Anatomic–echocardiographic correlates: an introduction to normal and congenitally malformed hearts

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With a sound understanding of anatomy, detecting and diagnosing most congenital heart malformations with echocardiography need not be an onerous task. Using all the modalities of ultrasound, the examination should define as completely as possible the morphological and physiological aspects of the malformation. The cross sectional imaging modality is a superb tool for displaying the morphology of intracardiac and vascular anomalies. In this review we provide the anatomical correlates to the more common forms of congenital cardiac defects.

Description and display

Transthoracic echocardiography accesses the heart through the parasternal, suprasternal, apical, and subcostal windows. Similar views of the heart can also be obtained by the transoesophageal and transgastric windows. Innumerable series of planes can be displayed through each window. Since the orthogonal planes of the heart are different from the major axes of the body it is conventional to describe the window of access as well as the reference plane—for example, the parasternal long axis plane (fig 1). It is also appropriate to display the image in as near anatomical orientation as possible. The sector fan can be inverted on the modern ultrasound equipment. This is a most useful facility when interrogating complex malformations. Operators no longer need to reorient the images mentally or stand on their heads in order to figure out the relative position of structures!

The basic planes

The body and the heart can be described in terms of their three orthogonal planes. The planes of the body are the sagittal and coronal long axis planes and the short axis planes. Because the long axis of the normal heart is at an angle to that of the body, the sagittal and coronal planes of the body are also at an angle to the long axis planes of the heart (fig 1). Instead, the long axis planes of the heart transect the heart so that they display all four cardiac chambers (“four chamber plane”) or only the right or left heart chambers (“two chamber planes”). Owing to the spiral arrangement of the ventricular outflow tracts, the “two chamber” planes often include the outflow tract of the other side of the heart.1

Short axis planes through the heart are at an angle to short axis planes through the body. Such planes can be obtained from the parasternal window. Another series of short axis planes can be obtained from the subcostal window but these are in the parasagittal planes. The subcostal window also allows the heart to be viewed along its long axis but in the paracoronal plane. While the subcostal short axis and long axis cuts approximate to the parasternal short axis planes and the four chamber planes, respectively, there are significant differences.

Figure 1  Upper panel: Diagrams showing the orthogonal planes of the body at an angle to that of the heart and the transthoracic windows for accessing the heart. Middle panels: The parasternal approach produces cuts through the short and long axes of the heart itself whereas the apical approach yields long axis two and four chamber cuts. Lower panels: Access from the subcostal window yields short and long axis cuts through the heart that are in parasagittal and paracoronal planes of the body.
Figure 2  The middle panel shows the endocast viewed from the front displaying mainly the right heart chambers (in blue) and very little of the left heart chambers (in red). When viewed from the right (A), the right heart chambers are to either side of the aorta. The view from the left (B) shows the relation between the right and left ventricular outflow tracts and the posterior location of the left atrium. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle; PT, pulmonary trunk; RS, right cusp.

Table 1  Morphological features of chambers and great arteries

<table>
<thead>
<tr>
<th>Chambers</th>
<th>Morphologically right</th>
<th>Morphologically left</th>
</tr>
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<tbody>
<tr>
<td>Atria</td>
<td>triangular, broad based appendage</td>
<td>terminal crest</td>
</tr>
<tr>
<td></td>
<td>extensive pectinate muscles</td>
<td>rim around oval fossa</td>
</tr>
<tr>
<td>Ventricles</td>
<td>tubular, narrow based appendage</td>
<td>no terminal crest</td>
</tr>
<tr>
<td></td>
<td>limited pectinate muscles</td>
<td>valve of oval fossa</td>
</tr>
<tr>
<td>Great arteries</td>
<td>fine apical trabeculations</td>
<td>moderator band</td>
</tr>
<tr>
<td></td>
<td>leaflet of AV valve attached directly to septum (septal leaflet of TV)</td>
<td>septomarginal trabeculation</td>
</tr>
<tr>
<td></td>
<td>morphologically right</td>
<td>very coarse trabeculations</td>
</tr>
<tr>
<td></td>
<td>fine apical trabeculations</td>
<td>no ventricular septum (solitary ventricle)</td>
</tr>
<tr>
<td></td>
<td>smooth upper part of septum (without attachment of mitral valve)</td>
<td></td>
</tr>
</tbody>
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Basic anatomy of the normal heart

