 13% of cases, lesion resorption was observed, accompanied by focal atrophic changes, without significant visual decline. The authors hypothesized that resorption of vitelliform material may occur as a result of substantial photoreceptor loss and gradual removal of the material by the RPE. In addition, some patients exhibited cuticular or reticular drusen as well as RPE detachment. The findings suggest that both photoreceptor integrity and the degree of subretinal material accumulation are key determinants of visual function. The authors assumed that both dysfunction of the retinal pigment epithelium (RPE) and loss of adhesion of photoreceptor terminals to the RPE may impair phagocytosis of shed photoreceptor outer segments. Both the accumulated material and pigment-laden macrophages and RPE cells contribute to the yellowish coloration of vitelliform lesions. In some cases, progressive loss of photoreceptors may be associated with spontaneous lesion resorption [21].

Optical coherence tomography angiography (OCTA)

OCTA represents a valuable method for assessing macular vascularization in patients with AFVD, particularly in the context of detecting neovascularization. Studies have shown that OCTA can detect CNV at the pseudohypopyon stage, even when there are no definitive signs of neovascularization on fluorescein angiography (FA). For example, in one of eight eyes (12.5%) at this stage, CNV was visible only on OCTA. In another study, CNV was revealed in two cases, although conventional imaging did not show it due to masking of the vessels by vitelliform material. At the same time, OCTA is subject to characteristic artifacts, which are observed in almost all patients with AFVD. The most common of these are segmentation errors (observed in approximately 86% of eyes), which can falsely suggest vascular loss in the deep capillary plexus, and flow-signal blockage artifacts beneath the vitelliform lesion, resulting in the apparent disappearance of choriocapillaris vasculature [26, 27].

Electrophysiological studies

Electrophysiological examinations, including electrooculography (EOG), electroretinography (ERG), and multifocal ERG, were also performed in patients with AFVD to assess retinal and RPE function. In the original description of the phenotype, Gass noted that the EOG recording is normal in most cases, although it may be reduced [2].

In AFVD, the Arden ratio in EOG may be either normal or reduced, regardless of the presence of mutations in the *PRPH2* and *BEST1* genes, which limits its diagnostic value. This finding suggests that some mutations may affect RPE function, while others may not, although the mechanism responsible for these differences remains unknown. Although the EOG result is often normal, focal RPE dysfunction in the fovea cannot be excluded [28].

In AFVD, full-field ERG is generally normal, although in some patients a slight decrease of rod-cone responses is observed. Macular dysfunction is revealed by reduced amplitudes in both full-field and multifocal ERG. This indicates that in AFVD, damage occurs primarily at the level of foveal photoreceptors, which correlates with OCT findings, while peripheral retinal function is preserved and the EOG is usually normal [12].

Differentiation

AFVD may mimic or coexist with various retinal disorders, which often makes it difficult to distinguish from other macular

pathologies. The most frequent differential diagnosis is age-related macular degeneration (AMD), particularly in older patients. Reports have described vitelliform lesions occurring alongside cuticular drusen, soft drusen, and subretinal drusenoid deposits – features typical of AMD [29–31]. Studies by Gass and subsequent authors demonstrated that 20–30% of patients with cuticular drusen also exhibit vitelliform lesions, often accompanied by reduced visual acuity. These changes exhibit a clinical, autofluorescence, and OCT appearance similar to classic AFVD, and their subretinal location suggests a shared pathogenic mechanism arising from dysfunction of the photoreceptor-RPE complex. In some cases, regression of vitelliform material has been observed, typically associated with atrophy and visual decline, which further resembles advanced AMD [30, 31].

Another condition requiring differentiation is Best disease, the classic inherited macular dystrophy caused by mutations in the *BEST1* gene. Compared with AFVD, the condition typically presents earlier and is characterized by larger vitelliform lesions, although, like AFVD, it may follow a multifocal course. Genetic studies have shown that a small proportion of patients with AFVD carry *BEST1* mutations; in such cases, the disease resembles a milder form of Best dystrophy, often with a normal EOG [32].

Vitelliform lesions may also occur in a secondary process due to mechanical separation of the photoreceptors from the RPE. They have been described in the course of central serous chorioretinopathy, epiretinal membrane, vitreomacular traction, and after retinal detachment surgery. In these cases, the accumulation of vitelliform material results from impaired phagocytosis of photoreceptor outer segments and represents a secondary process [33].

Conclusions

AFVD represents a common phenotype of various retinal disorders, both hereditary and acquired, and their diagnosis requires the use of multimodal imaging. A key role in pathogenesis is played by dysfunction of the retinal pigment epithelium and loss of contact between photoreceptor terminals and the RPE, which disrupts normal phagocytosis of outer segments. The accumulated subretinal material, along with migrating pigment-laden macrophages and RPE cells, accounts for the characteristic yellowish appearance of the lesions. With progression, there is loss of IS/OS junction integrity, pigment migration into the inner layers, and, in some cases, fluid accumulation, reflecting gradual photoreceptor damage. This process may lead to spontaneous resorption of the vitelliform lesion, often under conditions of advanced photoreceptor loss.

Imaging modalities, particularly OCT, are the cornerstone of AFVD diagnosis. OCT enables accurate localization of deposits and evaluation of photoreceptor integrity, which is crucial for prognosis. Given the similar clinical appearance of vitelliform lesions in the course of AMD and other drusen-related pathologies, the diagnosis of AFVD should be based not only on lesion morphology but also on age at onset, the characteristic OCT findings, and, in selected patients, analysis of mutations in the *BEST1* and *PRPH2* genes.

All available data indicate that AFVD represents a phenotype of disorders at the photoreceptor-RPE interface, rather than a distinct, homogeneous disease entity. Multimodal imaging diagnostics combined with clinical analysis are therefore crucial for accurate differentiation, visual prognosis, and monitoring disease progression.

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