Understanding the normal anatomy is essential for interpretation of malformations. Viewing the patient from the front, the cardiac silhouette is more or less trapezoidal in shape, with the right atrium forming the right heart border and the left ventricle forming the sloping left heart border. The upper part of the trapezium is occupied by the arterial trunks emerging from the ventricles. With two thirds of the bulk of the heart lying to the left of the midline in the thorax, and the overlapping arrangement of the cardiac chambers, the so-called left and right heart structures do not occupy positions implied by their names. Overall, the “right” chambers are anteriorly located relative to their supposedly left sided counterparts, while the right ventricular outflow tract and the pulmonary trunk pass anterior and leftward to the aorta (fig 2). The atrial chambers are positioned superior and to the right relative to their respective ventricular chambers. The left atrium is also the most posteriorly situated cardiac chamber, lying anterior to the tracheal bifurcation.

Since the locations of cardiac chambers and great vessels can be abnormal in congenitally malformed hearts, it makes more sense to describe them in terms of their morphology. Thus, atrial and ventricular chambers have characteristic features ascribing “morphological rightness” and “morphological leftness”. Similarly, the great arteries can be distinguished according to their branching pattern. In the normal or relatively normal heart there are several recognisable features for each chamber. In the setting of malformations, however, features for determining what is morphologically right or left may be limited to the most constant anatomic markers or by inference. The anatomic characteristics for the cardiac chambers are listed in table 1.

The key to understanding the overall arrangement of the cardiac structures is the central location of the aortic valve (fig 3). Even though the aorta emerges from the left ventricle, its valve is in right inferior position relative to the pulmonary valve. The tricuspid valve, guarding the junction between right atrium and right ventricle, is more or less vertically orientated and is the most inferior of the cardiac valves. The aortic and mitral valves, adjoining one another, are hidden behind the right ventricle. It follows from the disposition of the cardiac chambers that the septal structures are not on one plane. The atrial septum is positioned obliquely behind the aorta whereas the ventricular septum is curved, allowing the right
Normal and congenitally malformed hearts

Dissections made into the sandwich risk exiting the heart. At the apex of the sandwich is the membranous septum which, together with the union of the rightward margin of fibrous continuity between aortic and mitral valves, forms the central fibrous body.

The atrial septum is not nearly as extensive as might be suggested by a cursory glance at the right aspect. Sections through this area demonstrate nicely its true extent that is confined to the floor of the oval fossa and its immediate surroundings (fig 4). Any excursion beyond the septal area will lead toward epicardial tissues instead of into the adjoining atrium. The right aspect of the septum can be identified by the rim of the fossa whereas the left aspect is relatively featureless.

For the anatomist, the atrial appendages are sufficiently different to allow the morphologically right atrium to be distinguished from the morphologically left atrium. The internal features are even more striking (fig 2). In the right atrium, an array of trabeculations branch out in comb-like fashion from the terminal crest. These pectinate muscles extend also into the posteroinferior wall. In contrast, the wall of the left atrium is much smoother since the pectinate muscles are mainly located within the finger-like appendage. The pulmonary veins enter the left atrium at the posterior part of the venous component. The valves of the inferior caval vein and the coronary sinus are additional features of the right atrium. The course of the coronary sinus passing beneath the inferior wall of the left atrium revealed by cuts angled toward the diaphragmatic surface is another useful marker.

Ventricles are best considered in terms of possessing three components: inlet, apical trabecular, and outlet. It is the apical trabecular portion that most consistently permits distinction between right and left morphologies, even in the absence of one or more of the other portions. The coarse apical trabeculations and a prominent moderator band crossing the ventricular cavity are characteristics of the right ventricle. Trabeculations in the apical portion of the left ventricle are fine by contrast. The nature of the ventricular inlets, the atrioventricular valves, are also helpful markers. The tricuspid valve in the right ventricle has a circular orifice guarded by septal, anterosuperior, and mural (inferior) leaflets. Direct chordal attachments of the septal leaflet to the ventricular septum readily distinguish the tricuspid valve. The mitral valve with its kidney shaped orifice is guarded by mural and aortic (anterior) leaflets. Since the leaflets are attached entirely to paired papillary muscles situated in anterolateral and posteromedial positions, the septal surface is smooth and devoid of valvar insertions. The smooth muscular septum is one half of the left ventricular outflow tract as seen in short axis sections (fig 5). The other half is formed by the curtain-like aortic (anterior) leaflet of the mitral valve. In long axis, two chamber sections, the inlet and outlet lengths of the left ventricle are equal (fig 5).

Figure 4  (A) The membranous septum is revealed by transillumination in this view of the right atrium and right ventricle. The hingeline of the tricuspid valve divides the membranous septum into atrioventricular (AV) and interventricular (IV) components. (B) This four chamber section shows the more apical attachment of the tricuspid valve compared to the mitral valve resulting in an offset arrangement (arrows). The cardiac septum is profiled showing the thin valve of the oval fossa. Superiorly, the rim is an infolding of the atrial wall (arrow). (C) A cut taken slightly more cephalad passes through the aortic root producing the so-called five chambers section that shows the aortic valve wedged between the atrioventricular valves. Note the thin membranous septum (small arrow) and the moderator band (thick yellow arrow).

Figure 5  (A) This short axis section shows the mitral valve (MV) in fibrous continuity with the aortic valve. The corresponding echocardiographic section shows the arrangement of the mitral leaflet. (B) This cut shows a long axis section that replicates a parastral or apical view. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle; S, septum.
The trileaflet arrangement of the semilunar leaflets is characteristic of the arterial valves. Distinction of aorta from the pulmonary trunk is based on branching pattern (fig 6). The pulmonary trunk branches early into right and left pulmonary arteries with a third vessel, the arterial duct, seen at the bifurcation in fetuses and neonates (fig 6). The aorta gives rise to the coronary arteries and then ascends to branch into the neck and arm arteries at the aortic arch before continuing as the descending thoracic aorta. The normal aortic arch curves leftward and crosses superiorly relative to the bifurcation of the pulmonary trunk. In its rightward course, the right pulmonary artery therefore passes underneath the aortic arch. This view can be displayed using the supersternal approach (fig 6).

**Sequential segmental analysis**

The majority of patients with congenital malformation exhibit normal relations and connections of the cardiac chambers. In order to be able to identify cases with abnormal connections, however, it is necessary to adopt a systematic method of diagnosis. A relatively simple approach termed sequential segmental analysis is described here (fig 7). Based on morphological features rather than presumed embryological derivatives, even seemingly complex malformations can be described.

The first step in analysis is the determination of the arrangement of the atrial appendages (situs). Four variants are possible: usual (situs solitus), mirror image of normal (situs inversus), isomeric right, and isomeric left. The morphology of the atrial appendages can be observed from parasternal and subcostal positions. Although previously described as syndromes of heterotaxy, the correlation between isomeric arrangement of the atrial appendages and arrangement of abdominal organs and status of the spleen is not always consistent. Arrangement of the atrial appendages corresponds more frequently to arrangement of the branching pattern of the main bronchi and their relation to the left and right pulmonary arteries. Inference can also be made from the spatial relationship between the abdominal great vessels and the spine (fig 8). Usually, the abdominal aorta descends in front or slightly to the left of the spine with the inferior caval vein occupying a position to the right of the spine and anterior to the aorta (fig 8). Reversal of this pattern is present in cases with mirror imagery of normal arrangement of the atrial appendages. When the venous and arterial trunks are on the same side of the spine with the aorta in posterior position, there is usually right isomerism. Approximately 70% of cases with left isomerism can be distinguished by the associated interruption of the suprarenal portion of the inferior caval vein. Blood from the lower body drains via the azygos (or hemiazygos) vein into a superior caval vein. The enlarged azygos vein will be to the same side of the spine as the aorta but in a posterior position. The hepatic veins connect bilaterally to the atrial chambers but occasionally via a common vessel.

The second step in segmental analysis examines the atrioventricular junction in terms of biventricular and univentricular connections. Biventricular atrioventricular connections describe the arrangement whereby each atrium is connected to its own ventricle, albeit that one of the ventricles may be hypoplastic or the atrioventricular valve is imperforate. By contrast, univentricular atrioventricular connections describe hearts where only one ventricle is connected to the atrial mass. The combinations of chambers for biventricular connections are concordant or discordant in hearts with
usual and mirror image arrangement of the atrial appendages, but ambiguous in those with isomeric atrial appendages (fig 9). Invoking the concept of ventricular topology further aids description of ambiguous atrioventricular connections.

There are also three specific patterns to univentricular atrioventricular connections. Existing with any of the four possible atrial arrangements, the patterns are double inlet ventricle and absent right and absent left atrioventricular connections. The atrioventricular junction itself can be connected to one of three possible morphologies in the ventricular mass (fig 10). The receiving ventricle is usually large and described as dominant. Most frequently, the dominant ventricle is of left ventricular morphology recognised by its fine apical trabeculations. Accompanying the dominant left ventricle, there is usually a rudimentary right ventricle in the anterosuperior portion of the ventricular mass that can be revealed by short axis sections. Occasionally, the dominant ventricle is of right morphology with coarse trabeculations. On short axis sections, the rudimentary left ventricle will be seen toward the diaphragmatic aspect of the ventricular mass. The third possible morphology is solitary and indeterminate ventricle which contains muscle bundles thicker than those found in the morphologically right ventricle.

While double inlet will show both atrial chambers, or their major portions, entering the same dominant ventricle in long axis sections, hearts with absence of the right or left atrioventricular connections will show only one atrium opening to the dominant ventricle (fig 11). Absence of connection will be revealed by the blind muscular floor of the atrium overlying the ventricular mass, with the fatty tissue plane of the atrioventricular groove sandwiched in between.

When analysing the atrioventricular junction, it is also necessary to assess the valvar morphology, noting straddling and overriding, stenosis or imperforateness, etc. All hearts with overriding atrioventricular junctions produce a spectrum of anomalies between the extremes of double inlet and biventricular atrioventricular connections. When coexisting with absence of right or left atrioventricular connection, it can produce the very rare situation of uniatrial with biventricular connections. Since the potential chamber connections in the presence of overriding are legion, the degree of override at greater or less than 50% is used to determine the type of atrioventricular connection (fig 12).

The third step in segmental analysis is at the junction between ventricles and great arteries. Ventricle-arterial connections are concordant when the arterial trunks arise from the appropriate ventricles, but discordant when
they are connected to inappropriate ventricles, irrespective of the relations of the trunks and the musculature or infundibulum of the outflow tract. When both arterial trunks are connected to the same ventricle, this situation is described as double outlet from either the right, the left, or the indeterminate ventricle accordingly. Occasionally, the blind origin of one of the great arteries cannot be traced back to a specific ventricle. When this occurs as in some cases with aortic or pulmonary atresia, the term single outlet is used. Single outlet of the heart can also be produced by a common arterial trunk exiting through a common valve (fig 13). Alternatively, there is a solitary arterial trunk with complete absence of intrapericardial remnants of the pulmonary trunk and its branches (so-called truncus type IV).

The morphology of the arterial valves need to be noted. In cases with overriding of an arterial valve, the 50% rule is used in adjudicating ventriculo-arterial connections.

Segmental analysis is not complete until all the associated malformations have been examined. These include anomalies of systemic and pulmonary venous connections, septal defects, valvar anomalies, coronary abnormalities, aortic arch anomalies, anomalies of pulmonary arteries, position of the heart and apex, etc.

Abnormal position of the heart is not a diagnosis in itself but it alerts the investigator to the likely presence of abnormal arrangement of the body organs and possibility of intracardiac lesions. Unusual echocardiographic views may be needed to analyse these hearts sequentially.

Echocardiographic anatomy of congenital malformations

As described above, most congenital heart malformations occur with normal connections of the cardiac chambers but still the chambers are analysed sequentially before the specific lesion or lesions are highlighted. As it is not possible in this review to present all forms and variations of malformations, we will restrict our discussion to the more common entities.

**atrial septal defects**

Conventionally, four types of “atrial septal defects” are described: secundum, sinus venosus, coronary sinus, and ostium primum. Although most defects that permit interatrial shunting are at the site of the oval fossa (“secundum defects”) and involve the atrial septum, the less common defects are outside of the confines of the atrial septum (see above). The “secundum defect” relates to the putative “ostium secundum” and on echocardiography is seen as an area of dropout in the oval fossa. The morphology of oval fossa defects ranges from small inadequacy of the valve, multiple fenestrations, filigreed appearance, to complete absence of the valve (fig 14). Parasternal and apical views can produce dropout artefacts leading to false positive diagnosis. Scans from a subcostal parasagittal plane or a lower right parasternal approach will show the thin atrial septum in axial rather than lateral resolution.

The sinus venosus defect is more commonly located superiorly at the junction of the superior caval vein and the right atrium than inferiorly related to the orifice of the inferior caval vein. In hearts with superior sinus venosus defects, the right upper pulmonary vein or the superior caval vein connects to both atria and the interatrial communication is superior to the true atrial septum (fig 14).

The coronary sinus defect, or partially unroofed coronary sinus, is rare. It is usually associated with persistence of the left superior caval vein, and the lesion is a spectrum of deficiencies between the apposing walls of the coronary sinus and the left atrium. Scanning subcostally in a parasagittal plane may reveal its presence.

The “ostium primum defect” is more common but involves the atroventricular junction, not a deficiency of the atrial septum since in many cases the valve of the fossa is adequate and competent. This lesion will be considered in the section on atroventricular septal defects.

**Ventricular septal defects**

Over the years, numerous taxonomies have been proposed for the classification of ventricular septal defects. Our preference is to use a simple descriptive categorisation that can alert
the surgeon or the interventional cardiologist repairing the defect to the proximity or remoteness of the major bundles of the atrioventricular conduction system. A fuller description should include defect size and presence or absence of septal malalignment.

All ventricular septal defects, whether existing in isolation or as an integral part of a combination of lesions, can be described as belonging to one of three anatomic groups (fig 15). The most common is described as perimembranous since the defect abuts the area of the membranous septum, or its remnant, and an area of fibrous continuity involving the atrioventricular valves forms part of its border. This fibrous border harbours the penetrating atrioventricular bundle and its continuation into the ventricular bundles. The normal wedged position of the aortic outflow tract accounts for the subaortic position of such perimembranous defects in the otherwise normal heart (fig 15). The defects can extend so that they open from the subaortic area mostly into the inlet, outlet or trabecular portions of the right ventricle, or they can be large and confluent. Several views are used to obtain an image of perimembranous defects. The parasternal short axis and apical four chamber views demonstrate proximity to the tricuspid valve while parasternal and apical long axis views show the relation to the aortic valve. Perimembranous defects extending to the inlet portions are best shown in apical four chamber or subcostal paracoronal views.

The second group, muscular defects, is distinguished from perimembranous defects by their muscular borders (fig 15). Again, these can be described according to their locations as inlet, outlet, or trabecular. Multiple muscular defects give the septum a “swiss cheese” appearance. Apically situated defects may be small and difficult to detect among the trabeculations without careful interrogation across the entire septum using apical four chamber and parasternal short axis or subcostal paracoronal views. With posterior angulation, these views also allow display of muscular inlet defects that are roofed by a ridge of muscle separating the atrioventricular valves. The presence of the ridge and normal offsetting of the valves distinguish muscular from perimembranous inlet defects (fig 15).

The least common group, doubly committed and juxta-arterial defects, is characterised by fibrous continuity between the adjacent leaflets of the aortic and pulmonary valves (fig 15). Superiorly, these defects are roofed by the arterial valves while postero-inferiorly the margin may be muscular or perimembranous. The parasternal long axis or subcostal parasaggital planes will display the continuity of both arterial valves and also the presence, if any, of herniation of the right facing aortic sinus.

**ATRIOVENTRICULAR SEPTAL DEFECTS**

Anatomically, the feature unifying hearts with atrioventricular septal defect is the common

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**Figure 14** (A) Right atrial view of an oval fossa defect and a similar defect (arrow) seen on four chamber section. (B) Right atrial view shows the superior sinus venous defect (thick yellow arrow) beneath the orifice of the superior caval vein (SCV) and anomalous insertion of the right pulmonary vein (PV). The oval fossa (OF) is intact. The echocardiographic section shows the defect (arrow) superior to the intact atrial septum and anomalous right upper pulmonary vein (RUVP). ICV, inferior caval vein; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.

**Figure 15** (A) The locations of perimembranous (red circles), muscular (black circles), and doubly committed (blue circles) subtypes of ventricular septal defects are superimposed on this right ventricular view. (B) This four chamber cut through a perimembranous inlet ventricular septal defect shows the fibrous continuity (red dotted line) between tricuspid (TV) and mitral (MV) valves forming the roof of the defect. The normal offset between the valves is lost. (C) The muscular defect (arrow) has complete muscular borders. (D) The doubly committed and aortic-arterial defect (arrow) is roofed by fibrous continuity (red dotted line) between the aortic and pulmonary valves.
atrioventricular junction guarded by a common valve. Unlike the normal figure-of-eight arrangement, the atrioventricular junction is typically oval shaped in these hearts. The abnormal junctional anatomy is associated with an elongated and anteriorly positioned aortic outflow tract and notable disproportion between the inlet and outlet lengths in the left ventricle. The atrioventricular valve is abnormal, having a five leaflet configuration, two of which bridge the ventricular septum anterosuperiorly and posteroinferiorly. Two other leaflets are exclusive to the right ventricle. The bridging leaflets together with the remaining leaflet guard the left ventricular inlet in a tri-leaflet configuration (fig 16).

Atrioventricular septal defects represent a spectrum of lesions with potential for shunting at the atrial level as exemplified by “ostium primum defects”, at the ventricular level, or at both levels. The size of the defect is variable as is the arrangement of the valvar leaflets to each other and to the septum.

The variability in anatomy requires imaging in multiple planes properly to define the entirety of the lesion. The apical or subcostal four chamber views can display the loss of offset arrangement of valvar leaflets at the septum, a particularly useful feature in antenatal screening. Anterior angulation of the transducer can display the anterosuperior bridging leaflet (fig 16). Short axis parasagittal planes can show the five leaflet arrangement of the common valve, highlighting the abnormal trileaflet left component and location of the papillary muscles.

TETRALOGY OF FALLOT

The classical four features of this anomaly are: ventricular septal defect, biventricular connection (overriding) of the aorta, subpulmonary stenosis, and right ventricular hypertrophy.

The anatomical hallmark is anterior and cephalad deviation of the septal insertion of the outlet septum relative to the septomarginal trabeculation in the right ventricle (fig 17). Compared with the normal heart, this feature accounts for both the ventricular septal defect and subpulmonary stenosis. The latter is exacerbated by hypertrophy of septoparietal trabeculations in the outflow tract. Variations exist in the morphology and number of ventricular septal defect, pulmonary valvar and arterial stenosis, degree of aortic override, aortic arch anomalies and coronary arterial anomalies. The association of Fallot with absence or underdevelopment of the pulmonary valve (so-called absent pulmonary valve syndrome) leads to grossly enlarged pulmonary arteries that can cause bronchial obstruction.

Biventricular connection of the aorta is readily apparent in the precordial long axis section (fig 17). Rotation from long axis to short axis planes can demonstrate the ventricular septal defect, deviation of the outlet septum, and the right ventricular outflow tract. In infants especially, the malformation is best shown from the subcostal window along para coronal and parasagittal planes (fig 17). The pulmonary arteries and aorta can be evaluated from suprasternal or high precordial cuts.

COMPLETE TRANSPOSITION OF THE GREAT ARTERIES

Segmentally, this malformation is defined as the combination of discordant atrioventricular connections with discordant ventriculo-arterial connections. When occurring without intracardiac shunts, the so-called simple form, this condition is not compatible with life without early palliation. Recognising this condition prenatally or in the neonate is particularly helpful in planning management. The aorta arises from the morphologic right ventricle via a muscular infundibulum, while the pulmonary valve arising from the morphologic left ventricle is usually in fibrous continuity with the mitral valve. Although not diagnostic, the frequently abnormal spatial relation between the great arteries alerts the investigator to the possibility of this malformation or other defects. Most often, the aorta arises anteriorly and to the right of the pulmonary trunk and the proximal great arteries run in parallel instead of
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